PRECISION HEALTH

PRECISYA

SKIN PANEL

ENGLISH VERSION

Name	Sample				
Age		Gender	F	Report date	12/09/2025
Prescriber		Health insurance			

Sample

WARNING

The values of the results of genetic tests are not diagnostic, but show trends that are influenced by physiological, pathological conditions, use of medications and other personal conditions of the examinee.

Only your clinician is able to correctly interpret these results and to prescribe the most appropriate treatment for you, and the company is not responsible for any treatment based on the results.

If necessary, our science team is available to discuss the results with the attending clinician upon request.

The genetic test

The genetic examination is the most current and advanced technological leap in the health area, mainly for the clinical area because DNA is the true Instruction Manual for the individual.

The exam shows conditions, determined by genetics, that may or may not develop at some point in life, as in DNA, all individual needs, susceptibilities and psycho-behavioral and structural characteristics are determined with high precision, functional and reactive that an individual has and will have throughout his life.

Today science considers Epigenetics, a term that encompasses countless factors such as the state and emotional relationships, nutrition, physical activity and environmental factors, among others, as of fundamental value for development (expression), or not (silencing), of these conditions.

Hence the importance of genetic examination. It allows each person to know what their tendencies are and thus be able to work epigenetically to prevent them from developing (genetic silencing), thus maintaining their Health, Vitality, Beauty and Longevity.

The information found in the DNA, which determines the individual differences and the conditions analyzed in the exams, are called Polymorphisms (SNPs). In each condition our exam can find and analyze up to several dozen polymorphisms.

The current level of our technology, allows the high level of precision and reliability of our exams in the fundamental aspects for a genetic exam.

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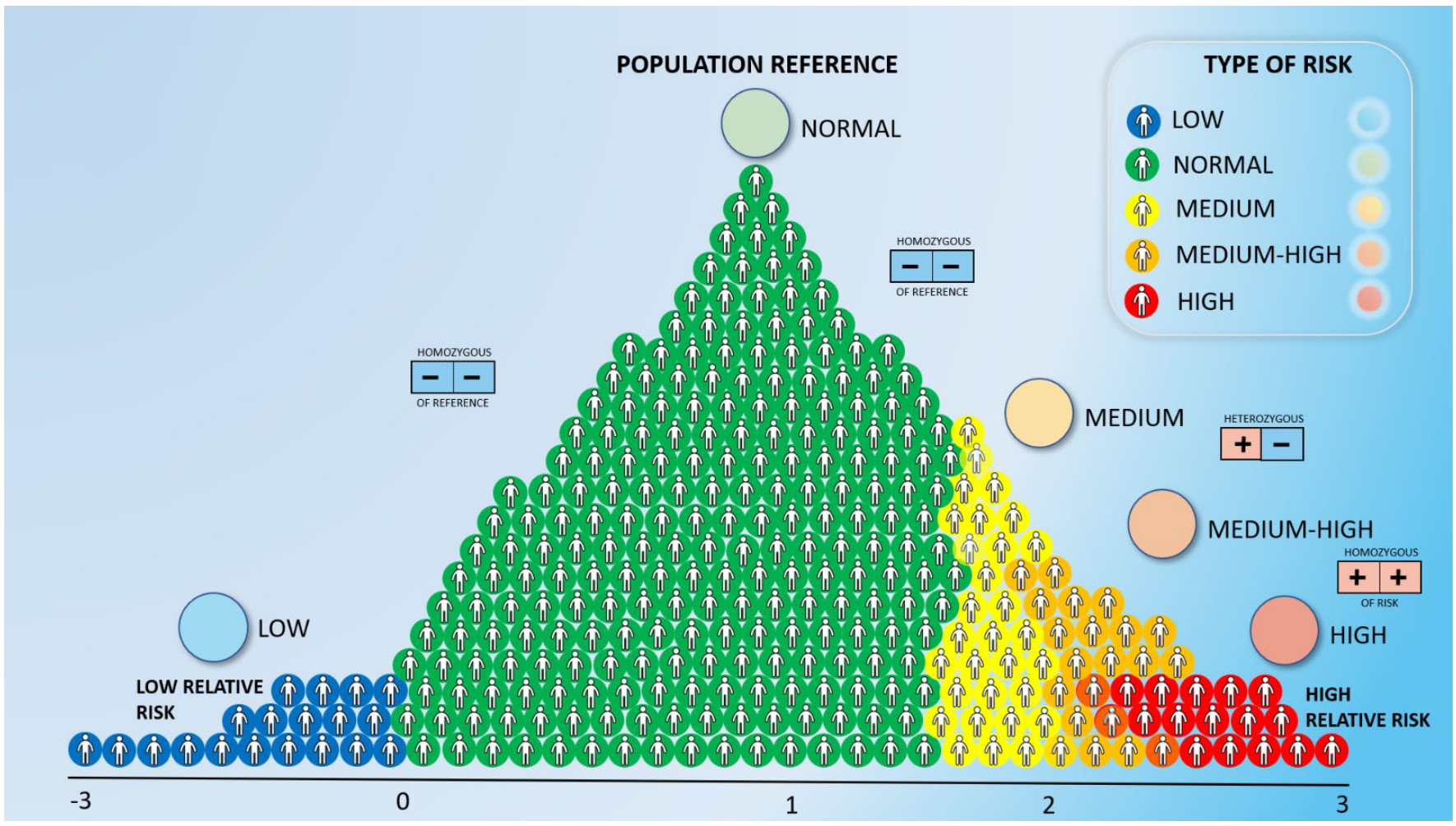
How to interpret the exam:

FIRST PART

The analyzed genetic CONDITIONS are grouped into CATEGORIES.

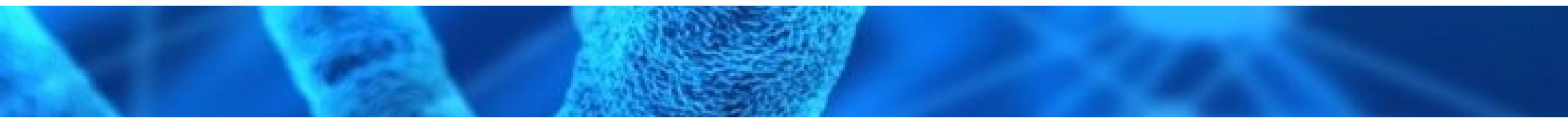
Each CONDITION is presented according to its MAGNITUDE. That is, what is the genetic susceptibility (intensity or possibility) of the analyzed condition to express itself (happen).

- If the susceptibility is TOO HIGH, a RED dot will appear
- If the susceptibility is HIGH, an ORANGE dot will appear
- If the susceptibility is AVERAGE, a YELLOW dot will appear
- If the susceptibility is NORMAL a GREEN dot will appear
- If the susceptibility is LOW, a BLUE dot will appear
- If the condition is not identified GRAY dot



PART TWO

In the second part the CATEGORIES and CONDITIONS are shown again in more detail and presenting the analyzed genes



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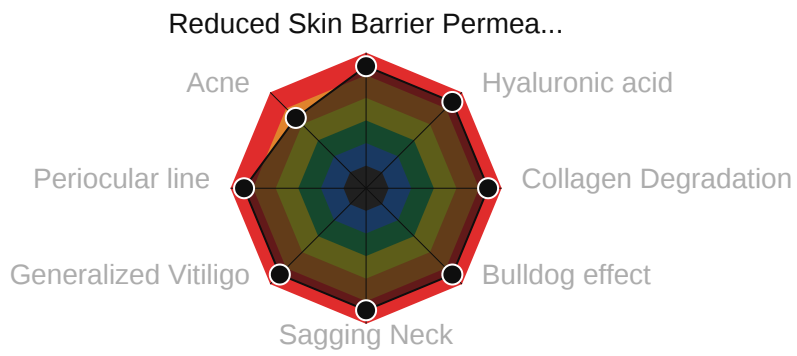
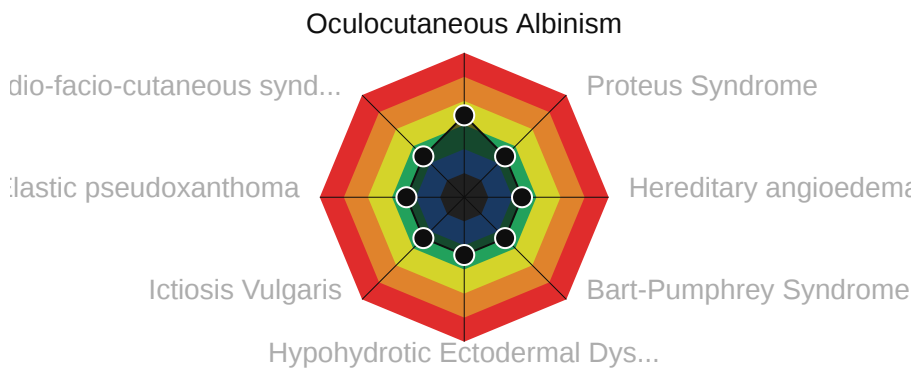
MOST RELEVANT CONDITIONS BY CATEGORY

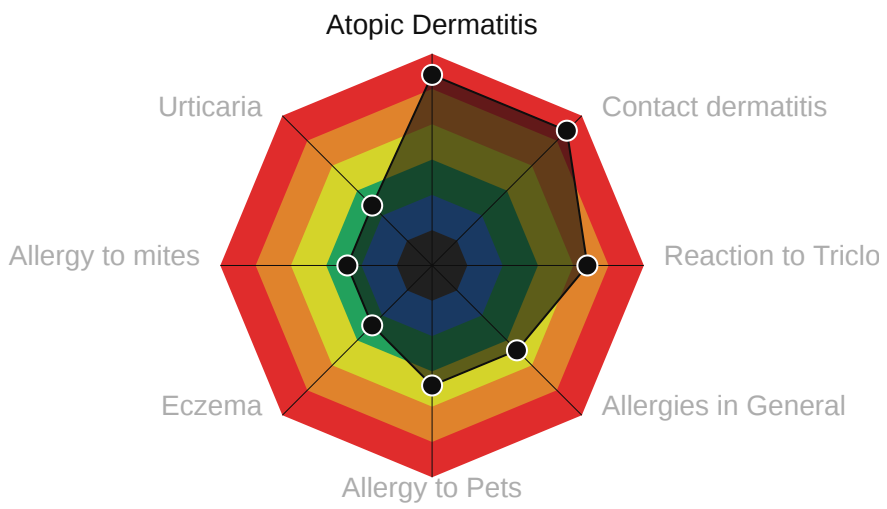
SKIN



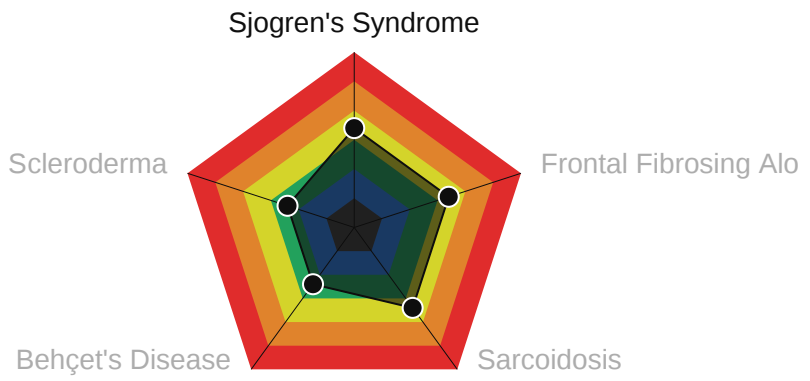
GENETIC DISEASES



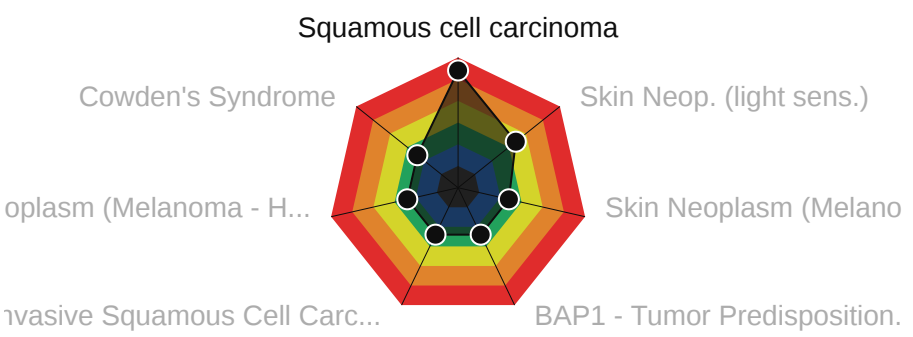
ALLERGIES



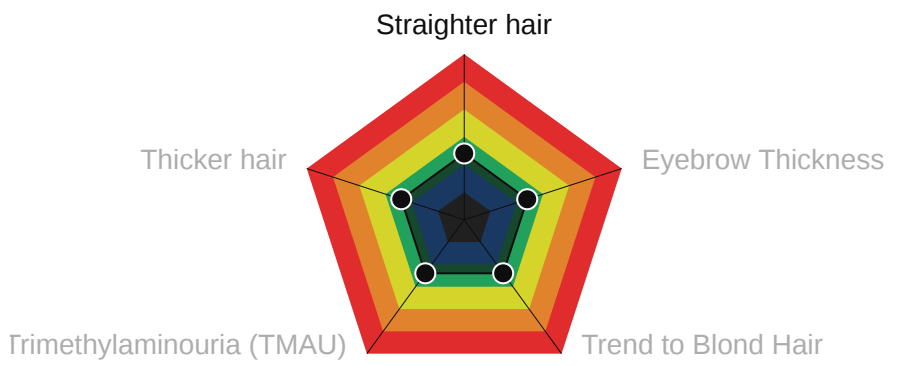
CHRONIC SUBCLINICAL INFLAMMATION



CANCER



PERSONAL CHARACTERISTICS



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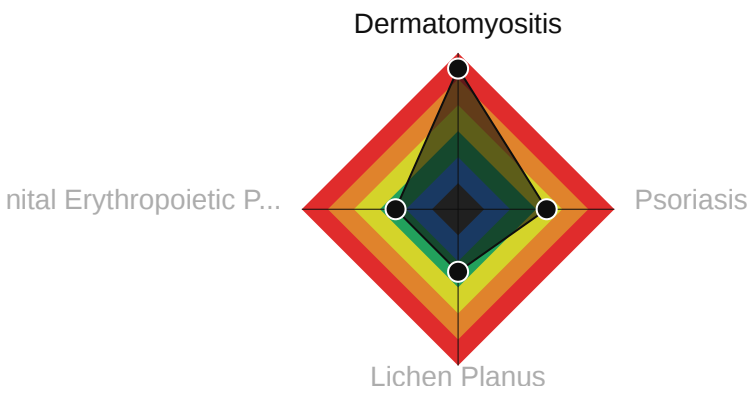
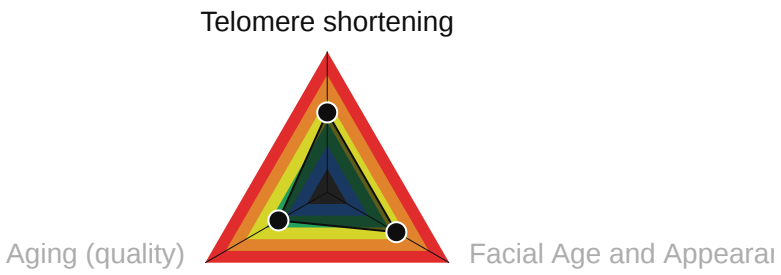
MOST RELEVANT CONDITIONS BY CATEGORY

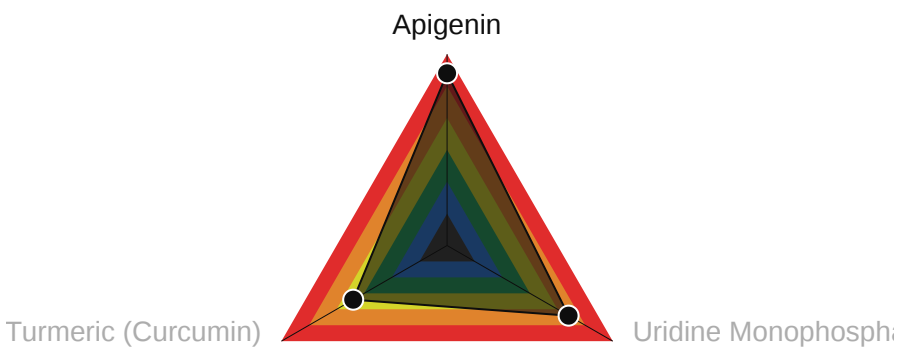
IMMUNE SYSTEM



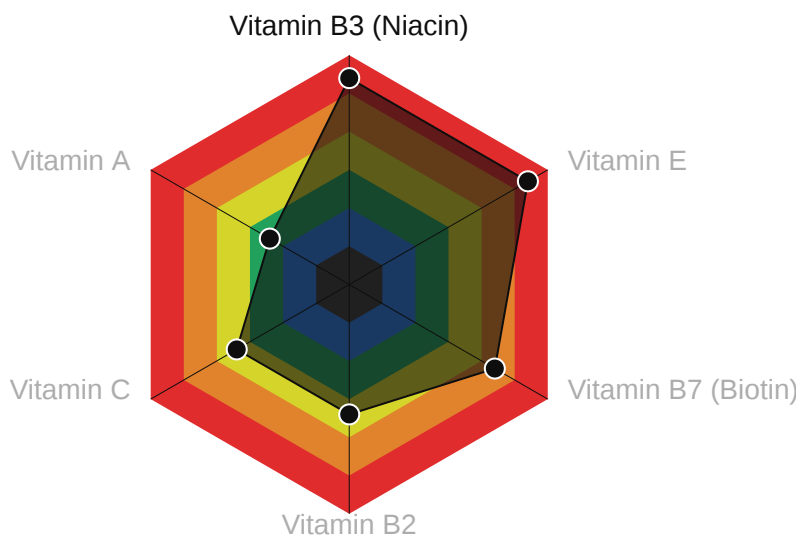
AGING



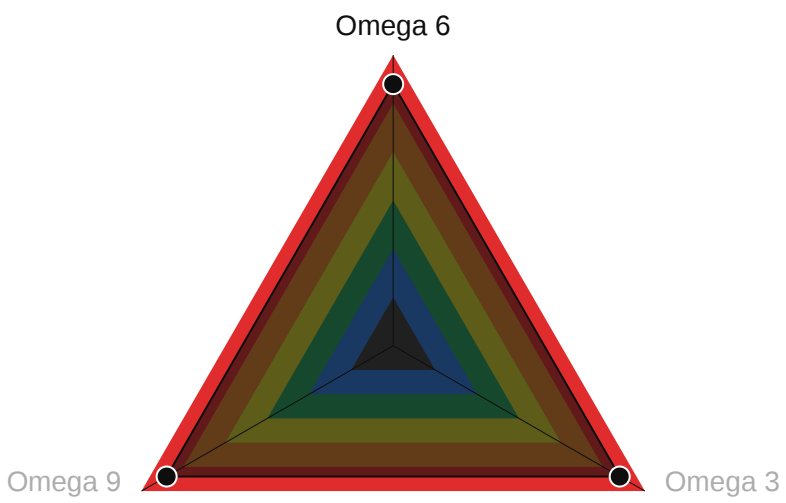
NEED FOR NUTRIENTS



VITAMINS



FATTY ACIDS



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SUMMARY OF RESULTS

Aging

Telomere shortening	17	-	-	8	+	-	6	+	+	MEDIUM
Facial Age and Appearance (greater aging)	16	-	-	2	+	-	0	+	+	MEDIUM
Aging (quality)	28	-	-	3	+	-	0	+	+	NORMAL

Allergies

Atopic Dermatitis	27	-	-	3	+	-	2	+	+	HIGH
Contact dermatitis	9	-	-	1	+	-	1	+	+	HIGH
Reaction to Triclosan	0	-	-	5	+	-	0	+	+	MEDIUM-HIGH
Allergies in General	4	-	-	1	+	-	0	+	+	MEDIUM
Allergy to Pets	5	-	-	0	+	-	1	+	+	MEDIUM
Eczema	18	-	-	1	+	-	0	+	+	NORMAL
Allergy to mites	5	-	-	0	+	-	0	+	+	NORMAL
Urticaria	4	-	-	0	+	-	0	+	+	NORMAL
Allergy to pollen	3	-	-	1	+	-	0	+	+	LOW

Amino acids

L-arginine	11	-	-	0	+	-	0	+	+	NORMAL

Antioxidants / Supplements

Coenzyme Q10	150	-	-	13	+	-	11	+	+	HIGH

Cancer

Squamous cell carcinoma	20	-	-	1	+	-	1	+	+	HIGH
Skin Neoplasm (light sensitivity)	1	-	-	0	+	-	1	+	+	MEDIUM
Skin Neoplasm (Melanoma)	21	-	-	1	+	-	1	+	+	NORMAL
BAP1 - Tumor Predisposition Syndrome	1	-	-	0	+	-	0	+	+	NORMAL
Invasive Squamous Cell Carcinoma	3	-	-	0	+	-	0	+	+	NORMAL
Skin Neoplasm (Melanoma - Hereditary)	1	-	-	0	+	-	0	+	+	NORMAL
Cowden's Syndrome	10	-	-	0	+	-	0	+	+	NORMAL

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Chronic Subclinical Inflammation

Sjogren's Syndrome	2	-	-	4	+	-	0	+	+	MEDIUM
Frontal Fibrosing Alopecia	0	-	-	1	+	-	0	+	+	MEDIUM
Sarcoidosis	2	-	-	1	+	-	0	+	+	MEDIUM
Behçet's Disease	5	-	-	1	+	-	0	+	+	NORMAL
Scleroderma	2	-	-	0	+	-	0	+	+	NORMAL

Digestive system

Canker sores	3	-	-	2	+	-	0	+	+	NORMAL

Fatty acids

Omega 6	312	-	-	33	+	-	24	+	+	HIGH
Omega 3	107	-	-	23	+	-	9	+	+	HIGH
Omega 9	144	-	-	24	+	-	12	+	+	HIGH

General

Loss of hair	14	-	-	3	+	-	0	+	+	MEDIUM

Genetic diseases

Oculocutaneous Albinism	1	-	-	1	+	-	0	+	+	MEDIUM
Proteus Syndrome	1	-	-	0	+	-	0	+	+	NORMAL
Hereditary angioedema	4	-	-	1	+	-	0	+	+	NORMAL
Bart-Pumphrey Syndrome	58	-	-	0	+	-	0	+	+	NORMAL
Hypohydrotic Ectodermal Dysplasia	3	-	-	0	+	-	0	+	+	NORMAL
Ictiosis Vulgaris	1	-	-	0	+	-	0	+	+	NORMAL
Elastic pseudoxanthoma	1	-	-	0	+	-	0	+	+	NORMAL
Cardio-facio-cutaneous syndrome	2	-	-	0	+	-	0	+	+	NORMAL
Blau's Syndrome	2	-	-	0	+	-	0	+	+	NORMAL

Hematologic system

Hyperbilirubinemia	2	-	-	0	+	-	1	+	+	MEDIUM
Post-Transfusion Purpura (PPT)	2	-	-	0	+	-	0	+	+	NORMAL

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Immune system

Dermatomyositis	2	-	-	0	+	-	1	+	+	HIGH
Psoriasis	42	-	-	4	+	-	3	+	+	MEDIUM
Lichen Planus	2	-	-	0	+	-	0	+	+	NORMAL
Congenital Erythropoietic Porphyria (Gunther's Disease)	5	-	-	0	+	-	0	+	+	NORMAL

Metabolic

Glycation	5	-	-	1	+	-	0	+	+	MEDIUM-HIGH

Metabolic disorders

Acute Intermittent Porphyria	3	-	-	0	+	-	0	+	+	NORMAL
Lipodystrophy	17	-	-	0	+	-	0	+	+	NORMAL

Microbiome

Staphylococcus aureus infections	0	-	-	1	+	-	0	+	+	HIGH

Need for Nutrients

Apigenin	1	-	-	0	+	-	1	+	+	HIGH
Uridine Monophosphate	32	-	-	3	+	-	2	+	+	MEDIUM-HIGH
Turmeric (Curcumin)	1	-	-	1	+	-	0	+	+	MEDIUM

Oxidation

Oxidative stress	29	-	-	11	+	-	5	+	+	HIGH
Antioxidant Capacity	23	-	-	4	+	-	1	+	+	MEDIUM

Personal characteristics

Straighter hair	3	-	-	0	+	-	0	+	+	NORMAL
Eyebrow Thickness	1	-	-	0	+	-	0	+	+	NORMAL
Trend to Blond Hair	1	-	-	0	+	-	0	+	+	NORMAL
Trimethylaminouria (TMAU)	3	-	-	1	+	-	0	+	+	NORMAL
Thicker hair	1	-	-	0	+	-	0	+	+	NORMAL

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Reactions to Treatments

Glucocorticoid Resistance	4	-	-	0	+	-	0	+	+	NORMAL
Glucocorticoid Therapy	0	-	-	1	+	-	0	+	+	NORMAL

Skin

Reduced Skin Barrier Permeability Function	0	-	-	0	+	-	1	+	+	HIGH
Hyaluronic acid	5	-	-	0	+	-	1	+	+	HIGH
Collagen Degradation	673	-	-	9	+	-	10	+	+	HIGH
Bulldog effect	673	-	-	8	+	-	10	+	+	HIGH
Sagging Neck	684	-	-	7	+	-	10	+	+	HIGH
Generalized Vitiligo	10	-	-	7	+	-	1	+	+	HIGH
Periocular line	18	-	-	2	+	-	1	+	+	HIGH
Acne	14	-	-	9	+	-	1	+	+	MEDIUM-HIGH
Melasma	4	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Dark circles	23	-	-	3	+	-	1	+	+	MEDIUM-HIGH
Chicken feet	19	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Bags	18	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Gravitational Wrinkles	18	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Forehead wrinkles	19	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Chin Wrinkles	18	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Alopecia Areata	1	-	-	3	+	-	1	+	+	MEDIUM-HIGH
Nasogenian Line	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Perioral Line	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Puppet Lines	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Loss of Facial Volume	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Fall from the Corner of the Mouth	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH

PRECISION HEALTH PRECISYA

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Nasal wrinkles	17	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Cellulitis	0	-	-	0	+	-	1	+	+	MEDIUM-HIGH
Inflammatory Skin Response	0	-	-	2	+	-	0	+	+	MEDIUM-HIGH
Response to tanning	6	-	-	2	+	-	1	+	+	MEDIUM-HIGH
Collagen Synthesis	2	-	-	1	+	-	0	+	+	MEDIUM
Skin Hydration	5	-	-	2	+	-	0	+	+	MEDIUM
Varicose veins	3	-	-	1	+	-	0	+	+	MEDIUM
Stretch marks	13	-	-	2	+	-	2	+	+	MEDIUM
Wrinkles in the lap	6	-	-	1	+	-	0	+	+	MEDIUM
Baldness (Androgenetic Alopecia)	14	-	-	1	+	-	2	+	+	MEDIUM
Permanent Elastic Wrinkles	7	-	-	1	+	-	0	+	+	MEDIUM
Antioxidant capacity of the skin	6	-	-	3	+	-	0	+	+	MEDIUM
Ephelides (Freckles)	4	-	-	0	+	-	1	+	+	MEDIUM
Lentigos (Sun Spots)	9	-	-	0	+	-	1	+	+	MEDIUM
Ehlers Danlos Syndrome (collagen)	8	-	-	1	+	-	0	+	+	MEDIUM
Eyelid sagging	11	-	-	2	+	-	0	+	+	MEDIUM
Expression Marks (Glabella)	17	-	-	1	+	-	0	+	+	MEDIUM
Atypical Mycobacteriosis	7	-	-	0	+	-	1	+	+	MEDIUM
Hyperchromias	0	-	-	1	+	-	0	+	+	NORMAL
Skin Aging	9	-	-	3	+	-	0	+	+	NORMAL
Dermatochalasis (Excess Skin on the Eyelids)	1	-	-	0	+	-	0	+	+	NORMAL
Junctional bullous epidermolysis atresia of the pylorus	1	-	-	0	+	-	0	+	+	NORMAL
Elastin Deficiency	56	-	-	0	+	-	0	+	+	NORMAL
KID Syndrome	4	-	-	0	+	-	0	+	+	NORMAL
Family candidiasis	5	-	-	0	+	-	0	+	+	NORMAL
Dystrophic Bullosa Epidermolysis	27	-	-	0	+	-	0	+	+	NORMAL
Simple Bullous Epidermolysis	2	-	-	0	+	-	0	+	+	NORMAL
Armpit odor	1	-	-	0	+	-	0	+	+	NORMAL
Pseudofolliculitis barbae	1	-	-	0	+	-	0	+	+	NORMAL
Keloids	5	-	-	1	+	-	0	+	+	NORMAL
Hereditary Trichilemmal Cysts	1	-	-	0	+	-	0	+	+	NORMAL
Wrinkles	3	-	-	1	+	-	0	+	+	LOW

Leprosy	3	-	-	0	+	-	0	+	+	LOW
Skin elasticity	2	-	-	2	+	-	0	+	+	LOW
Ceramides	10	-	-	2	+	-	0	+	+	LOW
Alcohol Erythema Reaction	1	-	-	0	+	-	0	+	+	LOW

Vitamins

Vitamin B3 (Niacin)	3	-	-	0	+	-	4	+	+	HIGH
Vitamin E	323	-	-	32	+	-	16	+	+	HIGH
Vitamin B7 (Biotin)	94	-	-	11	+	-	3	+	+	MEDIUM-HIGH
Vitamin B2	11	-	-	3	+	-	0	+	+	MEDIUM
Vitamin C	123	-	-	18	+	-	4	+	+	MEDIUM
Vitamin A	4	-	-	0	+	-	0	+	+	NORMAL

Weight

Adiposity	186	-	-	10	+	-	1	+	+	MEDIUM-HIGH

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Aging

Telomere shortening

MEDIUM

Telomere shortening can lead to premature aging, in terms of increased risk of age-associated diseases such as heart disease and at least some forms of cancer, and therefore also reduced longevity. Result and orange or red indicate greater telomere shortening. Blue result indicates larger telomeres.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ACYP2	rs11125529	CC+	A	+ +	
ATM	rs1801516	AG+	A	+ -	
BHMT	rs3733890	GG+	A	- -	
C2	rs558702	CC-	T	- -	
CBS	rs5742905	TT-	G	- -	
CLPTM1L	rs401681	CT+	T	+ -	
CTC1	rs3027234	GG-	T	- -	
INTERGENIC	rs398652	GG+	A	- -	
INTERGENIC	rs6028466	GG+	A	+ +	
MTR	rs1805087	GG+	G	- -	
MYNN	rs10936599	CT+	T	+ -	
NAF1	rs7675998	GG+	G,T	- -	
PXK	rs6772228	TT+	A	- -	
STN1	rs9420907	AA+	A	+ +	
TERC	rs12696304	CG+	G	+ -	
TERF2	rs4783704	CC+	C	+ +	
TERT	rs2242652	CC-	A	- -	
TERT	rs2736098	AG-	T	- -	
TERT	rs2736100	TT-	A	+ +	
TNKS	rs11249943	AA+	C	- -	
TRPM6	rs11144134	CT+	C	+ -	
UCP2	rs659366	CT+	T	+ -	
UCP2	rs660339	CT-	T	+ -	
VDR	rs7975232	AC+	A	+ -	
VN1R85P	rs412658	TT+	C	- -	
ZBTB46	rs755017	AA+	G	+ +	

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Facial Age and Appearance (greater aging)

MEDIUM

Physical appearance. Tendency to look younger, taking into account age and other aging factors. Result in red and orange indicates greater aging.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
TERC	rs12696304	CG+	G	+ -	

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Aging (quality)

NORMAL

Aging, in human beings, is the process of wearing out the body. Results in orange and/or red indicate greater aging.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
APOB	rs676210	AG+	A,T	- -	
APOB	rs1367117	GG+	A	- -	
B3GALT1	rs13020412	AA+	G	- -	
CDKN2A	rs3731249	GG-	A,G,T	- -	
CETP	rs5882	AG+	A	+ -	
CFH	rs1061147	CC+	C	- -	
DEF8	rs4268748	CT+	C	- -	
ERI1	rs96621	CT+	C	- -	
HDAC4	rs3791406	CT+	C	+ -	
INTERGENIC	rs9287638	CC+	A	- -	
INTERGENIC	rs12661968	TT+	C	- -	
IRF4	rs12203592	CC+	T	- -	
KL	rs9536314	TT+	A,G	- -	
LMNA	rs553016	GG-	G,T	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
PPARG	rs17036170	GG+	A	- -	
SLC45A2	rs185146	CT+	T	+ -	
UCP2	rs660339	CT-	T	- -	

Allergies

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Atopic Dermatitis

HIGH

Skin inflammation that causes itching.

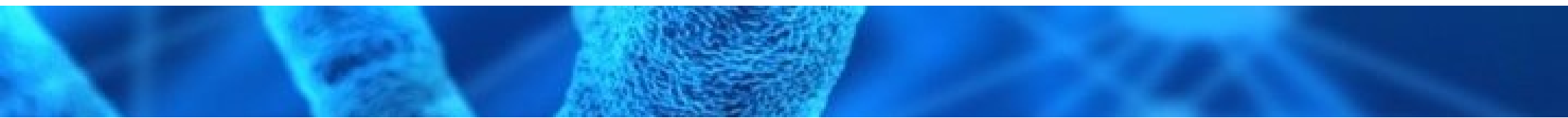
Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CLEC16A	rs12708716	AG+	G	- -	
FLG	rs3814300	CC+	C	+ +	
FLG	rs61816761	GG+	A,T	- -	
FLG	rs121909626	CC-	C	- -	
IL-13	rs1800925	CC+	G,T	- -	
IL-13	rs2066960	AC+	A	- -	
IL-18RAP	rs917997	AG-	A,C	- -	
IL-6R	rs2228145	AA+	C,T	- -	
IL-6R	rs4129267	CC+	G,T	- -	
LEP	rs2167270	AG+	A	+ -	
OVOL1	rs479844	CT-	G	- -	
RTEL1	rs6010620	AG+	C,G	+ -	
SPINK5	rs2303067	AA+	G	- -	
ZBTB10	rs1051920	CT+	T	- -	
ZNF365	rs2393903	AG-	A,C	- -	
ZNF365	rs7076156	GG+	C,G	- -	
ZNF365	rs7089814	TT+	G,T	+ +	

Contact dermatitis

HIGH

Skin rash caused by contact with a substance.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FLG	rs3814300	CC+	C	+ +	
FLG	rs61816761	GG+	A,T	- -	
FLG	rs121909626	CC-	C	- -	
LTA	rs1799724	CT+	T	+ -	
TNF	rs1800610	CT-	A	- -	
TNF	rs1800629	GG+	A	- -	



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Sample

Reaction to Triclosan

MEDIUM-HIGH

This substance is present in numerous products, such as soaps, toothpaste, deodorants and bactericidal soaps, being used as an antibacterial. The indiscriminate use (without proper need) of products with triclosan increases the occurrence of bacterial resistance, which disrupts the human body's defense system, facilitating contact with bacteria that are harmful to health. In addition to causing other harmful effects to health, such as the reduction of muscle functions, which can affect the heart, in addition to polluting water bodies, which affects the quality of the water.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
GSDMB	rs7224129	AG+	A	+ -	
GSDML	rs7216389	CT+	T	+ -	
IKZF3	rs9303277	CT+	T	+ -	
LRRC3C	rs3744246	CT+	T	+ -	
LRRC3C	rs4794820	AG+	G,T	+ -	

Allergies in General

MEDIUM

Tendency to allergic reactions in general.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FCER1A	rs2251746	TT+	C	- -	
HLA-DRA	rs7192	GG+	G	- -	
INTERGENIC	rs9275596	TT+	T	- -	
TGFB1	rs1800469	CT-	G	+ -	

Allergy to Pets

MEDIUM

Some people have allergies to domestic animals, such as dogs, rabbits or cats, which causes symptoms such as constant sneezing, dry cough or itchy nose, eyes and skin, whenever they come into contact with them, their objects or the area where the animal was.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
C11ORF30	rs2155219	TT+	T	+ +	
HLA	rs6902544	AA+	G	- -	
HLA-DQB1	rs7775228	TT+	C	- -	
IL-1RL1	rs10197862	AA+	G	- -	

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Eczema

NORMAL

Eczema is a type of dermatosis that is characterized by having several types of lesions. It can be acute, subacute or chronic. The acute version has lesions that start with reddish marks with water bubbles on the surface that, when they break, eliminate a clear liquid, which characterizes the subacute phase of eczema. In the chronic phase, the secretion starts to dry, leading to the formation of crusts. At this stage, an increase in skin thickness is also observed. The patient may have acute eczema, acute/subacute, subacute/chronic or just chronic. Thus, it is not necessary for the same patient to have all phases of an eczema to make a diagnosis of the dermatosis.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CLEC16A	rs2041733	TT+	C,G	- -	
INTERGENIC	rs2212434	TT+	T	- -	
INTERGENIC	rs2918307	AA+	G,T	- -	
INTERGENIC	rs4713555	GG+	T	- -	
INTERGENIC	rs7512552	CC+	C	- -	
INTERGENIC	rs61813875	CC+	G	- -	
OVOL1	rs10791824	AG+	G,T	+ -	
ZNF365	rs7076156	GG+	C,G	- -	

Allergy to mites

NORMAL

Allergy to mites translates into respiratory symptoms (rhinitis and/or asthma and/or conjunctivitis) and results from genetic predisposition.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HLA-DRA	rs1041885	AA-	T	- -	
IL-1RL1	rs3771175	TT+	A	- -	
IL-1RL1	rs10197862	AA+	G	- -	
INTERGENIC	rs10174949	GG+	A	- -	

Urticaria

NORMAL

Urticaria is a disease characterized by the appearance of erythematous, diffuse or localized papules, and was initially considered an essentially allergic condition.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FCER1A	rs2298805	GG+	A	- -	

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Allergy to pollen

LOW

Many people know pollen allergy as hay fever, but health experts often refer to it as "seasonal allergic rhinitis."

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HLA	rs6902544	AA+	G	- -	
HLA-DQB1	rs7775228	TT+	C	- -	
IL-1RL1	rs10197862	AA+	G	- -	
TSLP	rs1837253	CT+	C	+ -	

Allergy to Nuts

UNDEFINED

Along with peanuts and shellfish, walnuts are one of the food allergens most often linked to anaphylaxis.

Dermographism

UNDEFINED

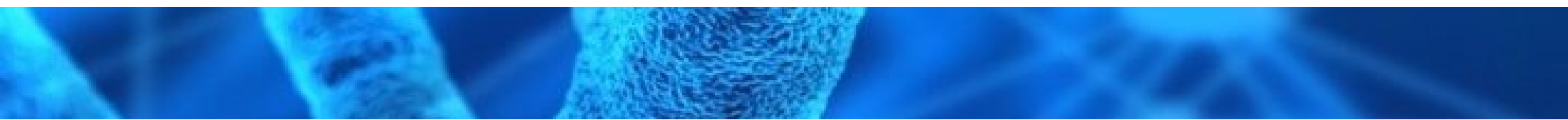
Dermographism is a type of skin allergy, characterized by the appearance of swelling after a stimulus caused by a scratch or contact on the skin, which may be accompanied by itching and redness in the surrounding region.

Mast Cell Activation Syndrome (MCAS)

UNDEFINED

MCAS is a condition in which the patient experiences repeated episodes of the symptoms of anaphylaxis – allergic symptoms such as hives, swelling, low blood pressure, difficulty breathing and severe diarrhea. High levels of mast cell mediators are released during those episodes.

Amino acids



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L-arginine

NORMAL

Arginine is a basic amino acid, also known as 2-amino-5-guanidino-pentanoic acid or l-arginine. Stimulates the production of lymphocytes (which play an important role in the body's defense). In addition, arginine helps to inhibit the growth of many types of tumors; stimulates growth hormone secretion, helps to heal wounds; inhibits the loss of muscle mass after surgery; treats liver problems and diseases; it helps in the production of sperm and the building of new bone and tendon cells, treating arthritis and connective tissue disorders. Arginine is still present in the process of synthesis of nitric oxide, which produces vasodilation, taking more blood and oxygen to tissues and organs, contributing to their health. Research has even shown that arginine is effective in reducing anxiety symptoms. Result in orange or red indicates greater need.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ASL	rs28941472	AA+	G	- -	
INTERGENIC	rs2545801	AG-	C	- -	
SLC7A2	rs56335308	GG+	A	- -	

Antioxidants / Supplements

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Sample

Coenzyme Q10

HIGH

Coenzyme Q10, also known as ubiquinone or ubidecarenone, is a molecule naturally produced by our body and is present in high amounts in the cells of the heart, liver, kidneys and pancreas. This molecule is important for the proper functioning of the body, performing several functions: It increases energy production, reducing fatigue (tiredness); It has an antioxidant effect, protecting cells from possible damage; Improves cardiovascular function; Stimulates the immune system; Promotes skin cell renewal, delaying skin aging. However, the production of coenzyme Q10 decreases with age: up to 20 or 30 years this molecule is released by our body in high amounts, but after that age, its production is increasingly reduced. Thus, it is important to compensate for this decrease with the intake of foods that contain the molecule, such as meat, fish and vegetable oils.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCB1	rs1128503	CT-	G	- -	
ABCB1	rs3213619	TT-	G	- -	
ACE	rs4343	AG+	A	+ -	
ADD1	rs4961	GT+	A,T	+ -	
ADRB2	rs1800888	CC+	T	- -	
APOE	rs7412	TT+	T	+ +	
APOE4	rs429358	TT+	C	- -	
ATP2B1	rs2681472	CT-	G	- -	
BAG3	rs2234962	CT+	C	- -	
BRAP	rs3782886	AA-	C	- -	
BTD	rs104893686	TT+	G	- -	
BTD	rs104893687	CC+	T	- -	
CAT	rs1001179	AA-	T	+ +	
CCL2	rs1024611	CT-	G	+ -	
CDH13	rs8055236	GG+	A,C,T	+ +	
CDKN2B-AS1	rs1011970	GT+	T	- -	
CDKN2B-AS1	rs1063192	TT-	A,T	- -	
CDKN2B-AS1	rs10116277	TT+	T	- -	
CDKN2B-AS1	rs10757272	TT+	T	+ +	
CDKN2B-AS1	rs10757274	GG+	G	+ +	
CELSR2	rs4970834	CC+	T	- -	
CETP	rs5882	AG+	A	+ -	
CETP	rs2303790	AA+	G	- -	
COQ2	rs121918230	AA-	C	- -	
COQ2	rs121918231	GG-	T	- -	
COQ2	rs121918232	AA-	C	- -	

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COQ2	rs121918233	GG-	T	- -	
COQ4	rs143441644	CC+	T	- -	
COQ6	rs189840848	CC+	G,T	- -	
COQ8A	rs119468004	GG+	A,C	- -	
COQ8A	rs119468005	CC+	T	- -	
COQ8A	rs145034527	CC+	T	- -	
COQ8A	rs387906298	DD+	G	- -	
COQ8A	rs387906299	II+		- -	
COQ8A	rs578189699	CC+	T	- -	
COQ8A	rs771578775	CC+	T	- -	
COQ9	rs267606751	CC+	T	- -	
COQ9	rs786205897	II+		- -	
DMD	rs1800278	AA-	C	- -	
DMD	rs1801187	GG-	T	- -	
DNAJC5B	rs13279522	TT+	C	- -	
DPYD	rs1801266	CC-	A	- -	
DPYD	rs1801267	GG-	T	- -	
DPYD	rs1801268	GG-	A	- -	
DSG2	rs2230234	AA+	G,T	- -	
F12	rs1801020	CT-	G	+ -	
F7	rs6046	CT-	A	- -	
FMN2	rs17672135	TT+	C	+ +	
HLA-DRA	rs3135391	CC-	A	- -	
HLA-DRB1	rs660895	GG+	G	+ +	
IL-4	rs2243250	CC+	T	- -	
INTERGENIC	rs501120	AG-	C	+ -	
INTERGENIC	rs1333049	CC+	C	+ +	
INTERGENIC	rs2383207	GG+	G	+ +	
INTERGENIC	rs2943634	AC+	C,G	- -	
INTERGENIC	rs10757278	GG+	G	+ +	
ITGB3	rs5918	TT+	C	- -	
KCNE1	rs1805127	AG-	A,C,G	+ -	
KCNE2	rs2234916	AA+	G	- -	
KCNE3	rs2270676	CT-	G	- -	
KL	rs9536314	TT+	A,G	- -	

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LIPC	rs1800588	CC+	T	- -	
LPL	rs326	AG+	G	- -	
LRP8	rs5174	GG-	T	- -	
MAOA	rs909525	GG-	T	+ +	
MAOA	rs1137070	TT+	C	- -	
MAOA	rs3027399	GG+	C	- -	
MAT1A	rs118204001	TT-	C	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
MC4R	rs2229616	GG-	C	- -	
MMP3	rs3025058	DI+	G	+ -	
MRAS	rs2306374	TT+	C	- -	
MTHFD1	rs1076991	AG-	C,G	+ -	
MTHFD1L	rs6922269	AG+	A	+ -	
MTHFD1L	rs11754661	GG+	A,T	- -	
MTHFD1L	rs17349743	TT+	C	- -	
MYBPC3	rs11570112	CC-	A,C	- -	
NAF1	rs7675998	GG+	G,T	- -	
NOS3	rs1800779	AA+	G	- -	
NPPA	rs5065	AA+	G	- -	
NQO1	rs1800566	CC-	A	- -	
NQO2	rs1143684	TT+	C	- -	
PACERR	rs689466	AG-	C	+ -	
PDSS2	rs118203956	CC-	A	- -	
PHACTR1	rs9349379	AA+	A	- -	
PLPP3	rs17114036	AA+	G	- -	
PSMA6	rs1048990	CG+	G,T	- -	
PSRC1	rs599839	AA+	A,C	- -	
RYR2	rs34967813	AA+	G	- -	
SCN5A	rs1805124	GG-	T	- -	
SH2B3	rs3184504	CC+	A,C,G	- -	
SMAD3	rs17228212	TT+	C	- -	
STAT4	rs10181656	CG+	C	+ -	

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Sample

TLR4	rs4986790	AA+	G,T	- -	●
TLR4	rs4986791	CC+	T	- -	●
TMPO	rs17028450	CC+	T	- -	●
TNF	rs1800610	CT-	A	- -	●
TNF	rs1800629	GG+	A	- -	●
TNFSF4	rs1234313	AG+	G	+ -	●
TNFSF4	rs3861950	TT+	C	- -	●
TTN	rs2244492	GG-	T	- -	●
TXNRD2	rs5748469	AA+	A	- -	●

Cancer

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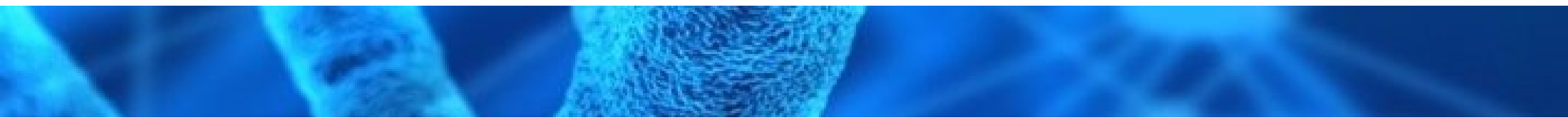
Sample

Squamous cell carcinoma

HIGH

Squamous cells (keratinocytes) are the main structural cells of the epidermis (outer layer of the skin). Squamous cell carcinoma is cancer of these cells. Each year in the United States, more than one million people are diagnosed with squamous cell carcinoma and 2,500 people die from it. Squamous cell carcinoma, the second most common type of skin cancer after basal cell carcinoma, usually develops in sun-exposed areas but can grow anywhere on the skin or in the mouth, where sun exposure is minimal. However, people who have had their skin more exposed to the sun are at greater risk of developing squamous cell skin cancer. Light-skinned people are much more susceptible to squamous cell carcinoma than dark-skinned people. Squamous cell carcinoma can appear on normal skin, but is more likely to appear on damaged skin. These damages include: Pre-cancerous skin tumors caused by previous sun exposure (actinic keratoses), Chronic ulcers on the skin or mucous membranes (such as those lining the eyes, nose, and lungs) or genitals, Scarred skin, mainly from burns.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BRAF	rs121913357	GG-	G	- -	
BRCA2	rs11571833	AA+	T	- -	
CDKN2A	rs587778189	AA-	A	- -	
DENND5A	rs7939541	CT+	C	+ -	
IRF4	rs12203592	CC+	T	- -	
SLC45A2	rs16891982	CC+	C	+ +	
TP53	rs11540652	GG-	T	- -	
TP53	rs28934573	CC-	A	- -	
TP53	rs28934574	CC-	A	- -	
TP53	rs121912651	CC-	A	- -	
TP53	rs121912657	GG-	A	- -	
TP53	rs397516436	CC-	A	- -	
TP53	rs587778720	GG-	T	- -	
TP53	rs587780070	CC-	A	- -	
TP53	rs587781525	AA-	A	- -	
TP53	rs587782289	TT-	C	- -	
TP53	rs730882008	GG-	A	- -	
TP53	rs730882025	GG-	T	- -	
TP53	rs786201057	CC-	T	- -	
TP53	rs786201059	GG-	T	- -	
TYR	rs1126809	GG+	A	- -	



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Sample

Skin Neoplasm (light sensitivity)

MEDIUM

Exposure to light increasing the risk of Skin Cancer, taking into account the incidence of UV (ultraviolet) rays

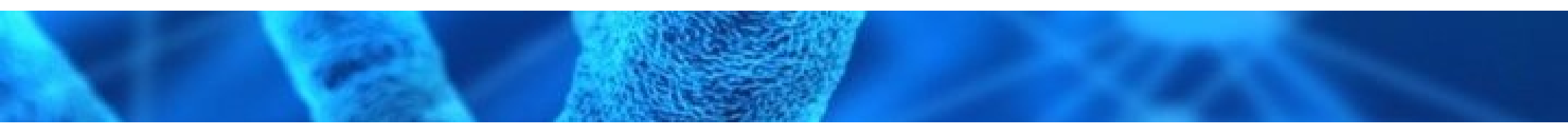
Gene	RSID	Genotype	Minor Allele	Alteration	Result
ASIP	rs1015362	GG-	T	+ +	
ASIP	rs4911414	GG+	G	- -	

Skin Neoplasm (Melanoma)

NORMAL

Melanoma is the most serious type of skin cancer.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BRAF	rs113488022	TT-	C,G,T	- -	
BRCA2	rs1799944	AA+	G	- -	
CDK4	rs11547328	CC-	A,T	- -	
CDKN2A	rs587776716	II+		- -	
ERCC2	rs13181	TT+	A,G	- -	
FTO	rs16953002	GG+	A	- -	
INTERGENIC	rs1408799	CT+	C	+ -	
MC1R	rs1110400	TT+	C	- -	
MC1R	rs1805006	CC+	A,G	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs2228479	GG+	A,C	- -	
MCR1R	rs2228478	AG+	G	- -	
NCOA6	rs4911442	AA+	A	- -	
OGG1	rs1052133	GG+	G	+ +	
PIGU	rs910873	GG+	A,C	- -	
TYR	rs1393350	GG+	A	- -	



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Sample

BAP1 - Tumor Predisposition Syndrome

NORMAL

Patients with germline mutations in BAP1 can develop several atypical intradermal tumors with mutations in BAP1, melanocytic and flesh color (MBAITs). These tumors generally develop earlier than other BAP1-associated tumors, highlighting an important role for dermatologists in identifying and screening patients with a history suggestive of germline mutation. Mutations of the BAP1 gene cause Tumor Predisposition Syndrome. People with this condition are at increased risk of developing many types of tumors, both benign and malignant, particularly certain skin tumors (atypical Spitz tumors, cutaneous melanoma, and basal cell carcinoma); eyes (uveal melanoma); kidneys (clear cell renal cell carcinoma); of a tissue called mesothelium that lines the chest, abdomen and internal organs (malignant mesothelioma).

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BAP1	rs387906848	CC-	A	- -	

Invasive Squamous Cell Carcinoma

NORMAL

This form of skin cancer arises in squamous cells, which make up most of the upper layers of the skin (epidermis). Squamous cell carcinomas can occur in all parts of the body, including the mucous membranes and genitals, although they develop more in areas that are constantly exposed to the sun, such as the arms, legs, neck, face and scalp. Skin in these regions often shows signs of sun damage, such as wrinkling, changes in pigmentation, and loss of elasticity.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HRAS	rs104894228	GG-	A	- -	
TP53	rs587778720	GG-	T	- -	

Skin Neoplasm (Melanoma - Hereditary)

NORMAL

Hereditary genetic mutations that increase the risk of melanoma are often passed from one generation to another

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CDKN2A	rs3731249	GG-	A,G,T	- -	

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Sample

Cowden's Syndrome

NORMAL

Cowden's Syndrome is an inherited disorder characterized by multiple equal tumor growths called hamartomas. Almost everyone with this condition has hamartomas. These small, non-cancerous tumors are commonly found on the skin and mucous membranes (such as the mouth and nose), but can also occur in the intestinal tract and other parts of the body. People with Cowden syndrome are at increased risk of developing a variety of cancers, including cancers of the breast, thyroid, and uterus. Women with Cowden syndrome have up to a 25-50% lifetime risk of developing breast cancer. (Robbins & Cotran (2004). Pathological Basis of Disease, 7th Edition. Elsevier, 1134.) Non-cancerous breast tumors and thyroid disease are also common.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
PTEN	rs121909218	GG+	A	- -	
PTEN	rs121909219	CC+	A,T	- -	
PTEN	rs121909222	AA+	G	- -	
PTEN	rs121909223	TT+	C	- -	
PTEN	rs121909225	TT+	G	- -	
PTEN	rs121909226	TT+	C	- -	
PTEN	rs587776668	TT+	C,G	- -	
PTEN	rs587776675	GG+	T	- -	

Chronic Subclinical Inflammation

Sjogren's Syndrome

MEDIUM

Chronic inflammatory disease, in which the immune system attacks and damages various organs and structures of the body itself, especially salivary, lacrimal and skin glands. People with this syndrome may have dry eyes and dry mouth. However, depending on the case, a patient diagnosed with Sjögren's syndrome may also experience: Pain, swelling and stiffness in the joints, Swelling of the salivary glands - particularly the joint located behind the jaw and in front of the ears, Rashes or dry skin , Vaginal dryness, Dry and persistent cough, Prolonged fatigue.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IRF5	rs10488631	CT+	C	+ -	
STAT4	rs7574865	GT+	G	+ -	
STAT4	rs10168266	CT+	T	+ -	
STAT4	rs10181656	CG+	C	+ -	
TNFAIP3	rs5029939	CC+	G	- -	

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Sample

Frontal Fibrosing Alopecia

MEDIUM

Frontal Fibrosing Alopecia is a chronic inflammatory disease that causes permanent hair loss. After the 1990s it became a growing phenomenon worldwide, as the first case was described in 1994. The cause may involve genetic, hormonal, immunological and environmental factors. Most women who experience hair loss notice a gradual broadening of the part in their hair (female-pattern hair loss). An increasingly common type of hair loss in older women is a receding hairline, called frontal fibrosing alopecia (FFA). It's a form of lichen planopilaris.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CYP1B1	rs1800440	AG-	C	+ -	

Sarcoidosis

MEDIUM

Sarcoidosis is a disease characterized by the formation of abnormal aggregates of inflammatory cells called granulomas. The disease usually starts in the lungs, skin or lymph nodes. Although less common, it can affect the eyes, liver, heart and brain. However, it is possible to affect any organ.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ANXA11	rs7091565	TT+	T	- -	
BTNL2	rs2076530	AG-	C	+ -	
TNF	rs1800629	GG+	A	- -	

Behçet's Disease

NORMAL

Behçet's disease is a chronic inflammation of the blood vessels (vasculitis) that can cause painful sores in the mouth and genitals, skin lesions and eye problems. The joints, nervous system and digestive tract can also become inflamed.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IL-10	rs1800871	CT-	G	+ -	
IL-23R	rs1495965	AA-	T	- -	
PTPN22	rs2476601	GG+	G	- -	

Scleroderma

NORMAL

Scleroderma is a chronic inflammatory disease of the connective tissue, linked to autoimmune factors. Its main characteristic is the hardening (scleral) of the skin (dermia), which becomes thicker, brighter and darker in the affected areas.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
DNASE1L3	rs35677470	GG+	A	- -	
PTPN22	rs2476601	GG+	G	- -	

Digestive system

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Sample

Canker sores

NORMAL

Canker sores are lesions caused by an inflammatory process, usually oval-shaped and white or yellow in color. Although they do not represent something more serious, they are one of the main causes of discomfort and complaints related to problems in the mouth.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IL-1B	rs1143634	CT-	A	+ -	
IL-6	rs1800795	CG+	G	- -	
SELE	rs5361	AC-	G	+ -	
TNF	rs1799964	TT+	C	- -	

Fatty acids

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

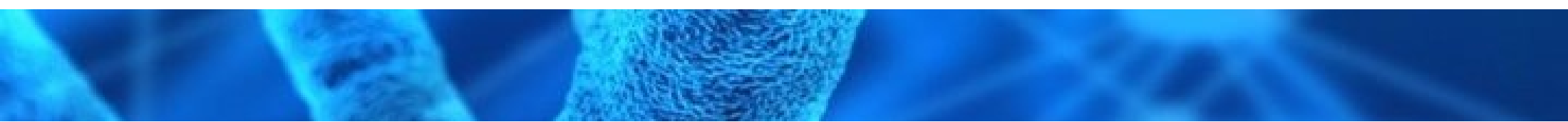
Sample

Omega 6

HIGH

Omega 6 mainly serves to reduce bad cholesterol (LDL) and triglycerides, helping to prevent cardiovascular disease, heart attack and stroke. The benefits of omega 6 are mainly related to its effect on blood vessels, preventing the formation of clots that can detach from the artery wall and obstruct blood flow (thrombosis), causing myocardial infarction (heart attack) and accident cerebral vascular ("stroke"). Omega 6 also prevents the deposition of fat (cholesterol) in artery walls, a condition called atherosclerosis, considered one of the main causes of myocardial infarction. In addition, omega 6 acts positively on the immune system, body temperature regulation and body water loss. Also known as linoleic acid, omega 6 is a polyunsaturated fatty acid, a type of "good" fat found in some foods in the form of oils. Like omega 3, omega 6 is considered essential as it is not produced by the body and therefore needs to be ingested through food. The main sources of omega 6 are sunflower, corn and soybean oils, nuts and walnuts.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCA1	rs1883025	AG-	T	+ -	
ABCB1	rs1128503	CT-	G	- -	
ABCB1	rs2032583	TT-	G	+ +	
ABCB1	rs2235015	GG-	A,T	+ +	
ABCB1	rs2235040	GG-	A,G,T	+ +	
ABCB1	rs2235067	GG-	T	+ +	
ABCB1	rs3213619	TT-	G	- -	
ABCB1	rs4148739	AA-	C	+ +	
ABCB1	rs10248420	AA+	G,T	+ +	
ABCB1	rs11983225	TT+	C	+ +	
ABCG2	rs2231137	GG-	T	+ +	
ABCG2	rs72552713	GG+	A	- -	
ABCG8	rs6544713	CC+	C	- -	
ACE	rs4343	AG+	A	+ -	
ACP7	rs472265	AA+	G	- -	
ADA	rs73598374	GG-	A,G,T	- -	
ADAMTS12	rs469568	TT-	C	+ +	
ADAMTS12	rs1364044	TT+	A,T	+ +	
ADCY5	rs11708067	AA+	G	- -	
ADD1	rs4961	GT+	A,T	+ -	
ADD1	rs4963	CG+	G,T	- -	
ADD2	rs3755351	CC-	T	- -	
ADIPOQ	rs17366743	TT+	C	- -	
ADRB2	rs1042714	GT+	C,T	- -	
ADRB2	rs1800888	CC+	T	- -	



Name Sample

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Prescriber | Health insurance

Sample

AGT	rs699	CT-	G	+ -	
AGTR1	rs5186	AA+	C	- -	
APOC1	rs4420638	AA+	G	- -	
APOE	rs7412	TT+	T	+ +	
APOE4	rs429358	TT+	C	- -	
AR	rs5031002	GG+	A	- -	
ATP2B1	rs2681472	CT-	G	- -	
ATP2B1	rs7965584	AG+	G	- -	
BAG3	rs2234962	CT+	C	- -	
BAG6	rs3117583	TT-	G	- -	
BDNF	rs6265	GG-	T	- -	
BRAP	rs3782886	AA-	C	- -	
BRCA2	rs4942486	CT+	C	+ -	
BTD	rs104893686	TT+	G	- -	
BTD	rs104893687	CC+	T	- -	
CAPN10	rs3792267	GG+	A	- -	
CASZ1	rs880315	AA-	C	- -	
CBS	rs2851391	CT+	C	+ -	
CBS	rs5742905	TT-	G	- -	
CBS	rs28934891	GG-	T	- -	
CCL2	rs1024611	CT-	G	+ -	
CDH13	rs8055236	GG+	A,C,T	+ +	
CDKAL1	rs4712523	AG+	G	- -	
CDKAL1	rs7756992	AG+	G,T	- -	
CDKN2A	rs10811661	TT+	T	+ +	
CDKN2A/B	rs2383208	AA+	G,T	- -	
CDKN2B-AS1	rs1011970	GT+	T	- -	
CDKN2B-AS1	rs1063192	TT-	A,T	- -	
CDKN2B-AS1	rs10116277	TT+	T	- -	
CDKN2B-AS1	rs10757272	TT+	T	+ +	
CDKN2B-AS1	rs10757274	GG+	G	+ +	
CETP	rs2303790	AA+	G	- -	
CHRM2	rs324650	AT+	A	+ -	
CHRM2	rs1824024	GT-	A	+ -	
CHRM2	rs2061174	CT-	A,C	- -	

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

CLOCK	rs1801260	AA+	C,G,T	- -	🟢
CPS1	rs1047891	CC+	A	- -	🟢
CR1L	rs4844614	GT+	T	+ -	🟡
CYP51A1	rs1041296	TT-	G	- -	🟢
DAPK1	rs11141915	AC+	C	- -	🟢
DMD	rs1800278	AA-	C	- -	🟢
DMD	rs1801187	GG-	T	- -	🟢
DNAH11	rs12670798	TT+	C	- -	🟢
DNAJC5B	rs13279522	TT+	C	- -	🟢
DNER	rs1861612	AG+	A,C	- -	🟢
DSG2	rs2230234	AA+	G,T	- -	🟢
F12	rs1801020	CT-	G	+ -	🟡
F13B	rs6003	AA-	T	- -	🟢
F2	rs1799963	GG+	A	- -	🟢
F5	rs4524	AG-	C	- -	🟢
F5	rs6025	GG-	T	- -	🟢
F5	rs7542281	CT+	T	- -	🟢
F7	rs6046	CT-	A	- -	🟢
FABP2	rs1799883	GG-	A,C,G	- -	🟢
FADS1	rs174546	CC+	T	- -	🟢
FADS2	rs174570	CC+	T	- -	🟢
FKBP5	rs1360780	CC+	A,C	- -	🟢
FMN2	rs17672135	TT+	C	+ +	🟠
FTO	rs1121980	CC-	A	- -	🟢
GAD1	rs701492	TT+	T	- -	🟡
GAD1	rs2241165	AA-	T	- -	🟢
GCK	rs4607517	GG+	A,C	- -	🟢
GCKR	rs780094	AG-	C	- -	🟢
GPHN	rs104894470	CC+	T	- -	🟢
GPX1	rs1050450	CC-	A	- -	🟢
HABP2	rs7080536	GG+	A	- -	🟢
HDAC9	rs2073963	GT+	G	+ -	🟠
HHEX	rs5015480	TT+	T	- -	🟢
HIVEP2	rs761993070	CC+	A,G,T	- -	🟢
HTR2A	rs6314	CC-	A	+ +	🟠

Name Sample

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Prescriber | Health insurance

Sample

HTR2A	rs1328674	AG-	C,G	- -	🟢
HTR2C	rs3813929	CC+	G,T	- -	🟢
IGF2BP2	rs4402960	GG+	T	- -	🟢
IL-1B	rs16944	AG+	G	+ -	🟡
IL-4	rs2243250	CC+	T	- -	🟢
INSIG2	rs7566605	GG+	C	- -	🟢
INTERGENIC	rs501120	AG-	C	+ -	🟡
INTERGENIC	rs751891	CC-	T	- -	🟢
INTERGENIC	rs791595	AG+	G	+ -	🟡
INTERGENIC	rs1333049	CC+	C	+ +	🔴
INTERGENIC	rs1545843	AG+	A	+ -	🟢
INTERGENIC	rs2383207	GG+	G	+ +	🟠
INTERGENIC	rs2820037	AA+	T	- -	🟢
INTERGENIC	rs7923837	AA+	A,T	- -	🟢
INTERGENIC	rs10757278	GG+	G	+ +	🔴
IRS1	rs2943641	CT+	C	+ -	🟡
ITGA11	rs7174755	TT+	C,G	- -	🟢
ITGB3	rs5918	TT+	C	- -	🟢
JAZF1	rs1635852	CT+	C	- -	🟢
KCNE1	rs1805127	AG-	A,C,G	+ -	🟡
KCNE2	rs2234916	AA+	G	- -	🟢
KCNE3	rs2270676	CT-	G	- -	🟢
KCNJ11	rs5215	TT+	T	- -	🟢
KCNJ11	rs5219	CC+	T	- -	🟢
KCNQ1	rs2283228	AA+	C	- -	🟢
KCNQ1	rs104894252	GG+	A,C	- -	🟢
KL	rs9536314	TT+	A,G	- -	🟢
LDLR	rs6511720	GG+	T	- -	🟢
LEPR	rs1137101	AA+	G	- -	🟢
LPL	rs268	AA+	G	- -	🟢
LRP8	rs5174	GG-	T	- -	🔵
MAOA	rs909525	GG-	T	+ +	🟠
MAOA	rs1137070	TT+	C	- -	🟢
MAOA	rs3027399	GG+	C	- -	🟢
MMP3	rs3025058	DI+	G	+ -	🟢

Name Sample

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Sample

MOV10	rs2932538	CC-	C,G	- -	🟢
MRAS	rs2306374	TT+	C	- -	🟢
MTHFD1	rs1076991	AG-	C,G	+ -	🟢
MTHFD1L	rs6922269	AG+	A	+ -	🟡
MTHFD1L	rs11754661	GG+	A,T	- -	🟢
MTHFD1L	rs17349743	TT+	C	- -	🟢
MTHFR	rs1476413	AG-	G,T	+ -	🟢
MTHFR	rs1801131	AC-	G	+ -	🟠
MTHFR	rs1801133	CC-	A	- -	🟢
MTNR1B	rs10830963	CC+	G	- -	🟢
MTRR	rs1801394	AA+	G	- -	🟢
MYBPC3	rs11570112	CC-	A,C	- -	🟢
NAA25	rs17696736	AA+	G	- -	🟢
NAF1	rs7675998	GG+	G,T	- -	🟢
NEDD4L	rs2288774	CT+	A,C	- -	🟢
NEDD4L	rs3865418	CC+	C	- -	🟢
NEDD4L	rs4149601	AA+	A	- -	🟢
NFE2L2	rs6721961	GG+	C,G	- -	🟢
NFE2L2	rs35652124	CT+	C	- -	🟢
NGF	rs6330	CC-	A	- -	🟢
NGF	rs11466112	CC-	A	- -	🟢
NINJ2	rs12425791	AG+	A,C	+ -	🟡
NOS3	rs1799983	GG+	T	- -	🟢
NOS3	rs1800779	AA+	G	- -	🟢
NOV	rs2071518	CC+	T	- -	🟢
NPPA	rs5065	AA+	G	- -	🟢
OPRM1	rs1799971	GG+	G	+ +	🔴
OXTR	rs237899	GG+	A,C	- -	🟢
OXTR	rs2254298	GG+	A	- -	🟢
PAX4	rs2233580	GG-	T	- -	🟢
PDE4D	rs966221	CT-	G	- -	🟢
PEX5L	rs7630877	GG+	A,C	- -	🟢
PHACTR1	rs9349379	AA+	A	- -	🟢
PITX2	rs2200733	CT+	T	+ -	🟠
PITX2	rs12646447	CT+	C	+ -	🟡

PRECISION HEALTH PRECISYA

Name Sample

Age

Gender F

Report date 12/09/2025

Prescriber

Health insurance

Sample

PLPP3	rs17114036	AA+	G	- -	
PPARG	rs1801282	CG+	C	+ -	
PPARGC1A	rs8192678	GG-	T	- -	
PSMA6	rs1048990	CG+	G,T	- -	
PSRC1	rs599839	AA+	A,C	- -	
RASGRP1	rs7403531	CC+	C,G	- -	
RHOU	rs6426514	GG+	A	- -	
RPSAP52	rs1531343	CC+	C,T	- -	
RYR2	rs34967813	AA+	G	- -	
SCN5A	rs1805124	GG-	T	- -	
SDHAF4	rs1048886	AA+	G	- -	
SELE	rs5361	AC-	G	- -	
SH2B3	rs3184504	CC+	A,C,G	- -	
SHMT1	rs1979277	AG+	A	- -	
SLC30A8	rs13266634	CC+	A,T	+ +	
SMAD3	rs17228212	TT+	C	- -	
SOD2	rs4880	CT-	G	- -	
SPSB4	rs16851055	GG+	A	- -	
STK39	rs6749447	GT+	G	+ -	
SUPT3H	rs556621	AC-	G	+ -	
TAP2	rs241428	AA-	A,C,G	- -	
TAP2	rs1800454	GG-	T	- -	
TCF7L2	rs7901695	TT+	C	- -	
TCF7L2	rs7903146	CC+	G,T	- -	
TCF7L2	rs12255372	GG+	T	- -	
TGFBR3	rs1805110	CC-	A	- -	
THADA	rs7578597	TT+	C	- -	
THBD	rs1042580	AG-	C	- -	
TLR4	rs4986790	AA+	G,T	- -	
TLR4	rs4986791	CC+	T	- -	
TMPO	rs17028450	CC+	T	- -	
TNF	rs361525	GG+	A	+ +	
TNF	rs1800629	GG+	A	- -	
TNFSF4	rs1234313	AG+	G	+ -	
TNFSF4	rs3861950	TT+	C	- -	

Name Sample

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Sample

TPH1	rs1799913	AC-	A,T	- -	●
TRIB3	rs2295490	AG+	G,T	+ -	●
TRIM29	rs2084898	GG+	A	- -	●
TRPM6	rs11144134	CT+	C	+ -	●
TTN	rs2244492	GG-	T	- -	●
TXNRD2	rs5748469	AA+	A	- -	●
UBE2E2	rs7612463	AC+	A,G	- -	●
VCAN	rs173686	CT-	G,T	+ -	●
VPS26A	rs4812829	AG+	A	- -	●
WDR12	rs6725887	TT+	C	- -	●
WSCD2	rs9739493	CC+	C,G	- -	●
ZFHX3	rs7193343	CC+	A,C	- -	●

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

Omega 3

HIGH

Omega-3s are nutrients you get from food (or supplements) that help build and maintain a healthy body. They're key to the structure of every cell wall you have. They're also an energy source and help keep your heart, lungs, blood vessels, and immune system working the way they should. Two crucial ones -- EPA and DHA -- are primarily found in certain fish. ALA (alpha-linolenic acid), another omega-3 fatty acid, is found in plant sources such as nuts and seeds. DHA levels are especially high in retina (eye), brain, and sperm cells. Not only does your body need these fatty acids to function, they also deliver some big health benefits. fish oil can lower elevated triglyceride levels. Having high levels of this blood fat puts you at risk for heart disease and stroke. Also fish oil supplements (EPA+DHA) may curb stiffness and joint pain. Omega-3 supplements also seem to boost the effectiveness of anti-inflammatory drugs. Omega-3s build up cell walls, and when levels are too low, you may experience dry skin, brittle hair, and thin nails that peel and crack. Omega-3 deficiency can cause rashes on the skin and dandruff as well. Fatigue and trouble sleeping. Symptoms of omega-3 fatty acid deficiency include fatigue, poor memory, dry skin, heart problems, mood swings or depression, and poor circulation. It is important to have the proper ratio of omega-3 and omega-6 (another essential fatty acid) in the diet.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCA1	rs1883025	AG-	T	+ -	
ADRB2	rs1042713	GG+	A	+ +	
ADRB2	rs1800888	CC+	T	- -	
AGT	rs699	CT-	G	+ -	
APOA1	rs670	AG-	T	+ -	
APOA5	rs651821	TT+	T	- -	
APOA5	rs662799	AA+	T	- -	
APOA5	rs2266788	TT-	A	- -	
APOA5	rs3135506	GG+	C	- -	
APOC3	rs5128	CG-	G	+ -	
APOE	rs7412	TT+	T	+ +	
APOE4	rs429358	TT+	C	- -	
ARMS2	rs10490924	GT+	T	+ -	
BRAP	rs3782886	AA-	C	- -	
C2	rs547154	CC-	T	- -	
C2	rs9332739	GG+	A,C	- -	
C3	rs2230199	CG-	C,T	+ -	
C3	rs2230205	AG-	T	- -	
CDKN2B-AS1	rs10116277	TT+	T	- -	
CETP	rs5880	GG+	C	- -	
CETP	rs5882	AG+	A	+ -	
CETP	rs183130	TT+	T	+ +	
CETP	rs708272	CT-	A	+ -	

Name Sample

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Prescriber | Health insurance

Sample

CETP	rs1532624	TT-	A	- -	🟢
CETP	rs1864163	GG+	A	- -	🔵
CETP	rs2303790	AA+	G	- -	🟢
CETP	rs4783961	AA+	A	- -	🟢
CFB	rs4151667	TT+	A	- -	🟢
CFH	rs800292	CT-	A	+ -	🔵
CFH	rs1061147	CC+	C	- -	🔵
CFH	rs1061170	TT+	T	- -	🔵
CFH	rs1065489	GT+	T	- -	🟢
CFH	rs1329428	AG-	T	- -	🟢
CFH	rs3753394	CT+	T	- -	🟢
COL8A1	rs13081855	GG+	T	- -	🟢
CRYBB3	rs74315490	GG+		- -	🟢
CX3CR1	rs3732378	AG+	A	+ -	🔵
CX3CR1	rs3732379	CT+	T	- -	🟢
DOCK7	rs10889353	AC+	C,T	+ -	🟢
EPHA2	rs3754334	CC-	A	- -	🟢
EPHA2	rs116506614	CC+		- -	🟢
F12	rs1801020	CT-	G	+ -	🟡
F7	rs6046	CT-	A	- -	🟢
FADS1	rs174546	CC+	T	+ +	🔴
FADS1	rs174547	TT+	C	- -	🟢
FADS2	rs174570	CC+	T	- -	🟢
GCKR	rs1260326	CT+	C	+ -	🟡
HTRA1	rs11200638	AG+	A	+ -	🟠
IL-4	rs2243250	CC+	T	- -	🟢
IL-6	rs1800795	CG+	G	+ -	🟢
IL-6	rs2069845	AG+	G	+ -	🔴
INTERGENIC	rs493258	AG-	C	+ -	🟠
INTERGENIC	rs1333049	CC+	C	+ +	🔴
INTERGENIC	rs2383207	GG+	G	+ +	🟠
INTERGENIC	rs10757278	GG+	G	+ +	🔴
ITGB3	rs5918	TT+	C	- -	🟢
LPL	rs285	CT+	T	- -	🟢
LPL	rs320	GT+	G	+ -	🟢

Name Sample

Age

Gender F

Report date 12/09/2025

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Health insurance

Sample

LPL	rs13702	AG-	A,C	+ -	
LRP8	rs5174	GG-	T	- -	
MMP3	rs3025058	DI+	G	+ -	
MTHFR	rs1801131	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	
MYRF	rs174537	GG+	T	- -	
NOS3	rs1799983	GG+	T	- -	
NOS3	rs1800779	AA+	G	- -	
NOTCH4	rs422951	AG-	C	- -	
OR4A46P	rs7395662	GG+	A	- -	
OXTR	rs2254298	GG+	A	- -	
PCIF1	rs7679	TT+	C	- -	
PITX2	rs6533526	GG+	A	- -	
PPARA	rs1800206	CC+	G	- -	
PSMA6	rs1048990	CG+	G,T	- -	
PSRC1	rs599839	AA+	A,C	- -	
RAB11B	rs2967605	GG-	T	- -	
REST	rs1713985	AC-	T	- -	
REST	rs2227902	GG+	T	- -	
SERPINF1	rs1136287	CT+	T	+ -	
SH2B3	rs3184504	CC+	A,C,G	- -	
SKIV2L	rs429608	GG+	A	- -	
SKIV2L	rs2734331	TT-	G	- -	
TLR3	rs3775291	GG-	G,T	+ +	
TLR4	rs4986790	AA+	G,T	- -	
TMEM241	rs9949617	CC+	T	- -	
TNF	rs361525	GG+	A	+ +	
TNF	rs1800629	GG+	A	- -	
TNFSF4	rs1234313	AG+	G	+ -	
TNFSF4	rs3861950	TT+	C	- -	
VEGFA	rs3025039	CC+	T	- -	
ZPR1	rs964184	CC+	C	- -	

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

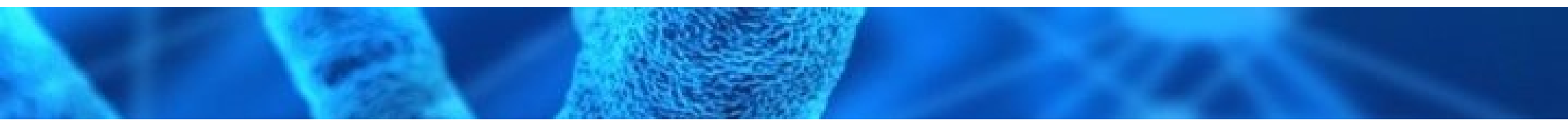
Sample

Omega 9

HIGH

Omega 9 or omega 9 (ω-9 fatty acids) are fatty acids, aid in human development, as do omega 3 and omega 6 fatty acids. The name omega 9 means they have a C=C double bond on the ninth carbon from the end opposite the carboxyl. We can find it in vegetable oils. The most important omega 9 acids are: oleic acid - with 18 carbons, erucic acid - with 22 carbons, nervonic acid - with 24 carbons

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCA1	rs1883025	AG-	T	+ -	
ABCG2	rs2231137	GG-	T	+ +	
ABCG2	rs72552713	GG+	A	- -	
ABCG8	rs6544713	CC+	C	- -	
ACE	rs4343	AG+	A	+ -	
ADAMTS12	rs469568	TT-	C	+ +	
ADAMTS12	rs1364044	TT+	A,T	+ +	
ADD1	rs4961	GT+	A,T	+ -	
ADIPOQ	rs17366743	TT+	C	- -	
ADRB2	rs1800888	CC+	T	- -	
APOA5	rs2266788	TT-	A	- -	
APOA5	rs3135506	GG+	C	- -	
APOC1	rs4420638	AA+	G	- -	
APOE	rs7412	TT+	T	+ +	
APOE4	rs429358	TT+	C	- -	
AR	rs5031002	GG+	A	- -	
ATP2B1	rs2681472	CT-	G	- -	
BAG3	rs2234962	CT+	C	- -	
BRAP	rs3782886	AA-	C	- -	
BRCA2	rs4942486	CT+	C	+ -	
BTD	rs104893686	TT+	G	- -	
BTD	rs104893687	CC+	T	- -	
CCL2	rs1024611	CT-	G	+ -	
CDH13	rs8055236	GG+	A,C,T	+ +	
CDKN2B-AS1	rs1011970	GT+	T	- -	
CDKN2B-AS1	rs1063192	TT-	A,T	- -	
CDKN2B-AS1	rs10116277	TT+	T	- -	
CDKN2B-AS1	rs10757272	TT+	T	+ +	
CDKN2B-AS1	rs10757274	GG+	G	+ +	



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CETP	rs2303790	AA+	G	- -	🟢
CPS1	rs1047891	CC+	A	- -	🟢
CR1L	rs4844614	GT+	T	+ -	🟡
CYP51A1	rs1041296	TT-	G	- -	🟢
DMD	rs1800278	AA-	C	- -	🟢
DMD	rs1801187	GG-	T	- -	🟢
DNAH11	rs12670798	TT+	C	- -	🟢
DNAJC5B	rs13279522	TT+	C	- -	🟢
DOCK7	rs10889353	AC+	C,T	+ -	🟢
DSG2	rs2230234	AA+	G,T	- -	🟢
F12	rs1801020	CT-	G	+ -	🟡
F5	rs6025	GG-	T	- -	🟢
F5	rs7542281	CT+	T	- -	🟢
F7	rs6046	CT-	A	- -	🟢
FABP2	rs1799883	GG-	A,C,G	- -	🟢
FADS1	rs174547	TT+	C	- -	🟢
FADS2	rs174570	CC+	T	- -	🟢
FMN2	rs17672135	TT+	C	+ +	🟠
GCKR	rs1260326	CT+	C	+ -	🟡
GPX1	rs1050450	CC-	A	- -	🟢
HABP2	rs7080536	GG+	A	- -	🟢
HDAC9	rs2073963	GT+	G	+ -	🟠
IL-4	rs2243250	CC+	T	- -	🟢
IL-6	rs1800795	CG+	G	+ -	🟢
INTERGENIC	rs501120	AG-	C	+ -	🟡
INTERGENIC	rs1333049	CC+	C	+ +	🔴
INTERGENIC	rs2383207	GG+	G	+ +	🟠
INTERGENIC	rs10757278	GG+	G	+ +	🔴
ITGB3	rs5918	TT+	C	- -	🟢
KCNE1	rs1805127	AG-	A,C,G	+ -	🟡
KCNE2	rs2234916	AA+	G	- -	🟢
KCNE3	rs2270676	CT-	G	- -	🟢
KL	rs9536314	TT+	A,G	- -	🟢
LDLR	rs6511720	GG+	T	- -	🟢
LPL	rs285	CT+	T	- -	🟢

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LPL	rs320	GT+	G	+ -	🟢
LPL	rs13702	AG-	A,C	+ -	🟢
LRP8	rs5174	GG-	T	- -	🔵
MAOA	rs909525	GG-	T	+ +	🟠
MAOA	rs1137070	TT+	C	- -	🟢
MAOA	rs3027399	GG+	C	- -	🟢
MMP3	rs3025058	DI+	G	+ -	🟢
MRAS	rs2306374	TT+	C	- -	🟢
MTHFD1	rs1076991	AG-	C,G	+ -	🟢
MTHFD1L	rs6922269	AG+	A	+ -	🟡
MTHFD1L	rs11754661	GG+	A,T	- -	🟢
MTHFD1L	rs17349743	TT+	C	- -	🟢
MTRR	rs1801394	AA+	G	- -	🟢
MYBPC3	rs11570112	CC-	A,C	- -	🟢
NAA25	rs17696736	AA+	G	- -	🟢
NAF1	rs7675998	GG+	G,T	- -	🟢
NINJ2	rs12425791	AG+	A,C	+ -	🟡
NOS3	rs1799983	GG+	T	- -	🟢
NOS3	rs1800779	AA+	G	- -	🟢
NPPA	rs5065	AA+	G	- -	🟢
OR4A46P	rs7395662	GG+	A	- -	🟢
PCIF1	rs7679	TT+	C	- -	🟢
PDE4D	rs966221	CT-	G	- -	🟢
PHACTR1	rs9349379	AA+	A	- -	🟢
PITX2	rs2200733	CT+	T	+ -	🟠
PITX2	rs12646447	CT+	C	+ -	🟡
PLPP3	rs17114036	AA+	G	- -	🟢
PSMA6	rs1048990	CG+	G,T	- -	🟢
PSRC1	rs599839	AA+	A,C	- -	🟡
RAB11B	rs2967605	GG-	T	- -	🟢
RYR2	rs34967813	AA+	G	- -	🟢
SCN5A	rs1805124	GG-	T	- -	🟢
SH2B3	rs3184504	CC+	A,C,G	- -	🟢
SMAD3	rs17228212	TT+	C	- -	🟢
SPSB4	rs16851055	GG+	A	- -	🟢

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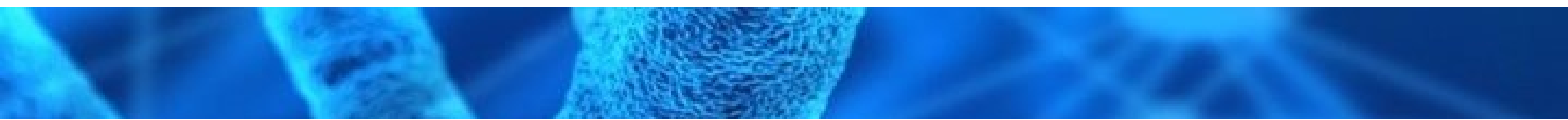
SUPT3H	rs556621	AC-	G	+ -	
THBD	rs1042580	AG-	C	- -	
TLR4	rs4986790	AA+	G,T	- -	
TLR4	rs4986791	CC+	T	- -	
TMEM241	rs9949617	CC+	T	- -	
TMPO	rs17028450	CC+	T	- -	
TNF	rs1800629	GG+	A	- -	
TNFSF4	rs1234313	AG+	G	+ -	
TNFSF4	rs3861950	TT+	C	- -	
TRIM29	rs2084898	GG+	A	- -	
TTN	rs2244492	GG-	T	- -	
TXNRD2	rs5748469	AA+	A	- -	
VCAN	rs173686	CT-	G,T	+ -	
WDR12	rs6725887	TT+	C	- -	
ZFHX3	rs7193343	CC+	A,C	- -	
ZPR1	rs964184	CC+	C	- -	

General



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Sample

Loss of hair

MEDIUM

Losing hair is normal. About 50 to 100 strands of hair fall out of everyone's head daily - which is not much compared to the 100,000+ strands on our scalp. As we get older, our hair strands become thinner and thinner. However, many people experience more severe hair loss, which could be a sign of a more serious health problem. But, even in very severe cases, those who suffer from hair loss have no reason to panic: this condition does not lead to baldness, as all the hair that falls out will be replaced by new hairs.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AR	rs6152	GG+	A	- -	
C1ORF127	rs2003046	CC+	A,G	- -	
CYP19A1	rs6493497	AG+	A	+ -	
CYP19A1	rs7176005	CT+	T	+ -	
EDA2R	rs1385699	TT+	T	- -	
HDAC9	rs2073963	GT+	G	+ -	
HR	rs7014851	TT+	C	- -	
IL-1B	rs1143634	CT-	A	- -	
INTERGENIC	rs925391	CC-	G,T	- -	
INTERGENIC	rs2180439	CC+	T	- -	
INTERGENIC	rs2223841	AA-	C	- -	
INTERGENIC	rs9287638	CC+	A	- -	
LINC01432	rs1160312	GG+	G	- -	

Genetic diseases

Oculocutaneous Albinism

MEDIUM

Human oculocutaneous albinism is a group of conditions that affect the pigmentation of hair, skin, and eyes.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MC1R	rs1805008	CC+	T	- -	
SLC45A2	rs26722	CT+	T	+ -	

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Sample

Proteus Syndrome

NORMAL

Proteus syndrome is a very rare congenital disease that causes pathological overgrowth of the skin with subcutaneous tumors, atypical development with macrodactyly and hemihypertrophy. It is an extremely rare disease: around 101 cases have been described worldwide. Because of this rarity, there are not many studies in the area today, and almost all the events, practically, have no solution. Features: partial gigantism of the hands and feet, pigmented nevus, hemihypertrophy, subcutaneous tumors, macrocephaly and other cranial and visceral anomalies.

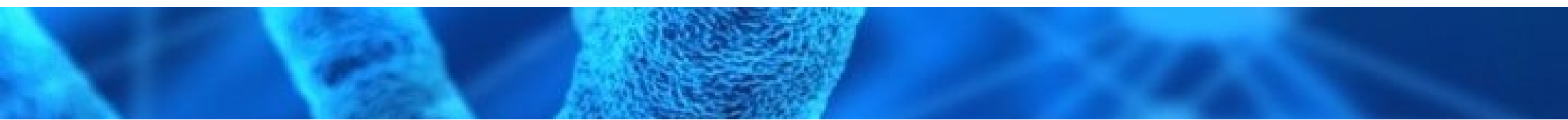
Gene	RSID	Genotype	Minor Allele	Alteration	Result
AKT1	rs2494732	TT+	C	- -	

Hereditary angioedema

NORMAL

It is a genetic, hereditary disease that affects both sexes. HAE patients have recurrent episodes of swelling throughout their lives, known by the term angioedema (angio = blood vessel and edema = swelling)

Gene	RSID	Genotype	Minor Allele	Alteration	Result
F12	rs1801020	CT-	G	+ -	
SERPING1	rs4926	AG+		- -	
SERPING1	rs28940870	CC+		- -	



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Bart-Pumphrey Syndrome

NORMAL

Bart-Pumphrey syndrome is characterized by nail and skin abnormalities and hearing loss. People with Bart-Pumphrey syndrome often have a white discoloration of the nails (leukonychia); nails can also be thick and brittle. Affected individuals often have wart-like (verrucous) skin growths called joints in the joints of the fingers and toes. They may also have thickened skin on the palms of the hands and soles of the feet (palmoplantar keratoderma). Skin abnormalities often become visible during childhood. Hearing loss associated with Bart-Pumphrey syndrome ranges from moderate to profound and is usually present from birth (congenital). The signs and symptoms of this disorder can vary even within the same family; while almost all affected individuals have hearing loss.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
GJB2	rs1801002	GG-	A	- -	
GJB2	rs2274084	CC+		- -	
GJB2	rs28931593	GG-	A	- -	
GJB2	rs28931594	GG-	T	- -	
GJB2	rs28931595	GG-	A,G,T	- -	
GJB2	rs35887622	AA+	C	- -	
GJB2	rs104894395	GG-	A	- -	
GJB2	rs104894396	GG-	A	- -	
GJB2	rs104894397	TT-	C	- -	
GJB2	rs104894398	GG-	T	- -	
GJB2	rs104894401	GG-	A	- -	
GJB2	rs104894402	CC-	T	- -	
GJB2	rs104894404	GG-	A	- -	
GJB2	rs104894407	GG-	A	- -	
GJB2	rs104894408	GG-		- -	
GJB2	rs104894409	GG-	A	- -	
GJB2	rs104894413	GG-	A	- -	
GJB2	rs111033190	GG-	A	- -	
GJB2	rs111033204	---		- -	
GJB2	rs111033217	AA-	C	- -	
GJB2	rs111033222	GG-	A	- -	
GJB2	rs111033253	---		- -	
GJB2	rs111033293	AA-	G	- -	
GJB2	rs111033294	AA-	G	- -	
GJB2	rs111033295	AA-	C	- -	
GJB2	rs111033296	CC-	A	- -	
GJB2	rs111033297	CC-	T	- -	

GJB2	rs111033299	GG-	A	- -	
GJB2	rs111033335	AA-		- -	
GJB2	rs111033401	GG-	A	- -	
GJB2	rs111033420	CC-	A	- -	
GJB2	rs111033451	CC-	T	- -	
GJB2	rs143343083	GG+	A	- -	
GJB2	rs150529554	CC+	T	- -	
GJB2	rs371024165	GG+	A	- -	
GJB2	rs397516874	CC-	T	- -	

Hypohydrotic Ectodermal Dysplasia

NORMAL

It is a genetic disease of X-linked recessive inheritance. People who suffer from this disease have few cone-shaped teeth, little hair, frailty, lack of eyebrows and do not sweat.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
EDAR	rs3827760	TT-	G	- -	
NFKBIA	rs8904	CC-	A,C,T	- -	

Ictiosis Vulgaris

NORMAL

It is a condition associated with an excessive accumulation of keratin that results in a scale-like appearance of the skin (ichthyus - fish). There is an accumulation of the stratum corneum with little or no inflammation. It can be genetically inherited in an autosomal dominant or acquired form. The two forms are similar, however, the acquired form is accompanied by systemic factors. The mutation occurs in the FLG gene, which encodes the epidermal protein filaggrin, responsible for the aggregation of keratin intermediate filaments. Clinically, there are thin scales that predominate in the extensor areas of the lower limbs. There is an association with atopy as well as with keratosis pilaris.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FLG	rs797045090	CC-	A	- -	

Elastic pseudoxanthoma

NORMAL

Rare genetic disease characterized by progressive calcification and fragmentation of elastic fibers in the skin, retina, and cardiovascular system.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCC6	rs2238472	GG-	T	- -	

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Cardio-facio-cutaneous syndrome

NORMAL

This syndrome is a genetic disorder characterized by multiple congenital anomalies and mental retardation. It affects various parts of the body such as the heart, face, skin and hair.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BRAF	rs121913355	GG-	A,G,T	- -	
MAP2K1	rs727504317	GG+	A	- -	

Blau's Syndrome

NORMAL

Blau's Syndrome is the term used for the familial forms of the disease, but sporadic forms that are known as Early Onset Sarcoidosis can also occur. It is a genetic disease whose patients suffer from a combination of dermatitis, arthritis and uveitis. Other organs may also be affected and there may be intermittent fever.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
NOD2	rs2066844	CC+	T	- -	
NOD2	rs2066845	GG+	C,T	- -	

Hematologic system

Hyperbilirubinemia

MEDIUM

Hyperbilirubinemia is divided into two types: unconjugated (indirect) and conjugated (direct). The diagnosis of conjugated hyperbilirubinemia can be confirmed by detecting bilirubin in urine. Unconjugated hyperbilirubin can be the result of hemolytic anemia, large hematomas, genetic conditions such as Gilbert's syndrome, long-term failure to eat, neonatal jaundice, or thyroid disease. Conjugated hyperbilirubin can be the result of liver disease such as cirrhosis or hepatitis, infections, some medications, or blockage of the bile duct.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
TKTL1	rs766420	GG-	G	+ +	
UGT1A1	rs887829	GG-	T	- -	
UGT1A1	rs6742078	GG+	T	- -	

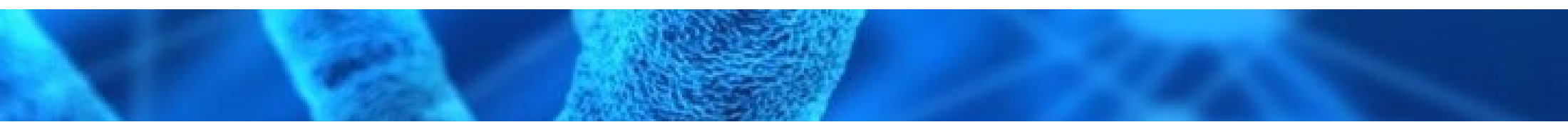
Post-Transfusion Purpura (PPT)

NORMAL

It is a late transfusion reaction, uncommon, self-limited to a few days or weeks, which occurs mainly in multiparous and/or previously transfused women, causing severe thrombocytopenia

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ITGB3	rs5918	TT+	C	- -	

Immune system



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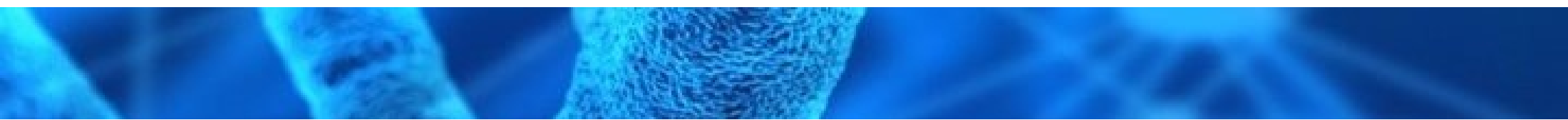
Dermatomyositis

HIGH

Dermatomyositis is a chronic inflammatory disease that affects the muscles. The most evident symptoms are muscle weakness that worsens over time and skin lesions, such as violet spots on the eyelids and scaly formations on the fingers, elbows and knees, called Gottron papules.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CCL2	rs4586	CC+	C	+ +	
IFIH1	rs1990760	CC+	T	- -	



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Psoriasis

MEDIUM

A disease in which skin cells accumulate and form scales and dry patches that cause itching.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FASLG	rs763110	TT+	T	- -	
FASLG	rs80358237	CC+	A	- -	
HLA-C	rs1265181	CC-	C	- -	
HLA-C	rs10484554	CC+	T	- -	
IL-12B	rs3212227	CC-	G	+ +	
IL-12B	rs3213094	AA-	C,T	- -	
IL-13	rs20541	CC-	G	- -	
IL-23R	rs2201841	CT-	G,T	+ -	
IL-23R	rs11209026	GG+	A	+ +	
INTERGENIC	rs2546890	GG+	G	- -	
INTERGENIC	rs4085613	AC-	G	+ -	
INTERGENIC	rs4112788	CT-	G,T	+ -	
INTERGENIC	rs4406273	GG+	A	- -	
INTERGENIC	rs6887695	CC+	G	- -	
KLRK1	rs1049174	CG-	C	- -	
POU5F1	rs1265159	CC-	A,C,T	- -	
PSORS1C3	rs887466	TT-	A,G	- -	
QTRT1	rs892085	CT-	A	- -	
STAT2	rs2066807	GG-	G	- -	
STAT2	rs2066808	TT-	G	- -	
TNF	rs361525	GG+	A	- -	
TNF	rs1800629	GG+	A	- -	
TNFAIP3	rs610604	AC-	G,T	+ -	
TNFAIP3	rs2230926	TT+	C,G	- -	
TRIM47	rs1055129	TT-	G	- -	
ZNF365	rs2393903	AG-	A,C	- -	
ZNF365	rs7076156	GG+	C,G	- -	
ZNF365	rs7089814	TT+	G,T	+ +	

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Lichen Planus

NORMAL

Lichen planus (Lichen planus) is a disease that affects mucosa and skin and has a chronic inflammatory nature, affecting about 2% of the population and affecting women over 40 years of age in a more usual way.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HMGB1	rs1045411	GG-	A	- -	
INTERGENIC	rs9461799	TT+	C	- -	

Congenital Erythropoietic Porphyria (Gunther's Disease)

NORMAL

Also known as Gunther's Disease, Congenital Erythropoietic Porphyria (CEP) causes serious damage to sun-exposed skin. The disease also causes excessive hair growth and light or dark patches on the skin. There are eight types of Porphyrias, a disease characterized by a defect in the "manufacture" of heme, a fundamental molecule of the organism. Heme can be synthesized in the liver or bone marrow. In the case of CEP, the problem causes skin photosensitivity and accumulation of heme in red blood cells, skin and bones. The disease usually manifests itself early in childhood, but EPC can also manifest later. Congenital Porphyria Erythropoietica is a disease of inborn error of metabolism, genetically and hereditary transmitted by a recessive mutation. That is, parents who have this mutation of the disease (but are asymptomatic) have 25% of having a child who develops it. Signs and Symptoms: The formation of blisters and ulcers on the skin in case of exposure to sunlight or even intense artificial light is usually the main symptom of CEP. The skin may have dark or light patches. It is also common for urine to turn red, due to the elimination of excess porphyrins. The eyes can also become very sensitive to light, leading to sores and falling eyelashes. Anemia, enlarged spleen and excess hair on the face and back of the hands, and darkening of the teeth are also reported.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FECH	rs3848519	CC+	A,T	- -	
UROS	rs28941775	AA-	C	- -	

Herpes

UNDEFINED

A virus that causes contagious sores, most often around the mouth or on Organs genitals.

Metabolic

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Glycation

MEDIUM-HIGH

Glycation is a process that joins a glucose molecule with a protein molecule, such as collagen and elastin - the same ones responsible for keeping the skin younger and firmer. This union destabilizes the protein and causes it to break down. It is an action as harmful as that of free radicals, promoting the formation of wrinkles and causing loss of elasticity and tone.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AGER	rs184003	GG-	A	- -	
AGER	rs1800624	TT-	T	- -	
AGER	rs1800625	CT-	G	+ -	
AGER	rs2070600	GG-	T	- -	
GLO1	rs4746	AA-	A,G	- -	

Metabolic disorders

Acute Intermittent Porphyria

NORMAL

It is a rare metabolic disorder in the production of heme, a prosthetic group of hemoglobin oxygen binding.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
DPAGT1	rs643788	CT+	C	- -	

Lipodystrophy

NORMAL

Lipodystrophy is an abnormal distribution of body fat. It occurs in patients who tend to develop insulin resistance, diabetes and have high levels of triglycerides. Some forms are hereditary and include Berardinelli-Seip syndrome. It is also common in HIV-positive patients.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
LMNA	rs28928902	CC+		- -	
LMNA	rs57520892	GG+		- -	
LMNA	rs60890628	CC+		- -	
PPARG	rs3856806	CC+	T	- -	
PPARG	rs72551362	GG+		- -	
PPARG	rs121909244	CC+	A,T	- -	

Variegate Porphyria

UNDEFINED

Variegata porphyria is one of the eight diseases of the porphyria group. It is caused by a deficiency in an enzyme called Protoporphyrinogen oxidase.

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Microbiome

Staphylococcus aureus infections

HIGH

Staphylococcus aureus is the most dangerous of all the most common staphylococcal bacteria. These Gram-positive, sphere-shaped (coconut) bacteria often cause skin infections, but can cause pneumonia, heart valve infections, and bone infections. The A allele of the SNP rs4321864 is associated with greater susceptibility to infection by Staphylococcus aureus.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
INTERGENIC	rs4321864	AC+	A	+ -	

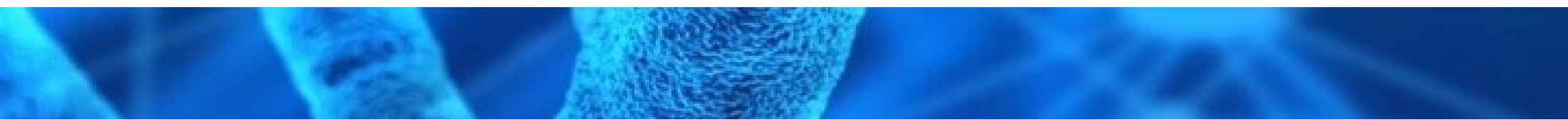
Need for Nutrients

Apigenin

HIGH

In recent years, a series of researches and studies have been carried out to prove the benefits of apigenin, a flavonoid found in parsley, thyme and chamomile, for example, with strong neurological effects and excellent antioxidant and anticancer properties. One of these studies, carried out by researchers at the federal universities of Rio de Janeiro and Bahia, showed that the substance has positive effects on memory and learning, in addition to protecting and strengthening brain connections, called synapses. According to experts, the more intense the synapses formed by our neurons, the easier it is to memorize information and recall them later. In addition to the benefits already mentioned, apigenin was also effective against ultraviolet radiation and visible light (all the light we see with the naked eye, including that from artificial lamps, computers and screens on tablets and smartphones, for example). According to studies, the substance minimizes the impacts of damage caused by these radiations, protecting the skin's DNA and regulating the photoaging process. Results in red means beneficial.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
GRIA3	rs687577	CC+	C	+ +	
GRIA3	rs3848874	GG+	A,T	- -	



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Sample

Uridine Monophosphate

MEDIUM-HIGH

It stimulates the growth of axons, stabilizes cell membranes in nerve cells and increases acetylcholine synthesis. It is used to prevent neurological disorders including Alzheimer's.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BDNF	rs6265	GG-	T	- -	
CLOCK	rs1801260	AA+	C,G,T	- -	
CLSTN2	rs6439886	AA+	G	+ +	
CLSTN2	rs17348572	TT+	C	+ +	
DBH	rs1611115	CT+	A,C,G	- -	
DRD4	rs1800955	CT+	C,G	+ -	
DTNBP1	rs2619522	GT-	C	- -	
GRK3	rs3730315	AA+	G	- -	
HES1	rs4686673	CC+	T	- -	
HTR1B	rs6296	CC+	G	- -	
HTR2A	rs6314	CC-	A	- -	
NT5C3A	rs104894025	AA-	A	- -	
NTF3	rs6332	GG+	A,T	- -	
SLC6A2	rs3785143	CC+	T	- -	
SLC6A3	rs27072	CT+	A,T	+ -	
SNAP25	rs3746544	AA-	T	- -	
TPH2	rs1843809	TT+	T	- -	
WWC1	rs17070145	CT+	T	+ -	

Turmeric (Curcumin)

MEDIUM

Benefit from regular turmeric intake, by stimulating the endogenous antioxidant system, and stimulating DNA repair.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BIN1	rs744373	CT-	G	+ -	
SOD3	rs1799895	CC+	G	- -	

Oxidation

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Sample

Oxidative stress

HIGH

Evaluation of a set of genes associated with the functioning of the reduction and oxidation (redox) system as a whole. Orange or red indicates poorer functioning, that is, greater risk of oxidative stress.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ADA	rs73598374	GG-	A,G,T	- -	
ALDH2	rs671	GG+	A	- -	
ATF1	rs11169571	CC+	C	+ +	
CAT	rs1001179	AA-	T	+ +	
CBS	rs234706	GG+	A	- -	
CBS	rs2851391	CT+	C	+ -	
EPHX1	rs1051740	TT+	C	- -	
EPHX1	rs2234922	AG+	G,T	+ -	
G6PD	rs1050829	AA-	C	- -	
G6PD	rs2230037	CC-	G	- -	
GCLC	rs17883901	CC-	A,T	- -	
GPX1	rs3448	CT+	T	+ -	
GPX1	rs1050450	CC-	A	- -	
GSR	rs1002149	TT+	T	- -	
GSR	rs2978663	TT+	T	+ +	
GSTM1	rs366631	CT-	G	+ -	
GSTP1	rs1695	GG+	G	+ +	
GSTP1	rs1138272	CT+	T	+ -	
HFE	rs1799945	GG+	G	+ +	
HFE	rs1800562	GG+	A	- -	
IL-6	rs1800795	CG+	G	+ -	
LCT	rs2322659	CT+	C	+ -	
LTA	rs909253	CT-	G,T	+ -	
NFE2L2	rs6721961	GG+	C,G	- -	
NQO1	rs1800566	CC-	A	- -	
SOD1	rs1041740	CT+	T	+ -	
SOD2	rs4880	CT-	G	+ -	
SOD3	rs1799895	CC+	G	- -	
SOD3	rs2855262	TT+	C	- -	
TLR4	rs4986790	AA+	G,T	- -	

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TNF	rs1800629	GG+	A	- -	
ZNF648	rs10911021	CT+	C	+ -	

Antioxidant Capacity

MEDIUM

Antioxidant compounds capable of inhibiting or retarding oxidation by inactivating free radicals thanks to the donation of hydrogen atoms or electrons, which transform the radicals into stable substances, are considered primary. Indication in orange or red indicates greater antioxidant capacity.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CAT	rs1001179	AA-	T	+ +	
CYP1B1	rs1056836	CC-	C	- -	
EPHX1	rs1051740	TT+	C	- -	
EPHX1	rs2234922	AG+	G,T	+ -	
G6PD	rs1050828	CC+	T	- -	
G6PD	rs1050829	AA-	C	- -	
GCLC	rs17883901	CC-	A,T	- -	
GPX1	rs1050450	CC-	A	- -	
GSTP1	rs1695	GG+	G	- -	
MTHFR	rs1476413	AG-	G,T	+ -	
MTHFR	rs1801131	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	
NAF1	rs7675998	GG+	G,T	- -	
NFE2L2	rs6706649	CC+	T	- -	
NFE2L2	rs6721961	GG+	C,G	- -	
NQO1	rs1800566	CC-	A	- -	
SOD2	rs4880	CT-	G	+ -	
SOD3	rs1799895	CC+	G	- -	
SOD3	rs2855262	TT+	C	- -	

Personal characteristics

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Sample

Straighter hair

NORMAL

Straighter hair is due to the parents' genetics.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
EDAR	rs3827760	TT-	G	- -	
TCHH	rs11803731	AT+	C,T	- -	

Eyebrow Thickness

NORMAL

There are different thickness and shapes of eyebrows. Thickness may be related to some genetic markers.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
EDAR	rs3827760	TT-	G	- -	

Trend to Blond Hair

NORMAL

Lighter hair tones.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
TYR	rs1393350	GG+	A	- -	

Trimethylaminouria (TMAU)

NORMAL

Also known as fish odor syndrome, it is a rare metabolic disease whose main symptoms are bad body odor, breath and breath, and bad taste in the mouth.

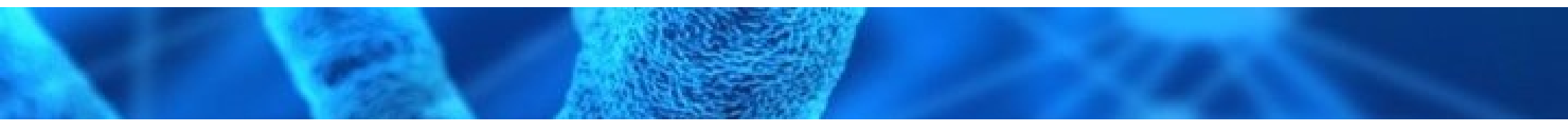
Gene	RSID	Genotype	Minor Allele	Alteration	Result
FMO3	rs909530	CC+	T	- -	
FMO3	rs1736557	AG+	A	+ -	
FMO3	rs2266780	AA+	G	- -	

Thicker hair

NORMAL

Thickness refers to the width of a single strand of hair, while density looks at how thin or thick strands are collectively, in a group. This means that someone can have fine hair that's also very dense as well. Alternatively, a person can also have thick hair that is not dense.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
EDAR	rs3827760	TT-	G	- -	



Double eyelids

UNDEFINED

Eyelids segmented by a fold; they look stretched from lashes to eyebrows.

Extreme Heat Resistance

UNDEFINED

Ability to withstand high temperatures without discomfort or risk of hyperthermia.

Reactions to Treatments

Glucocorticoid Resistance

NORMAL

The state of resistance or sensitivity to glucocorticoids, seen in patients with inflammatory autoimmune diseases. In orange or red, it indicates greater resistance to its action.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
NR3C1	rs6190	GG-	G,T	- -	

Glucocorticoid Therapy

NORMAL

Glucocorticoids are the primary anti-inflammatory therapy for asthma, but their effects are characterized by some interindividual variability that might have a genetic basis. Studies aimed to determine the relationship between pulmonary function change and the variant of the glucocorticoid-induced transcript 1 (GLCCI1) gene in patients with asthma receiving long-term ICS treatment, the association of GLCCI1 genotypes and the level of GLCCI1 expression and cytokines production. Individuals homozygous of SNP rs37973 for the wild-type allele who had a percent FEV1 change greater than 5% were more common than individuals homozygous for the rare allele. Patients with the A allele, the GLCCI1 expression was enhanced upon administration of low-dose dexamethasone; however, GG homozygotes required high-dose dexamethasone to achieve enhanced GLCCI1 expression. Furthermore, the levels of some cytokines were significantly reduced after glucocorticoid treatment in individuals with the AA and AG genotypes.The genetic variant rs37973 GG in GLCCI1 is associated with poorer clinical therapeutic response to inhaled glucocorticoids in asthma tested population. Results in red or orange indicate poorer clinical therapeutic response.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
GLCCI1	rs37973	AG+	A,C	+ -	

Skin

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Sample

Reduced Skin Barrier Permeability Function

HIGH

The skin barrier is a layer on the epidermis, made up of fats (mostly ceramides), which have the function of protecting and preserving the skin's hydration and health. The skin barrier has some functions, all related to the defense of the dermis and epidermis. It is responsible for regulating transdermal water loss, Determining the pH of the skin, creating favorable conditions for bacteria that live in harmony on its surface and that have a defense function against other unwanted microorganisms, Protecting the skin from external aggressions, such as the weather, pollution and sun exposure. For this layer to always remain healthy, it is important to avoid some harmful habits, such as using very hot water, very aggressive soaps or rubbing the skin with a towel. In addition, it is important to use a moisturizer that has the function of replacing ceramides and lipids, to restore damage to the barrier.

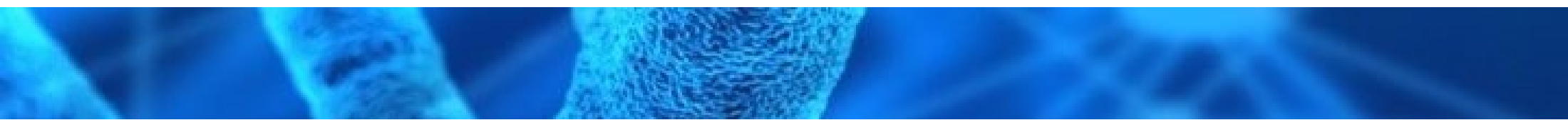
Gene	RSID	Genotype	Minor Allele	Alteration	Result
FCN1	rs11103631	GG+	G	+ +	

Hyaluronic acid

HIGH

Hyaluronic Acid is an active produced naturally by the body that has moisturizing and collagen stimulating properties. It is found in the body, but over time its production decreases, needing to be replaced in a treatment format. Result in Orange or Red indicate less production of Hyaluronic Acid.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FOXN4	rs12820283	AA+	A	+ +	
HYAL1	rs104893743	GG-	T	- -	



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Sample

Collagen Degradation

HIGH

In orange or red indicates the presence of polymorphisms associated with increased collagen degradation.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
B4GALT7	rs28937869	CC+	T	- -	
CBS	rs234706	GG+	A	- -	
CBS	rs2298758	GG+	A,C	- -	
COL11A1	rs2622848	TT+	C	- -	
COL11A1	rs3753841	AG+	A	+ -	
COL11A1	rs398122828	CC+	T	- -	
COL11A1	rs727503881	CC+	T	- -	
COL11A2	rs2076311	AA+	A	+ +	
COL11A2	rs2855429	CC+	C	+ +	
COL11A2	rs121912945	CC+	G,T	- -	
COL11A2	rs121912949	GG+	A,T	- -	
COL11A2	rs121912952	GG+	T	- -	
COL11A2	rs770888294	CC+	A,T	- -	
COL11A2	rs786205578	AA+	G,T	- -	
COL11A2	rs797044915	CC+	A	- -	
COL17A1	rs805698	TT+	G,T	+ +	
COL17A1	rs1320448	GG+	G	+ +	
COL1A1	rs1800012	GT-	A	- -	
COL1A1	rs2269336	CC-	A,C	+ +	
COL1A1	rs2586488	GG+	G	+ +	
COL1A1	rs67507747	CC+	A,G,T	- -	
COL1A1	rs72645328	CC+	G,T	- -	
COL1A1	rs72645347	GG+	A	- -	
COL1A1	rs72645353	CC+	A,T	- -	
COL1A1	rs72645357	GG-	T	- -	
COL1A1	rs72648320	CC+	T	- -	
COL1A1	rs139955975	CC+	T	- -	
COL1A1	rs144751329	CC+	A,T	- -	
COL1A1	rs193922140	CC+	G	- -	
COL1A1	rs193922143	II+		- -	

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Sample

COL1A1	rs193922144	GG+	A	- -	●
COL1A1	rs193922145	GG+	A	- -	●
COL1A1	rs193922147	CC+	A,G	- -	●
COL1A1	rs193922148	II+		- -	●
COL1A1	rs193922149	II+		- -	●
COL1A1	rs193922150	CC+	T	- -	●
COL1A1	rs193922151	II+		- -	●
COL1A1	rs193922152	TT+	C	- -	●
COL1A1	rs193922153	GG+	A	- -	●
COL1A1	rs193922155	TT+	C	- -	●
COL1A1	rs193922157	CC+	A,T	- -	●
COL1A1	rs193922158	TT+	C	- -	●
COL1A1	rs370865189	GG+	A,C,T	- -	●
COL1A2	rs42524	CC+	G	- -	●
COL1A2	rs441051	CT+	C	+ -	●
COL1A2	rs1801182	TT+	C	- -	●
COL1A2	rs1801182	TT+	C	- -	●
COL1A2	rs3736638	CC+	A	- -	●
COL1A2	rs72656355	AA+	G	- -	●
COL1A2	rs72658151	GG+	A	- -	●
COL1A2	rs72658154	GG+	A	- -	●
COL1A2	rs72658161	GG+	A	- -	●
COL1A2	rs72658176	GG+	A	- -	●
COL1A2	rs72659319	GG+	A,C	- -	●
COL1A2	rs139446305	GG+	A	- -	●
COL1A2	rs193922159	CC+	A,G	- -	●
COL1A2	rs193922162	GG+	A	- -	●
COL1A2	rs193922165	GG+	A	- -	●
COL1A2	rs193922168	GG+	C	- -	●
COL1A2	rs193922173	GG+	A	- -	●
COL1A2	rs768171831	CC+	T	- -	●
COL1A2	rs786205587	GG+	A	- -	●
COL1A2	rs794727470	GG+	C	- -	●
COL1A2	rs794727669	GG+	T	- -	●
COL1A2	rs797044949	GG+	T	- -	●

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Sample

COL27A1	rs946053	GG+	G	+ +	🔴
COL27A1	rs1249719	GG+	A	- -	🟢
COL27A1	rs7868992	AA+	A	+ +	🔴
COL27A1	rs140950220	GG+	C	- -	🟢
COL2A1	rs1793933	AC-	T	- -	🟢
COL2A1	rs121912866	GG+	A	- -	🟢
COL2A1	rs121912870	CC+	T	- -	🟢
COL2A1	rs121912873	II+		- -	🟢
COL2A1	rs121912874	GG+	A	- -	🟢
COL2A1	rs121912877	CC+	T	- -	🟢
COL2A1	rs121912880	CC+	A,T	- -	🟢
COL2A1	rs121912882	GG+	A	- -	🟢
COL2A1	rs121912884	GG+	A	- -	🟢
COL2A1	rs121912885	GG+	A,T	- -	🟢
COL2A1	rs121912886	GG+	A,T	- -	🟢
COL2A1	rs121912893	CC-	A,T	- -	🟢
COL2A1	rs398123628	II+		- -	🟢
COL2A1	rs727503882	CC+	G,T	- -	🟢
COL2A1	rs748459670	GG+	A,C	- -	🟢
COL2A1	rs786205477	CC+	A	- -	🟢
COL2A1	rs794727202	CC+	T	- -	🟢
COL2A1	rs794727225	II+		- -	🟢
COL2A1	rs794727261	GG+	T	- -	🟢
COL2A1	rs794727377	TT+	G	- -	🟢
COL2A1	rs794727438	CC+	A	- -	🟢
COL2A1	rs794727462	CC+	T	- -	🟢
COL2A1	rs794727472	CC+	A,T	- -	🟢
COL2A1	rs794727533	GG+	A,T	- -	🟢
COL2A1	rs794727546	CC+	G	- -	🟢
COL2A1	rs794727596	CC+	A	- -	🟢
COL2A1	rs794727684	CC+	T	- -	🟢
COL2A1	rs869312907	CC+	T	- -	🟢
COL3A1	rs1800255	AG+	A	+ -	🟠
COL3A1	rs1800255	AG+	A	+ -	🟡
COL3A1	rs111505097	GG+	A,T	- -	🟢

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Sample

COL3A1	rs111929073	GG+	A,C,T	- -	
COL3A1	rs112371422	CC+	G,T	- -	
COL3A1	rs112456072	GG+	A	- -	
COL3A1	rs113485686	GG+	A	- -	
COL3A1	rs113871730	GG+	A	- -	
COL3A1	rs121912913	GG+	A,T	- -	
COL3A1	rs121912914	GG+	A,T	- -	
COL3A1	rs121912915	GG+	T	- -	
COL3A1	rs121912916	GG+	A	- -	
COL3A1	rs121912917	GG+	A,T	- -	
COL3A1	rs121912918	GG+	A,T	- -	
COL3A1	rs121912919	GG+	A	- -	
COL3A1	rs121912920	GG+	A	- -	
COL3A1	rs121912921	GG+	A	- -	
COL3A1	rs121912922	GG+	A,T	- -	
COL3A1	rs121912923	GG+	A,C,T	- -	
COL3A1	rs121912924	GG+	A	- -	
COL3A1	rs121912925	GG+	A,T	- -	
COL3A1	rs121912926	GG+	A,C,T	- -	
COL3A1	rs121912927	GG+	A,T	- -	
COL3A1	rs121912928	GG+	A	- -	
COL3A1	rs193922176	GG+	C	- -	
COL3A1	rs267599120	GG+	A,C	- -	
COL3A1	rs387906557	GG+	C	- -	
COL3A1	rs397509369	GG+	A	- -	
COL3A1	rs397509370	GG+	A,T	- -	
COL3A1	rs397509371	GG+	T	- -	
COL3A1	rs397509372	GG+	A,T	- -	
COL3A1	rs397509373	GG+	A	- -	
COL3A1	rs397509375	TT+	A,C	- -	
COL3A1	rs397509376	GG+	A,T	- -	
COL3A1	rs397509377	DD+	T	- -	
COL3A1	rs553203474	GG+	A	- -	
COL3A1	rs587779416	GG+	T	- -	
COL3A1	rs587779417	GG+	A	- -	

Name Sample

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Sample

COL3A1	rs587779418	GG+	A	- -	
COL3A1	rs587779419	GG+	A	- -	
COL3A1	rs587779420	GG+	A,C	- -	
COL3A1	rs587779421	GG+	A	- -	
COL3A1	rs587779422	GG+	A	- -	
COL3A1	rs587779423	TT+	A,C	- -	
COL3A1	rs587779424	GG+	A	- -	
COL3A1	rs587779426	TT+	A,C	- -	
COL3A1	rs587779427	GG+	T	- -	
COL3A1	rs587779428	GG+	T	- -	
COL3A1	rs587779429	TT+	C	- -	
COL3A1	rs587779431	GG+	A,T	- -	
COL3A1	rs587779432	GG+	A	- -	
COL3A1	rs587779433	GG+	A,C	- -	
COL3A1	rs587779434	GG+	A	- -	
COL3A1	rs587779435	GG+	A,C	- -	
COL3A1	rs587779436	GG+	C	- -	
COL3A1	rs587779437	GG+	A	- -	
COL3A1	rs587779438	GG+	A	- -	
COL3A1	rs587779439	GG+	A	- -	
COL3A1	rs587779440	GG+	A,T	- -	
COL3A1	rs587779441	GG+	A,C	- -	
COL3A1	rs587779442	GG+	C	- -	
COL3A1	rs587779443	GG+	A,T	- -	
COL3A1	rs587779444	GG+	A,C,T	- -	
COL3A1	rs587779445	GG+	T	- -	
COL3A1	rs587779446	GG+	A	- -	
COL3A1	rs587779447	GG+	A	- -	
COL3A1	rs587779448	GG+	A	- -	
COL3A1	rs587779449	GG+	A,C	- -	
COL3A1	rs587779450	GG+	A,T	- -	
COL3A1	rs587779451	II+		- -	
COL3A1	rs587779452	GG+	T	- -	
COL3A1	rs587779453	AA+	C	- -	
COL3A1	rs587779454	GG+	A,T	- -	

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Sample

COL3A1	rs587779455	II+		- -	
COL3A1	rs587779456	GG+	A	- -	
COL3A1	rs587779457	GG+	A,T	- -	
COL3A1	rs587779458	GG+	A,T	- -	
COL3A1	rs587779459	GG+	A,C	- -	
COL3A1	rs587779460	GG+	A	- -	
COL3A1	rs1057518075	CC+	T	- -	
COL3A1	rs1057518372	GG+	A	- -	
COL3A1	rs1057521106	CC+	A,T	- -	
COL3A1	rs1057521930	GG+	A,T	- -	
COL3A1	rs1057523593	AA+	C	- -	
COL3A1	rs1060500193	GG+	A	- -	
COL3A1	rs1060500199	II+		- -	
COL3A1	rs1085307896	GG+	A	- -	
COL3A1	rs1085307964	GG+	T	- -	
COL4A3	rs7606754	AG+	G	+ -	
COL4A3	rs10178458	CC+	A,C	+ +	
COL4A3	rs11677877	AA+	G	- -	
COL4A3	rs34505188	GG+	A	- -	
COL4A3	rs121912824	CC+	T	- -	
COL4A3	rs121912825	CC+	G,T	- -	
COL4A3	rs121912827	GG+	A,T	- -	
COL4A3	rs201697532	CC+	T	- -	
COL4A3	rs759739044	GG+	A,T	- -	
COL4A4	rs2229813	CT+	G,T	+ -	
COL4A4	rs2272205	TT+	C	- -	
COL4A4	rs786205548	TT+	A	- -	
COL4A4	rs786205640	II+		- -	
COL4A5	rs104886096	GG+	A	- -	
COL4A5	rs104886142	GG+	A	- -	
COL4A6	rs769211787	AA+	C	- -	
COL5A1	rs12722	CC+	T	- -	
COL5A1	rs7044529	TT+	T	+ +	
COL5A1	rs7874142	AG+	A	+ -	
COL5A1	rs61735045	GG+	A	- -	

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Sample

COL5A1	rs80338764	GG+	C	- -	
COL5A1	rs113452150	GG+	A	- -	
COL5A1	rs183495554	TT+	A,C	- -	
COL5A1	rs374020067	CC+	T	- -	
COL5A1	rs377138881	GG+	A	- -	
COL5A1	rs387906606	CC+	T	- -	
COL5A1	rs557361751	CC+	T	- -	
COL5A1	rs564375308	CC+	T	- -	
COL5A1	rs764446683	CC+	A,G,T	- -	
COL5A1	rs765079080	TT+	G	- -	
COL5A1	rs777625241	CC+	T	- -	
COL5A1	rs794727114	GG+	C	- -	
COL5A1	rs794727760	II+		- -	
COL5A1	rs863223444	TT+	A	- -	
COL5A1	rs863223445	GG+	A	- -	
COL5A1	rs863223448	GG+	C	- -	
COL5A1	rs863223452	GG+	A	- -	
COL5A1	rs863223453	GG+	A,C	- -	
COL5A1	rs863223454	CC+	T	- -	
COL5A1	rs863223458	GG+	A	- -	
COL5A1	rs863223466	GG+	A	- -	
COL5A1	rs863223469	DD+	C	- -	
COL5A1	rs863223470	DD+	G	- -	
COL5A1	rs863223473	DD+	C	- -	
COL5A1	rs863223474	II+		- -	
COL5A1	rs863223475	DD+	T	- -	
COL5A1	rs863223478	CC+	T	- -	
COL5A1	rs863223483	TT+	G	- -	
COL5A2	rs121912930	CC+	G	- -	
COL5A2	rs747946828	CC+	A,T	- -	
COL5A2	rs762080305	GG+	A,C	- -	
COL5A2	rs770598613	CC+	G	- -	
COL5A2	rs773726323	CC+	T	- -	
COL5A2	rs779153546	CC+	T	- -	
COL5A2	rs780495441	CC+	T	- -	

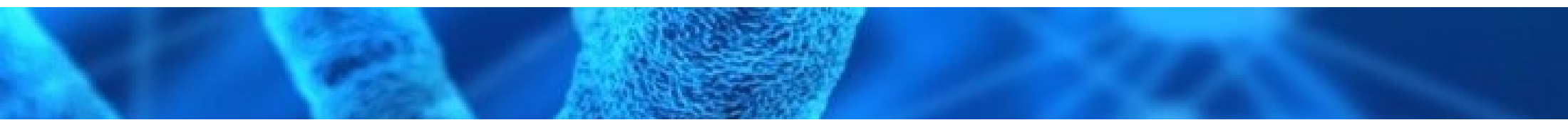
Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL5A2	rs863223491	CC+	T	- -	
COL5A2	rs863223501	CC+	T	- -	
MMP1	rs1799750	---	C	- -	
MMP3	rs679620	AG-	C	+ -	
MMP3	rs3025058	DI+	G	+ -	



Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

Bulldog effect

HIGH

Over the years, the skin loses water and collagen, resulting in sagging. On the face, the cheek tends to be the first part affected, with the skin sagging, which gives its name to the so-called bulldog effect.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
B4GALT7	rs28937869	CC+	T	- -	
CBS	rs234706	GG+	A	- -	
CBS	rs2298758	GG+	A,C	- -	
COL11A1	rs2622848	TT+	C	- -	
COL11A1	rs3753841	AG+	A	+ -	
COL11A1	rs398122828	CC+	T	- -	
COL11A1	rs727503881	CC+	T	- -	
COL11A2	rs2076311	AA+	A	+ +	
COL11A2	rs2855429	CC+	C	+ +	
COL11A2	rs121912945	CC+	G,T	- -	
COL11A2	rs121912949	GG+	A,T	- -	
COL11A2	rs121912952	GG+	T	- -	
COL11A2	rs770888294	CC+	A,T	- -	
COL11A2	rs786205578	AA+	G,T	- -	
COL11A2	rs797044915	CC+	A	- -	
COL17A1	rs805698	TT+	G,T	+ +	
COL17A1	rs1320448	GG+	G	+ +	
COL1A1	rs1800012	GT-	A	- -	
COL1A1	rs2269336	CC-	A,C	+ +	
COL1A1	rs2586488	GG+	G	+ +	
COL1A1	rs67507747	CC+	A,G,T	- -	
COL1A1	rs72645328	CC+	G,T	- -	
COL1A1	rs72645347	GG+	A	- -	
COL1A1	rs72645353	CC+	A,T	- -	
COL1A1	rs72645357	GG-	T	- -	
COL1A1	rs72648320	CC+	T	- -	
COL1A1	rs139955975	CC+	T	- -	
COL1A1	rs144751329	CC+	A,T	- -	
COL1A1	rs193922140	CC+	G	- -	
COL1A1	rs193922143	II+		- -	

Name Sample

Age

Gender F

Report date 12/09/2025

Prescriber

Health insurance

Sample

COL1A1	rs193922144	GG+	A	- -	●
COL1A1	rs193922145	GG+	A	- -	●
COL1A1	rs193922147	CC+	A,G	- -	●
COL1A1	rs193922148	II+		- -	●
COL1A1	rs193922149	II+		- -	●
COL1A1	rs193922150	CC+	T	- -	●
COL1A1	rs193922151	II+		- -	●
COL1A1	rs193922152	TT+	C	- -	●
COL1A1	rs193922153	GG+	A	- -	●
COL1A1	rs193922155	TT+	C	- -	●
COL1A1	rs193922157	CC+	A,T	- -	●
COL1A1	rs193922158	TT+	C	- -	●
COL1A1	rs370865189	GG+	A,C,T	- -	●
COL1A2	rs42524	CC+	G	- -	●
COL1A2	rs441051	CT+	C	+ -	●
COL1A2	rs1801182	TT+	C	- -	●
COL1A2	rs1801182	TT+	C	- -	●
COL1A2	rs3736638	CC+	A	- -	●
COL1A2	rs72656355	AA+	G	- -	●
COL1A2	rs72658151	GG+	A	- -	●
COL1A2	rs72658154	GG+	A	- -	●
COL1A2	rs72658161	GG+	A	- -	●
COL1A2	rs72658176	GG+	A	- -	●
COL1A2	rs72659319	GG+	A,C	- -	●
COL1A2	rs139446305	GG+	A	- -	●
COL1A2	rs193922159	CC+	A,G	- -	●
COL1A2	rs193922162	GG+	A	- -	●
COL1A2	rs193922165	GG+	A	- -	●
COL1A2	rs193922168	GG+	C	- -	●
COL1A2	rs193922173	GG+	A	- -	●
COL1A2	rs768171831	CC+	T	- -	●
COL1A2	rs786205587	GG+	A	- -	●
COL1A2	rs794727470	GG+	C	- -	●
COL1A2	rs794727669	GG+	T	- -	●
COL1A2	rs797044949	GG+	T	- -	●

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL27A1	rs946053	GG+	G	+ +	🔴
COL27A1	rs1249719	GG+	A	- -	🟢
COL27A1	rs7868992	AA+	A	+ +	🔴
COL27A1	rs140950220	GG+	C	- -	🟢
COL2A1	rs1793933	AC-	T	- -	🟢
COL2A1	rs121912866	GG+	A	- -	🟢
COL2A1	rs121912870	CC+	T	- -	🟢
COL2A1	rs121912873	II+		- -	🟢
COL2A1	rs121912874	GG+	A	- -	🟢
COL2A1	rs121912877	CC+	T	- -	🟢
COL2A1	rs121912880	CC+	A,T	- -	🟢
COL2A1	rs121912882	GG+	A	- -	🟢
COL2A1	rs121912884	GG+	A	- -	🟢
COL2A1	rs121912885	GG+	A,T	- -	🟢
COL2A1	rs121912886	GG+	A,T	- -	🟢
COL2A1	rs121912893	CC-	A,T	- -	🟢
COL2A1	rs398123628	II+		- -	🟢
COL2A1	rs727503882	CC+	G,T	- -	🟢
COL2A1	rs748459670	GG+	A,C	- -	🟢
COL2A1	rs786205477	CC+	A	- -	🟢
COL2A1	rs794727202	CC+	T	- -	🟢
COL2A1	rs794727225	II+		- -	🟢
COL2A1	rs794727261	GG+	T	- -	🟢
COL2A1	rs794727377	TT+	G	- -	🟢
COL2A1	rs794727438	CC+	A	- -	🟢
COL2A1	rs794727462	CC+	T	- -	🟢
COL2A1	rs794727472	CC+	A,T	- -	🟢
COL2A1	rs794727533	GG+	A,T	- -	🟢
COL2A1	rs794727546	CC+	G	- -	🟢
COL2A1	rs794727596	CC+	A	- -	🟢
COL2A1	rs794727684	CC+	T	- -	🟢
COL2A1	rs869312907	CC+	T	- -	🟢
COL3A1	rs1800255	AG+	A	+ -	🟠
COL3A1	rs1800255	AG+	A	+ -	🟡
COL3A1	rs111505097	GG+	A,T	- -	🟢

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL3A1	rs111929073	GG+	A,C,T	- -	
COL3A1	rs112371422	CC+	G,T	- -	
COL3A1	rs112456072	GG+	A	- -	
COL3A1	rs113485686	GG+	A	- -	
COL3A1	rs113871730	GG+	A	- -	
COL3A1	rs121912913	GG+	A,T	- -	
COL3A1	rs121912914	GG+	A,T	- -	
COL3A1	rs121912915	GG+	T	- -	
COL3A1	rs121912916	GG+	A	- -	
COL3A1	rs121912917	GG+	A,T	- -	
COL3A1	rs121912918	GG+	A,T	- -	
COL3A1	rs121912919	GG+	A	- -	
COL3A1	rs121912920	GG+	A	- -	
COL3A1	rs121912921	GG+	A	- -	
COL3A1	rs121912922	GG+	A,T	- -	
COL3A1	rs121912923	GG+	A,C,T	- -	
COL3A1	rs121912924	GG+	A	- -	
COL3A1	rs121912925	GG+	A,T	- -	
COL3A1	rs121912926	GG+	A,C,T	- -	
COL3A1	rs121912927	GG+	A,T	- -	
COL3A1	rs121912928	GG+	A	- -	
COL3A1	rs193922176	GG+	C	- -	
COL3A1	rs267599120	GG+	A,C	- -	
COL3A1	rs387906557	GG+	C	- -	
COL3A1	rs397509369	GG+	A	- -	
COL3A1	rs397509370	GG+	A,T	- -	
COL3A1	rs397509371	GG+	T	- -	
COL3A1	rs397509372	GG+	A,T	- -	
COL3A1	rs397509373	GG+	A	- -	
COL3A1	rs397509375	TT+	A,C	- -	
COL3A1	rs397509376	GG+	A,T	- -	
COL3A1	rs397509377	DD+	T	- -	
COL3A1	rs553203474	GG+	A	- -	
COL3A1	rs587779416	GG+	T	- -	
COL3A1	rs587779417	GG+	A	- -	

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL3A1	rs587779418	GG+	A	- -	●
COL3A1	rs587779419	GG+	A	- -	●
COL3A1	rs587779420	GG+	A,C	- -	●
COL3A1	rs587779421	GG+	A	- -	●
COL3A1	rs587779422	GG+	A	- -	●
COL3A1	rs587779423	TT+	A,C	- -	●
COL3A1	rs587779424	GG+	A	- -	●
COL3A1	rs587779426	TT+	A,C	- -	●
COL3A1	rs587779427	GG+	T	- -	●
COL3A1	rs587779428	GG+	T	- -	●
COL3A1	rs587779429	TT+	C	- -	●
COL3A1	rs587779431	GG+	A,T	- -	●
COL3A1	rs587779432	GG+	A	- -	●
COL3A1	rs587779433	GG+	A,C	- -	●
COL3A1	rs587779434	GG+	A	- -	●
COL3A1	rs587779435	GG+	A,C	- -	●
COL3A1	rs587779436	GG+	C	- -	●
COL3A1	rs587779437	GG+	A	- -	●
COL3A1	rs587779438	GG+	A	- -	●
COL3A1	rs587779439	GG+	A	- -	●
COL3A1	rs587779440	GG+	A,T	- -	●
COL3A1	rs587779441	GG+	A,C	- -	●
COL3A1	rs587779442	GG+	C	- -	●
COL3A1	rs587779443	GG+	A,T	- -	●
COL3A1	rs587779444	GG+	A,C,T	- -	●
COL3A1	rs587779445	GG+	T	- -	●
COL3A1	rs587779446	GG+	A	- -	●
COL3A1	rs587779447	GG+	A	- -	●
COL3A1	rs587779448	GG+	A	- -	●
COL3A1	rs587779449	GG+	A,C	- -	●
COL3A1	rs587779450	GG+	A,T	- -	●
COL3A1	rs587779451	II+		- -	●
COL3A1	rs587779452	GG+	T	- -	●
COL3A1	rs587779453	AA+	C	- -	●
COL3A1	rs587779454	GG+	A,T	- -	●

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL3A1	rs587779455	II+		- -	
COL3A1	rs587779456	GG+	A	- -	
COL3A1	rs587779457	GG+	A,T	- -	
COL3A1	rs587779458	GG+	A,T	- -	
COL3A1	rs587779459	GG+	A,C	- -	
COL3A1	rs587779460	GG+	A	- -	
COL3A1	rs1057518075	CC+	T	- -	
COL3A1	rs1057518372	GG+	A	- -	
COL3A1	rs1057521106	CC+	A,T	- -	
COL3A1	rs1057521930	GG+	A,T	- -	
COL3A1	rs1057523593	AA+	C	- -	
COL3A1	rs1060500193	GG+	A	- -	
COL3A1	rs1060500199	II+		- -	
COL3A1	rs1085307896	GG+	A	- -	
COL3A1	rs1085307964	GG+	T	- -	
COL4A3	rs7606754	AG+	G	+ -	
COL4A3	rs10178458	CC+	A,C	+ +	
COL4A3	rs11677877	AA+	G	- -	
COL4A3	rs34505188	GG+	A	- -	
COL4A3	rs121912824	CC+	T	- -	
COL4A3	rs121912825	CC+	G,T	- -	
COL4A3	rs121912827	GG+	A,T	- -	
COL4A3	rs201697532	CC+	T	- -	
COL4A3	rs759739044	GG+	A,T	- -	
COL4A4	rs2229813	CT+	G,T	+ -	
COL4A4	rs2272205	TT+	C	- -	
COL4A4	rs786205548	TT+	A	- -	
COL4A4	rs786205640	II+		- -	
COL4A5	rs104886096	GG+	A	- -	
COL4A5	rs104886142	GG+	A	- -	
COL4A6	rs769211787	AA+	C	- -	
COL5A1	rs12722	CC+	T	- -	
COL5A1	rs7044529	TT+	T	+ +	
COL5A1	rs7874142	AG+	A	+ -	
COL5A1	rs61735045	GG+	A	- -	

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL5A1	rs80338764	GG+	C	- -	
COL5A1	rs113452150	GG+	A	- -	
COL5A1	rs183495554	TT+	A,C	- -	
COL5A1	rs374020067	CC+	T	- -	
COL5A1	rs377138881	GG+	A	- -	
COL5A1	rs387906606	CC+	T	- -	
COL5A1	rs557361751	CC+	T	- -	
COL5A1	rs564375308	CC+	T	- -	
COL5A1	rs764446683	CC+	A,G,T	- -	
COL5A1	rs765079080	TT+	G	- -	
COL5A1	rs777625241	CC+	T	- -	
COL5A1	rs794727114	GG+	C	- -	
COL5A1	rs794727760	II+		- -	
COL5A1	rs863223444	TT+	A	- -	
COL5A1	rs863223445	GG+	A	- -	
COL5A1	rs863223448	GG+	C	- -	
COL5A1	rs863223452	GG+	A	- -	
COL5A1	rs863223453	GG+	A,C	- -	
COL5A1	rs863223454	CC+	T	- -	
COL5A1	rs863223458	GG+	A	- -	
COL5A1	rs863223466	GG+	A	- -	
COL5A1	rs863223469	DD+	C	- -	
COL5A1	rs863223470	DD+	G	- -	
COL5A1	rs863223473	DD+	C	- -	
COL5A1	rs863223474	II+		- -	
COL5A1	rs863223475	DD+	T	- -	
COL5A1	rs863223478	CC+	T	- -	
COL5A1	rs863223483	TT+	G	- -	
COL5A2	rs121912930	CC+	G	- -	
COL5A2	rs747946828	CC+	A,T	- -	
COL5A2	rs762080305	GG+	A,C	- -	
COL5A2	rs770598613	CC+	G	- -	
COL5A2	rs773726323	CC+	T	- -	
COL5A2	rs779153546	CC+	T	- -	
COL5A2	rs780495441	CC+	T	- -	

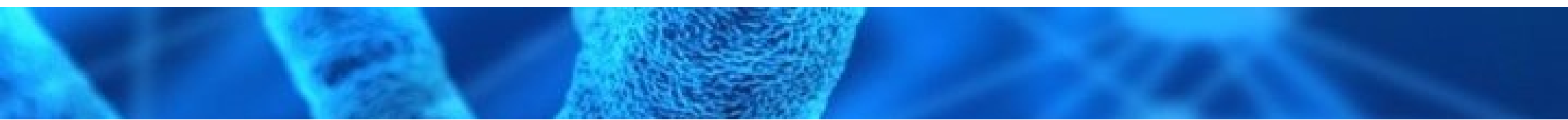
Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL5A2	rs863223491	CC+	T	- -	●
COL5A2	rs863223501	CC+	T	- -	●
MMP1	rs1799750	---	C	- -	●
MMP3	rs3025058	DI+	G	+ -	●



Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

Sagging Neck

HIGH

The sagging skin under the chin, popularly known as "double chin", is one of the signs of aging feared by women and men.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ADAMTS12	rs469568	TT-	C	- -	
ADAMTS12	rs469568	TT-	C	- -	
ADAMTS2	rs901254	CC+	T	- -	
ADAMTS2	rs901254	CC+	T	- -	
ADAMTS2	rs10039254	AA+	G	- -	
ADAMTS2	rs10039254	AA+	G	- -	
ADAMTS2	rs137853146	GG+	A	- -	
ADAMTS2	rs137853146	GG+	A	- -	
ADAMTS2	rs137853147	CC+	T	- -	
ADAMTS2	rs137853147	CC+	T	- -	
B4GALT7	rs28937869	CC+	T	- -	
CBS	rs234706	GG+	A	- -	
CBS	rs2298758	GG+	A,C	- -	
COL11A1	rs2622848	TT+	C	- -	
COL11A1	rs3753841	AG+	A	+ -	
COL11A1	rs398122828	CC+	T	- -	
COL11A1	rs727503881	CC+	T	- -	
COL11A2	rs2076311	AA+	A	+ +	
COL11A2	rs2855429	CC+	C	+ +	
COL11A2	rs121912945	CC+	G,T	- -	
COL11A2	rs121912949	GG+	A,T	- -	
COL11A2	rs121912952	GG+	T	- -	
COL11A2	rs770888294	CC+	A,T	- -	
COL11A2	rs786205578	AA+	G,T	- -	
COL11A2	rs797044915	CC+	A	- -	
COL17A1	rs805698	TT+	G,T	+ +	
COL17A1	rs1320448	GG+	G	+ +	
COL1A1	rs1800012	GT-	A	- -	
COL1A1	rs2269336	CC-	A,C	+ +	
COL1A1	rs2586488	GG+	G	+ +	

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

COL1A1	rs67507747	CC+	A,G,T	- -	●
COL1A1	rs72645328	CC+	G,T	- -	●
COL1A1	rs72645347	GG+	A	- -	●
COL1A1	rs72645353	CC+	A,T	- -	●
COL1A1	rs72645357	GG-	T	- -	●
COL1A1	rs72648320	CC+	T	- -	●
COL1A1	rs139955975	CC+	T	- -	●
COL1A1	rs144751329	CC+	A,T	- -	●
COL1A1	rs193922140	CC+	G	- -	●
COL1A1	rs193922143	II+		- -	●
COL1A1	rs193922144	GG+	A	- -	●
COL1A1	rs193922145	GG+	A	- -	●
COL1A1	rs193922147	CC+	A,G	- -	●
COL1A1	rs193922148	II+		- -	●
COL1A1	rs193922149	II+		- -	●
COL1A1	rs193922150	CC+	T	- -	●
COL1A1	rs193922151	II+		- -	●
COL1A1	rs193922152	TT+	C	- -	●
COL1A1	rs193922153	GG+	A	- -	●
COL1A1	rs193922155	TT+	C	- -	●
COL1A1	rs193922157	CC+	A,T	- -	●
COL1A1	rs193922158	TT+	C	- -	●
COL1A1	rs370865189	GG+	A,C,T	- -	●
COL1A2	rs42524	CC+	G	- -	●
COL1A2	rs441051	CT+	C	+ -	●
COL1A2	rs1801182	TT+	C	- -	●
COL1A2	rs3736638	CC+	A	- -	●
COL1A2	rs72656355	AA+	G	- -	●
COL1A2	rs72658151	GG+	A	- -	●
COL1A2	rs72658154	GG+	A	- -	●
COL1A2	rs72658161	GG+	A	- -	●
COL1A2	rs72658176	GG+	A	- -	●
COL1A2	rs72659319	GG+	A,C	- -	●
COL1A2	rs139446305	GG+	A	- -	●
COL1A2	rs193922159	CC+	A,G	- -	●

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COL1A2	rs193922162	GG+	A	- -	
COL1A2	rs193922165	GG+	A	- -	
COL1A2	rs193922168	GG+	C	- -	
COL1A2	rs193922173	GG+	A	- -	
COL1A2	rs768171831	CC+	T	- -	
COL1A2	rs786205587	GG+	A	- -	
COL1A2	rs794727470	GG+	C	- -	
COL1A2	rs794727669	GG+	T	- -	
COL1A2	rs797044949	GG+	T	- -	
COL27A1	rs946053	GG+	G	+ +	
COL27A1	rs1249719	GG+	A	- -	
COL27A1	rs7868992	AA+	A	+ +	
COL27A1	rs140950220	GG+	C	- -	
COL2A1	rs1793933	AC-	T	- -	
COL2A1	rs121912866	GG+	A	- -	
COL2A1	rs121912870	CC+	T	- -	
COL2A1	rs121912873	II+		- -	
COL2A1	rs121912874	GG+	A	- -	
COL2A1	rs121912877	CC+	T	- -	
COL2A1	rs121912880	CC+	A,T	- -	
COL2A1	rs121912882	GG+	A	- -	
COL2A1	rs121912884	GG+	A	- -	
COL2A1	rs121912885	GG+	A,T	- -	
COL2A1	rs121912886	GG+	A,T	- -	
COL2A1	rs121912893	CC-	A,T	- -	
COL2A1	rs398123628	II+		- -	
COL2A1	rs727503882	CC+	G,T	- -	
COL2A1	rs748459670	GG+	A,C	- -	
COL2A1	rs786205477	CC+	A	- -	
COL2A1	rs794727202	CC+	T	- -	
COL2A1	rs794727225	II+		- -	
COL2A1	rs794727261	GG+	T	- -	
COL2A1	rs794727377	TT+	G	- -	
COL2A1	rs794727438	CC+	A	- -	
COL2A1	rs794727462	CC+	T	- -	

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COL2A1	rs794727472	CC+	A,T	- -	●
COL2A1	rs794727533	GG+	A,T	- -	●
COL2A1	rs794727546	CC+	G	- -	●
COL2A1	rs794727596	CC+	A	- -	●
COL2A1	rs794727684	CC+	T	- -	●
COL2A1	rs869312907	CC+	T	- -	●
COL3A1	rs1800255	AG+	A	+ -	●
COL3A1	rs111505097	GG+	A,T	- -	●
COL3A1	rs111929073	GG+	A,C,T	- -	●
COL3A1	rs112371422	CC+	G,T	- -	●
COL3A1	rs112456072	GG+	A	- -	●
COL3A1	rs113485686	GG+	A	- -	●
COL3A1	rs113871730	GG+	A	- -	●
COL3A1	rs121912913	GG+	A,T	- -	●
COL3A1	rs121912914	GG+	A,T	- -	●
COL3A1	rs121912915	GG+	T	- -	●
COL3A1	rs121912916	GG+	A	- -	●
COL3A1	rs121912917	GG+	A,T	- -	●
COL3A1	rs121912918	GG+	A,T	- -	●
COL3A1	rs121912919	GG+	A	- -	●
COL3A1	rs121912920	GG+	A	- -	●
COL3A1	rs121912921	GG+	A	- -	●
COL3A1	rs121912922	GG+	A,T	- -	●
COL3A1	rs121912923	GG+	A,C,T	- -	●
COL3A1	rs121912924	GG+	A	- -	●
COL3A1	rs121912925	GG+	A,T	- -	●
COL3A1	rs121912926	GG+	A,C,T	- -	●
COL3A1	rs121912927	GG+	A,T	- -	●
COL3A1	rs121912928	GG+	A	- -	●
COL3A1	rs193922176	GG+	C	- -	●
COL3A1	rs267599120	GG+	A,C	- -	●
COL3A1	rs387906557	GG+	C	- -	●
COL3A1	rs397509369	GG+	A	- -	●
COL3A1	rs397509370	GG+	A,T	- -	●
COL3A1	rs397509371	GG+	T	- -	●

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COL3A1	rs397509372	GG+	A,T	- -	
COL3A1	rs397509373	GG+	A	- -	
COL3A1	rs397509375	TT+	A,C	- -	
COL3A1	rs397509376	GG+	A,T	- -	
COL3A1	rs397509377	DD+	T	- -	
COL3A1	rs553203474	GG+	A	- -	
COL3A1	rs587779416	GG+	T	- -	
COL3A1	rs587779417	GG+	A	- -	
COL3A1	rs587779418	GG+	A	- -	
COL3A1	rs587779419	GG+	A	- -	
COL3A1	rs587779420	GG+	A,C	- -	
COL3A1	rs587779421	GG+	A	- -	
COL3A1	rs587779422	GG+	A	- -	
COL3A1	rs587779423	TT+	A,C	- -	
COL3A1	rs587779424	GG+	A	- -	
COL3A1	rs587779426	TT+	A,C	- -	
COL3A1	rs587779427	GG+	T	- -	
COL3A1	rs587779428	GG+	T	- -	
COL3A1	rs587779429	TT+	C	- -	
COL3A1	rs587779431	GG+	A,T	- -	
COL3A1	rs587779432	GG+	A	- -	
COL3A1	rs587779433	GG+	A,C	- -	
COL3A1	rs587779434	GG+	A	- -	
COL3A1	rs587779435	GG+	A,C	- -	
COL3A1	rs587779436	GG+	C	- -	
COL3A1	rs587779437	GG+	A	- -	
COL3A1	rs587779438	GG+	A	- -	
COL3A1	rs587779439	GG+	A	- -	
COL3A1	rs587779440	GG+	A,T	- -	
COL3A1	rs587779441	GG+	A,C	- -	
COL3A1	rs587779442	GG+	C	- -	
COL3A1	rs587779443	GG+	A,T	- -	
COL3A1	rs587779444	GG+	A,C,T	- -	
COL3A1	rs587779445	GG+	T	- -	
COL3A1	rs587779446	GG+	A	- -	

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COL3A1	rs587779447	GG+	A	- -	
COL3A1	rs587779448	GG+	A	- -	
COL3A1	rs587779449	GG+	A,C	- -	
COL3A1	rs587779450	GG+	A,T	- -	
COL3A1	rs587779451	II+		- -	
COL3A1	rs587779452	GG+	T	- -	
COL3A1	rs587779453	AA+	C	- -	
COL3A1	rs587779454	GG+	A,T	- -	
COL3A1	rs587779455	II+		- -	
COL3A1	rs587779456	GG+	A	- -	
COL3A1	rs587779457	GG+	A,T	- -	
COL3A1	rs587779458	GG+	A,T	- -	
COL3A1	rs587779459	GG+	A,C	- -	
COL3A1	rs587779460	GG+	A	- -	
COL3A1	rs1057518075	CC+	T	- -	
COL3A1	rs1057518372	GG+	A	- -	
COL3A1	rs1057521106	CC+	A,T	- -	
COL3A1	rs1057521930	GG+	A,T	- -	
COL3A1	rs1057523593	AA+	C	- -	
COL3A1	rs1060500193	GG+	A	- -	
COL3A1	rs1060500199	II+		- -	
COL3A1	rs1085307896	GG+	A	- -	
COL3A1	rs1085307964	GG+	T	- -	
COL4A3	rs7606754	AG+	G	+ -	
COL4A3	rs10178458	CC+	A,C	+ +	
COL4A3	rs11677877	AA+	G	- -	
COL4A3	rs34505188	GG+	A	- -	
COL4A3	rs121912824	CC+	T	- -	
COL4A3	rs121912825	CC+	G,T	- -	
COL4A3	rs121912827	GG+	A,T	- -	
COL4A3	rs201697532	CC+	T	- -	
COL4A3	rs759739044	GG+	A,T	- -	
COL4A4	rs2229813	CT+	G,T	+ -	
COL4A4	rs2272205	TT+	C	- -	
COL4A4	rs786205548	TT+	A	- -	

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COL4A4	rs786205640	II+		- -	●
COL4A5	rs104886096	GG+	A	- -	●
COL4A5	rs104886142	GG+	A	- -	●
COL4A6	rs769211787	AA+	C	- -	●
COL5A1	rs12722	CC+	T	- -	●
COL5A1	rs7044529	TT+	T	+ +	●
COL5A1	rs7874142	AG+	A	+ -	●
COL5A1	rs61735045	GG+	A	- -	●
COL5A1	rs80338764	GG+	C	- -	●
COL5A1	rs113452150	GG+	A	- -	●
COL5A1	rs183495554	TT+	A,C	- -	●
COL5A1	rs374020067	CC+	T	- -	●
COL5A1	rs377138881	GG+	A	- -	●
COL5A1	rs387906606	CC+	T	- -	●
COL5A1	rs557361751	CC+	T	- -	●
COL5A1	rs564375308	CC+	T	- -	●
COL5A1	rs764446683	CC+	A,G,T	- -	●
COL5A1	rs765079080	TT+	G	- -	●
COL5A1	rs777625241	CC+	T	- -	●
COL5A1	rs794727114	GG+	C	- -	●
COL5A1	rs794727760	II+		- -	●
COL5A1	rs863223444	TT+	A	- -	●
COL5A1	rs863223445	GG+	A	- -	●
COL5A1	rs863223448	GG+	C	- -	●
COL5A1	rs863223452	GG+	A	- -	●
COL5A1	rs863223453	GG+	A,C	- -	●
COL5A1	rs863223454	CC+	T	- -	●
COL5A1	rs863223458	GG+	A	- -	●
COL5A1	rs863223466	GG+	A	- -	●
COL5A1	rs863223469	DD+	C	- -	●
COL5A1	rs863223470	DD+	G	- -	●
COL5A1	rs863223473	DD+	C	- -	●
COL5A1	rs863223474	II+		- -	●
COL5A1	rs863223475	DD+	T	- -	●
COL5A1	rs863223478	CC+	T	- -	●

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COL5A1	rs863223483	TT+	G	- -	
COL5A2	rs121912930	CC+	G	- -	
COL5A2	rs747946828	CC+	A,T	- -	
COL5A2	rs762080305	GG+	A,C	- -	
COL5A2	rs770598613	CC+	G	- -	
COL5A2	rs773726323	CC+	T	- -	
COL5A2	rs779153546	CC+	T	- -	
COL5A2	rs780495441	CC+	T	- -	
COL5A2	rs863223491	CC+	T	- -	
COL5A2	rs863223501	CC+	T	- -	
DLGAP1	rs11876749	TT+	C,G	- -	
MMP1	rs1799750	---	C	- -	
MMP3	rs3025058	DI+	G	+ -	

Generalized Vitiligo

HIGH

Localized: Appears on hands or face only; Segmental: the spot is a thick line that follows; Generalized: various spots appear in different areas of the body; but close together and Scattered: the different points are far apart.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
C1QTNF6	rs229527	GG-	A	- -	
CTLA4	rs231775	AG+	G	+ -	
GZMB	rs8192917	AA-	C	- -	
HCG9	rs3823355	CC+	T	- -	
IL2RA	rs706779	AG-	A	+ -	
IRF4	rs12203592	CC+	T	- -	
LPP	rs1464510	GT-	A,T	+ -	
PTPN22	rs2476601	GG+	G	- -	
RERE	rs4908760	GG+	G	+ +	
RNASET2	rs2236313	CC+	C	- -	
SMOC2	rs13208776	AG+	G	+ -	
TNFRSF11A	rs8083511	AA+	C	- -	
TYR	rs1393350	GG+	A	- -	
UBASH3A	rs11203203	AG+	A	+ -	
WASF5P	rs9468925	AG+	G	+ -	
XBP1	rs2269577	CG+	G	+ -	

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Sample

Periocular line

HIGH

The skin in the periocular region (around the eyes) is very delicate and requires special care. The bags and dark circles that form near the eyes are uncomfortable signs, in addition to bone resorption, which increases the appearance of aging and deep tiredness.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

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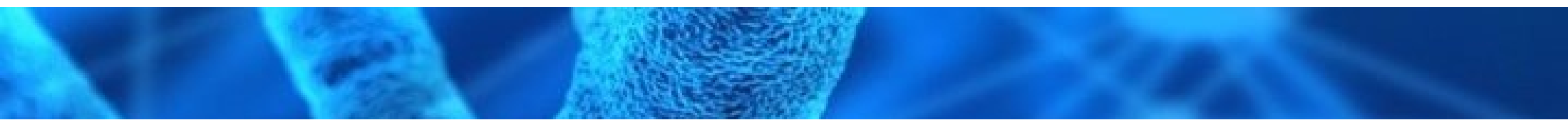
Sample

Acne

MEDIUM-HIGH

Acne is an inflammatory process that manifests itself in the skin when hair follicles are clogged by fat and dead cells.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BCO1	rs7501331	CC+	T	- -	
BCO1	rs12934922	AA+	G,T	- -	
C11ORF49	rs747650	AG-	T	+ -	
CTLA4	rs3087243	AG+	G	+ -	
CYP17A1	rs743572	AA+	G	- -	
CYP19A1	rs700518	AA-	C	- -	
HSD11B1	rs846910	AG+	A	+ -	
IL-1A	rs17561	GT-	A	+ -	
IL-1A	rs1800587	CT-	A,C	+ -	
IL-6	rs1800796	GG+	C	- -	
INTERGENIC	rs38055	CT-	G	+ -	
INTERGENIC	rs1159268	AG+	A	+ -	
INTERGENIC	rs7531806	AA+	A	- -	
LTA	rs1799724	CT+	T	+ -	
MCM6	rs4988235	AG+	A	- -	
OVOL1	rs479844	CT-	G	+ -	
RETN	rs3745367	AA+	A	+ +	
TNF	rs1800629	GG+	A	- -	
TYK2	rs33980500	CC+	T	- -	



Name Sample

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Sample

Melasma

MEDIUM-HIGH

Melasma is a condition characterized by the appearance of dark spots on the skin, most commonly on the face, but it can also be extrafacial, affecting the arms, neck and cervix. It affects women more often and can also be seen in men.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BNC2	rs10733310	GT+	A,G	+ -	
IRF4	rs12203592	CC+	T	- -	
MC1R	rs2228479	GG+	A,C	- -	
OCA2	rs1800414	AA-	C	- -	
OCA2	rs7495174	AA+	A	+ +	
SLC45A2	rs26722	CT+	T	+ -	

Dark circles

MEDIUM-HIGH

The most common type of dark circles is the one that darkens the lower part of the eye, which, in some cases, leaves spots much darker than the person's skin. There is another type called flabby eyelid dark circles.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
COL1A2	rs1801182	TT+	C	- -	
COL3A1	rs1800255	AG+	A	+ -	
DLGAP1	rs11876749	TT+	C,G	- -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

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Sample

Chicken feet

MEDIUM-HIGH

They are very unsightly wrinkles that form around the eyes. If, when smiling, you have already noticed small expression lines in this region, know that these are signs of skin aging, and they do not have a certain age to appear, but they happen when the skin starts to sag. Genetics is one of the great triggers of crow's feet, but we cannot forget external factors such as excessive sun, pollution, smoking and even myopia.

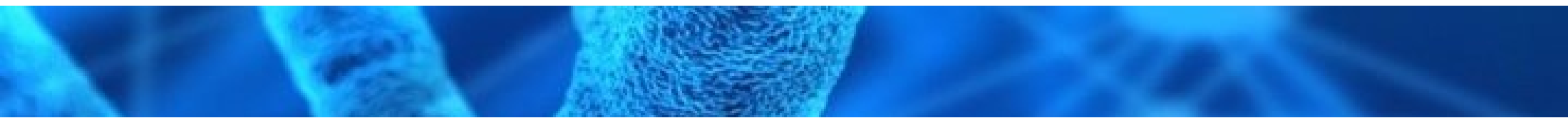
Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
MMP1	rs1799750	---	C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Bags

MEDIUM-HIGH

The bags in the eye region are mainly caused by fluid retention, high-fat diet, excessive alcohol and sleep deprivation. The eyes become swollen and give a heavy look to the face. These are characteristics that are normally also hereditary.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	



Name Sample

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Sample

Gravitational Wrinkles

MEDIUM-HIGH

Gravitational wrinkles are caused solely and exclusively by the impact of gravity on the skin, causing it to lose elasticity and look droopy. They are folds that form on the sides of the face, where the skin is thinner, forming unsightly creases and lines.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Forehead wrinkles

MEDIUM-HIGH

Forehead wrinkles can begin to appear around the age of 30, especially in people who, throughout their lives, have been exposed to the sun without protection, have lived in places with pollution or have neglected their diet.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
RBMS3	rs11711327	GG+	A	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

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Sample

Chin Wrinkles

MEDIUM-HIGH

Some people when they talk, or laugh, or when they do facial mimes, have “orange peel” or “golf ball” chin wrinkles.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
SHC4	rs28392847	CC-	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Alopecia Areata

MEDIUM-HIGH

Alopecia areata is an inflammatory disease that causes hair loss. Several factors are involved in its development, such as genetics and autoimmune participation. The strands begin to fall resulting more often in circular flaws with no hair or hair.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HLA-DQA2	rs9275572	GG+	G	+ +	
IL2RA	rs3130320	CT+	T	+ -	
INTERGENIC	rs1024161	CT+	T	+ -	
INTERGENIC	rs9479482	CT+	T	+ -	
PTPN22	rs2476601	GG+	G	- -	

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Sample

Nasogenian Line

MEDIUM-HIGH

Nasogeniana line or the "Chinese mustache" is that fold or wrinkle that appears on the face, from the side of the nasal wing to the corners of the mouth, on both sides, as if separating the cheeks from the lips, which may extend to the sides of the chin. It appears with age due to the flaccidity of the skin in this region, or due to a genetic factor, still in youth. They can be smooth or sharp lines.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Perioral Line

MEDIUM-HIGH

The perioral lines are expression marks that appear around the lips and are commonly known as a “bar code”. Repeated muscle contractions, along with skin aging, form these wrinkles, which, over time, can be seen even when the lips are relaxed. It is very common to find these lines, especially in smokers.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Name Sample

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Sample

Puppet Lines

MEDIUM-HIGH

They are usually an extension of the Chinese mustache. They occur due to sagging skin in the malar region (cheeks) and also due to the absorption of perioral fat compartments.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Loss of Facial Volume

MEDIUM-HIGH

The biggest changes in the face related to aging are due to the loss of fat areas and the reduction of the bony part of the face. The transformation of these two structures, which support and maintain the three-dimensionality of the face, makes the face acquire new shapes. The main area of fat lost with age is below the eye region (cheek). With this, the eyes end up getting deeper and dark circles and bags, more prominent.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

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Sample

Fall from the Corner of the Mouth

MEDIUM-HIGH

The drop in the corners of the mouth, leading to a sad mouth, is formed by a set of factors related to aging: loss of skin elasticity and firmness, loss of subcutaneous fat, reabsorption of the bones of the face. if we do nothing to soften them, the tendency is for them to become more and more fallen. The mouth angle depressor muscle, responsible for the drop in the corner of the mouth, is also related.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Nasal wrinkles

MEDIUM-HIGH

They are expression lines that many people make when smiling or talking, involuntarily. They are also known as "bunny lines".

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
STXBP5L	rs322458	GG-	T	+ +	

Name	Sample				
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Sample

Cellulitis

MEDIUM-HIGH

Cellulite is a complex cosmetic problem common to many post-adolescent women, characterized by changes that give the skin an orange-peel appearance. The rs11549465 polymorphism results in a C to T substitution. The T allele acts as a protective factor against the formation of subcutaneous septal fibrosis, thus reducing the risk of cellulite formation.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HIF1A	rs11549465	CC+	T	+ +	

Inflammatory Skin Response

MEDIUM-HIGH

We show a clear effect of the IL1 genotype on protein expression in the stratum corneum. This altered expression may be responsible for inter-individual differences in the skin's inflammatory response. The outermost layer of skin, called the stratum corneum or keratin layer, contains many layers of flat dead cells and acts as a barrier to protect the underlying tissues from injury and infection. By delaying evaporation, the oils present in this layer of skin help to keep moisture in the deeper layers, keeping the skin texture smooth and supple. The stratum corneum is just a part of the epidermis, which is a thin layer of skin that covers most of the body. In some places, such as the palms of the hands and the soles of the feet, the epidermis is naturally thick and the stratum corneum provides extra protection against impacts and abrasions. The epidermis can also be thick and hard in excessively dry areas. Disorders of the superficial layers of the skin involve the stratum corneum and the deeper layers of the epidermis and range from those that cause temporary discomfort to those that cause chronic disabilities. Orange or Red results indicate a greater tendency for skin inflammation.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IL-1A	rs1800587	CT-	A,C	+ -	
IL-1B	rs1143627	CT-	A	+ -	

Response to tanning

MEDIUM-HIGH

It refers to how each skin reacts to tanning. The graph on the side indicates greater response to tanning with orange and red colors.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ASIP	rs1015362	GG-	T	+ +	
ASIP	rs4911414	GG+	G	- -	
GRM5	rs10831496	AG+	A	+ -	
INTERGENIC	rs966321	AC-	T	- -	
PIGU	rs910873	GG+	A,C	- -	
SLC24A5	rs1426654	AG+	G,T	+ -	

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Sample

Collagen Synthesis

MEDIUM

Polymorphisms are associated with increased collagen synthesis. Result on the right indicates greater synthesis.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL1A1	rs72645357	GG-	T	- -	
COL3A1	rs1800255	AG+	A	+ -	

Skin Hydration

MEDIUM

The skin, in addition to being our body's first defense organ against the adversities of the external environment, has important roles, whose complexities and health contribute to the maintenance of the body's homeostasis. Such properties, however, are only performed with excellence if the integumentary tissue is in normal and full working and care conditions. Dehydrated skin is due to a lower than normal degree of imbibition, a condition often mediated by climate, such as wind and cold, in addition to low atmospheric humidity. The skin is an interface where exchanges with the environment are constant. The stratum corneum, a product of epidermal differentiation, forms an efficient barrier between the external environment and internal tissues. The amount of transepidermal water loss is a good measure of this barrier function. When this loss is very high, we observe dehydrated skin, xerosis, ichthyosis, itching, xeroderma and eczema. Orange or red result indicates worse skin hydration.

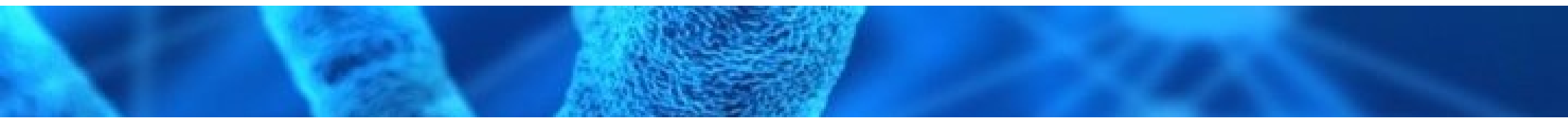
Gene	RSID	Genotype	Minor Allele	Alteration	Result
AHR	rs2066853	AG+	A	+ -	
AHR	rs10249788	CT+	T	+ -	
FGFR1	rs6996321	GG+	A	- -	
FLG	rs61816761	GG+	A,T	- -	

Varicose veins

MEDIUM

Twisted and dilated veins, which appear mainly on the legs and feet.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MTHFR	rs1801131	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	



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Sample

Stretch marks

MEDIUM

Stretch marks are scars that form when there is destruction of elastic and collagen fibers in the skin, usually caused by a stretching of the skin.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL1A2	rs42524	CC+	G	- -	
ELN	rs7787362	TT+	C,T	+ +	
HMCN1	rs10798036	GG+	A,G,T	+ +	
IL-6	rs1800795	CG+	G	+ -	
MMP1	rs1799750	---	C	- -	
MMP3	rs3025058	DI+	G	+ -	
PRDM6	rs59170767	CC+	T	- -	
TMEM18	rs6548238	CC+	C	- -	

Wrinkles in the lap

MEDIUM

The cervix, the area between the breasts and the neck, also suffers from wrinkles, largely due to exposure to the sun. They are common in older people, as they are more flaccid.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL1A2	rs1801182	TT+	C	- -	
COL3A1	rs1800255	AG+	A	+ -	
DLGAP1	rs11876749	TT+	C,G	- -	
STXBP5L	rs322458	GG-	T	- -	

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Sample

Baldness (Androgenetic Alopecia)

MEDIUM

Male pattern baldness or hair loss in women. Hair follicles are more vulnerable to the negative effects of high levels of dihydrotestosterone (DHT); a diet high in soy (for men) and zinc can lower the risk of AGA.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AR	rs6152	GG+	A	- -	
C1ORF127	rs2003046	CC+	A,G	- -	
EDA2R	rs1385699	TT+	T	+ +	
HDAC9	rs2073963	GT+	G	+ -	
HR	rs7014851	TT+	C	- -	
IL-1B	rs1143634	CT-	A	- -	
INTERGENIC	rs925391	CC-	G,T	- -	
INTERGENIC	rs2180439	CC+	T	- -	
INTERGENIC	rs2223841	AA-	C	- -	
INTERGENIC	rs6945541	TT+	C	- -	
INTERGENIC	rs9287638	CC+	A	- -	
IRF4	rs12203592	CC+	T	- -	
LINC01432	rs1160312	GG+	G	- -	
SLC14A2	rs8085664	CC+	C	+ +	

Permanent Elastic Wrinkles

MEDIUM

Permanent elastic wrinkles are a consequence of genetic aging and can be aggravated by unprotected sun exposure throughout life. They appear due to the thinning of the skin and also the intense loss of collagen, usually appearing on the cheeks, upper lip and base of the neck.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL1A2	rs1801182	TT+	C	- -	
COL3A1	rs1800255	AG+	A	+ -	
DLGAP1	rs11876749	TT+	C,G	- -	
GLIS1	rs702491	CC+	T	- -	
RBMS3	rs11711327	GG+	A	- -	

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Sample

Antioxidant capacity of the skin

MEDIUM

Free radicals are unstable molecules present in the body that damage the skin due to their high production. This happens, mainly, when we are exposed to pollution, solar radiation without photoprotection, genetic predisposition, environmental and nutritional conditions, among other factors, which increase the levels of free radicals, generating an oxidative cascade that results in the oxidation of cell membrane proteins, decreased synthesis of collagen and elastin, interfering with the quality and shine of the skin, leading to premature aging. Antioxidants help prevent collagen destruction by free radicals and stimulate the production of new fibers, thus having a corrective action in addition to prevention. In this way, they avoid wrinkles, skin sagging and, consequently, the emergence of new signs of aging. Orange or red results indicate a lower antioxidant capacity of the skin, thus recommending the use of antioxidants as an ally in the prevention and maintenance of skin homeostasis.

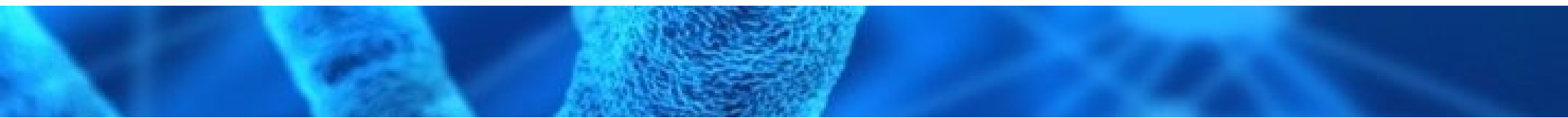
Gene	RSID	Genotype	Minor Allele	Alteration	Result
CAT	rs1001179	AA-	T	- -	
EPHX1	rs2234922	AG+	G,T	+ -	
GPX1	rs1050450	CC-	A	- -	
NFE2L2	rs6706649	CC+	T	- -	
NFE2L2	rs6721961	GG+	C,G	- -	
NFE2L2	rs35652124	CT+	C	+ -	
NQO1	rs1800566	CC-	A	- -	
SOD2	rs4880	CT-	G	+ -	

Ephelides (Freckles)

MEDIUM

Freckles or ephelides are spots caused by an increase in melanin (pigment that gives color to the skin) on the skin, which also has a familial tendency and appear mainly in fair-skinned people (phototype I and II) and redheads.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ASIP	rs1015362	GG-	T	+ +	
BNC2	rs2153271	GG-	T	- -	
IRF4	rs12203592	CC+	T	- -	
OCA2	rs4778138	GG+	G,T	- -	



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Lentigos (Sun Spots)

MEDIUM

Lentigo is a skin pigmentation similar to freckles, but not only dependent on the sun to appear. It is a brownish, circumscribed spot, with regular edges or not, composed of a localized proliferation of epidermal melanocytes.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ASIP	rs1015362	GG-	T	+ +	
ASIP	rs4911414	GG+	G	- -	
HERC2	rs12913832	AA+	G	- -	
INTERGENIC	rs12210050	CC+	T	- -	
IRF4	rs12203592	CC+	T	- -	
MC1R	rs1110400	TT+	C	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
NCOA6	rs4911442	AA+	A	- -	

Ehlers Danlos Syndrome (collagen)

MEDIUM

It is the name given to a group of 10 different inherited diseases, all of which involve a genetic defect in the synthesis and structure of collagen and connective tissue. EDS affects the skin, joint and blood vessels.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
B4GALT7	rs28937869	CC+	T	- -	
CBS	rs234706	GG+	A	- -	
CBS	rs2298758	GG+	A,C	- -	
COL1A2	rs1801182	TT+	C	- -	
COL3A1	rs1800255	AG+	A	+ -	
COL5A1	rs863223444	TT+	A	- -	
COL5A1	rs863223445	GG+	A	- -	

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Sample

Eyelid sagging

MEDIUM

The sagging eyelid deepens the eyes and casts shadows that make the eye region feel dark.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ADAMTS12	rs469568	TT-	C	- -	
ADAMTS2	rs901254	CC+	T	- -	
ADAMTS2	rs10039254	AA+	G	- -	
ADAMTS2	rs137853146	GG+	A	- -	
ADAMTS2	rs137853147	CC+	T	- -	
COL1A2	rs1801182	TT+	C	- -	
COL3A1	rs1800255	AG+	A	+ -	
DLGAP1	rs11876749	TT+	C,G	- -	
MACROH2A2	rs16927253	CT+	T	+ -	

Expression Marks (Glabella)

MEDIUM

The glabellar wrinkles are the furrows between the eyebrows, in the region called the glabella. They arise from the continuous contraction of the muscles in this region - corrective muscles - and over time, they can form deep fixed grooves, which do not disappear even with the muscles of the face relaxed.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CETP	rs5882	AG+	A	+ -	
FANCA	rs12931267	GG-	G	- -	
KL	rs9536314	TT+	A,G	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	

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Sample

Atypical Mycobacteriosis

MEDIUM

Skin diseases caused by species of the Mycobacterium genus are divided into cutaneous tuberculosis, whose etiological agents are Mycobacterium tuberculosis or Mycobacterium bovis, and atypical mycobacteriosis, caused by species other than the tuberculosis bacillus and Mycobacterium lepra

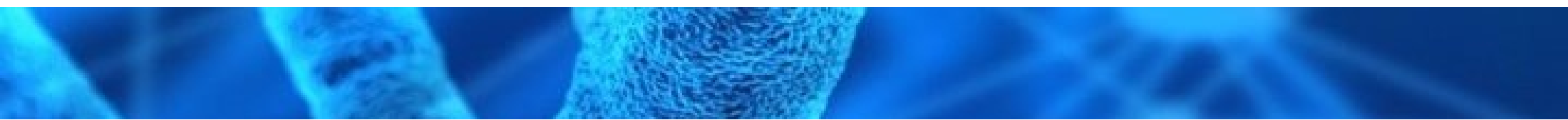
Gene	RSID	Genotype	Minor Allele	Alteration	Result
IFNGR1	rs3799488	TT+	C	- -	
IL-12B	rs3212227	CC-	G	+ +	
IL-12B	rs3213119	GG-	A	- -	
IL-12RB1	rs375947	AA+	G	- -	
IL-12RB1	rs436857	GG+	A	- -	

Hyperchromias

NORMAL

Hyperchromias are dark spots that appear on the skin and are produced by excess production of melanin (skin color).

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BNC2	rs10733310	GT+	A,G	+ -	



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Skin Aging

NORMAL

Skin changes are among the most visible signs of aging. Skin properties such as hydration, elasticity and antioxidant capacity play a key role in the skin aging process. Skin aging is a complex process influenced by hereditary and environmental factors. Recent twin studies have revealed that up to 60% of the variation in skin aging between individuals can be attributed to genetic factors, while the remaining 40% is due to non-genetic factors. Recent advances in genomics and bioinformatics approaches have led to the association of certain single nucleotide polymorphisms (SNPs) with skin properties. Our analysis assesses individuals based on a set of multiple polymorphisms associated with certain skin properties to provide personalized skin care and anti-aging therapies.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CAT	rs1001179	AA-	T	- -	
IL-6	rs1800795	CG+	G	+ -	
MMP1	rs1799750	---	C	- -	
MMP3	rs3025058	DI+	G	- -	
NFE2L2	rs6706649	CC+	T	- -	
NFE2L2	rs6721961	GG+	C,G	- -	
NFE2L2	rs35652124	CT+	C	+ -	
NQO1	rs1800566	CC-	A	- -	
SOD2	rs4880	CT-	G	+ -	

Dermatochalasis (Excess Skin on the Eyelids)

NORMAL

Dermatochalasis is an excess of skin that can occur on both the upper and lower eyelid. There may also be pockets of fat that increase the sensation of bulk in the eyelids. Therefore, the eyelids are heavier and make the face look tired. The extra weight accelerates the skin's loss of elasticity, causing sagging.

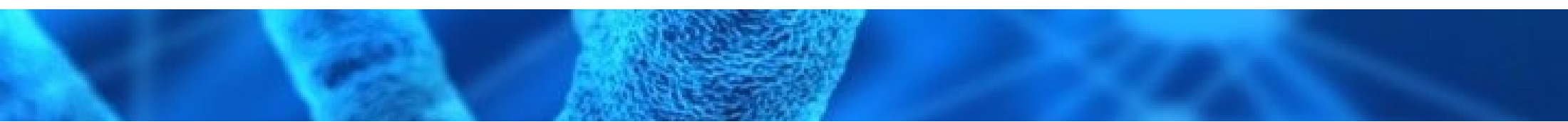
Gene	RSID	Genotype	Minor Allele	Alteration	Result
DLGAP1	rs11876749	TT+	C,G	- -	

Junctional bullous epidermolysis atresia of the pylorus

NORMAL

It is a basal subtype of simple bullous epidermolysis.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ITGA6	rs11895564	AA+	A	- -	



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Sample

Elastin Deficiency

NORMAL

With its main component being viscoelasticity, it completes collagen (which is a fiber that does not offer elasticity to the skin), forming a “support” network giving the epidermis the ability to return to its original shape after being stretched or deformed, giving elasticity, firmness, support and tone to the skin. It is also found in the walls of arteries, lungs and ligaments. The structure of elastin is composed of amino acid residues such as glycine, alanine, valine and proline (large amount), which are small, nonpolar amino acids, lysine and little hydroxyproline and no hydroxylysine.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ELN	rs2071307	GG+	A,C	- -	
ELN	rs3757587	CC+	T	- -	
ELN	rs7787362	TT+	C,T	- -	
ELN	rs41511151	GG+	A	- -	
ELN	rs137854452	CC+	T	- -	
ELN	rs137854453	CC+	G,T	- -	
ELN	rs200862792	GG+	A,T	- -	
ELN	rs201012726	CC+	T	- -	
ELN	rs397516433	CC+	G,T	- -	
ELN	rs527441189	GG+	A,T	- -	
ELN	rs727503022	DD+	C	- -	
ELN	rs727503023	II+		- -	
ELN	rs727503024	II+		- -	
ELN	rs727503026	II+		- -	
ELN	rs727503027	AA+	G	- -	
ELN	rs727503028	II+	G	- -	
ELN	rs727503029	GG+	A,T	- -	
ELN	rs727503030	GG+	A	- -	
ELN	rs727503031	II+		- -	
ELN	rs727503033	TT+	A,C	- -	
ELN	rs727503035	GG+	A	- -	
ELN	rs727503782	II+		- -	
ELN	rs727504581	II+		- -	
ELN	rs730880355	DD+	A	- -	
ELN	rs782755390	CC+	A,T	- -	
ELN	rs863223513	TT+	A	- -	
ELN	rs863223516	GG+	A,C	- -	
ELN	rs863223518	TT+	C	- -	

ELN	rs863223519	II+		- -	
ELN	rs863223522	II+		- -	
ELN	rs863223526	II+		- -	
ELN	rs863223528	DD+	C	- -	

KID Syndrome

NORMAL

KID syndrome is a rare congenital ectodermal dysplasia that affects the skin, corneal epithelium, and inner ear. Clinically, there are erythrokeratoderma plaques on the face and folds, usually present from birth, severe and bilateral sensorineural deafness, and corneal vascularization associated with progressively evolving keratitis that appears after skin and hearing changes in puberty. Given the condition of deafness, skin infections, the risk of blindness and malignant degeneration, early diagnosis of the syndrome is essential, as well as periodic clinical follow-up and genetic counseling.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
GJB2	rs28931594	GG-	T	- -	
GJB2	rs80338939	---		- -	
GJB2	rs104894408	GG-		- -	

Family candidiasis

NORMAL

Candidiasis is an infection caused by the fungus Candida albicans, which commonly lodges in the genital area, causing itching, secretion

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CARD9	rs10781499	GG+	A	- -	
CLEC7A	rs16910526	TT-	C,G	- -	
IL-17F	rs763780	TT+	C	- -	
IL-17F	rs2397084	TT+	C	- -	

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Sample

Dystrophic Bullosa Epidermolysis

NORMAL

It is a serious skin condition that causes blisters.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL7A1	rs121912839	GG-	T	- -	
COL7A1	rs121912845	CC-	A	- -	
COL7A1	rs121912847	CC-	A	- -	
COL7A1	rs121912849	CC-	A,C	- -	
COL7A1	rs121912852	CC-	A	- -	
COL7A1	rs121912854	CC-	A	- -	
COL7A1	rs121912855	CC-	A	- -	
COL7A1	rs143457874	CC-	A	- -	
LAMB3	rs80356680	CC-	A	- -	
LAMB3	rs80356681	CC-	A	- -	
LAMB3	rs80356682	CC-	A	- -	
LAMB3	rs786205451	GG-	T	- -	

Simple Bullous Epidermolysis

NORMAL

A disease of the hereditary epidermolysis bullosa (HEB) group, characterized by skin fragility, resulting in intra-epidermal blisters and erosions that occur spontaneously or after physical trauma.

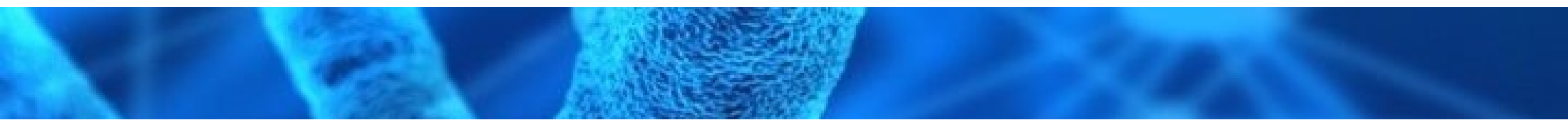
Gene	RSID	Genotype	Minor Allele	Alteration	Result
KRT5	rs11170164	GG-	T	- -	

Armpit odor

NORMAL

Research indicates that carriers of the G (C) allele of SNP rs17822931 are more susceptible to stronger underarm odors.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCC11	rs17822931	TT+	T	- -	



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Sample

Pseudofolliculitis barbae

NORMAL

Pseudofolliculitis barbae (razor bumps) is a common condition of the beard area and shaved areas, occurring in up to 60% African American men and other people with curly hair. The problem results when highly curved hairs grow back into the skin causing inflammation and a foreign body reaction.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
KRT75	rs2232387	GG-	A	- -	

Keloids

NORMAL

It occurs when there is an overgrowth of scar tissue at the site of a healed wound or surgery. Keloids are formed by raised lesions, usually red and can occur in any area of the skin.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BPESC1	rs940187	GG-	C	- -	
HYAL1	rs11130248	AA+	G	- -	
INTERGENIC	rs873549	AG-	T	+ -	
INTERGENIC	rs1511412	GG+	G,T	- -	
NEDD4	rs8032158	TT+	A,C	- -	

Hereditary Trichilemmal Cysts

NORMAL

Trichilemmal cysts are flesh-colored nodules or lumps that usually appear on your head. They can surface on your face, neck, arms and legs.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
PLCD1	rs75495843	GG+	A	- -	

Wrinkles

LOW

Rugas são sinais visíveis do envelhecimento da pele. Elas surgem quando a pele perde elasticidade, firmeza e hidratação. Com o tempo, os movimentos do rosto (como sorrir, franzir a testa ou mastigar) somados à perda de substâncias importantes fazem com que a pele quebre, formando linhas e sulcos.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IRF4	rs12203592	CC+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MMP1	rs1799750	---	C	- -	
MMP3	rs3025058	DI+	G	+ -	

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Leprosy

LOW

Chronic and curable infectious disease that mainly causes skin lesions and nerve damage.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CCR7	rs3136687	AA-	C	- -	
TNF	rs1800629	GG+	A	- -	

Skin elasticity

LOW

The property of skin that, when exposed to pressure, has the ability to return to its original state in a relatively acceptable period of time. This is made possible by two substances: elastin and collagen. That's why our skin is a viscoelastic medium. Elasticity parameters vary according to the area of the body and the person's age, because over the years the dermis produces less elastin and collagen.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IL-6	rs1800795	CG+	G	+ -	
MMP1	rs1799750	---	C	- -	
MMP3	rs3025058	DI+	G	+ -	

Ceramides

LOW

Ceramides are lipids (oils) naturally present in the skin, essential for its barrier and to retain hydration. Without ceramides, the skin can become dry, itchy and irritated. When the level of ceramides is low, the skin barrier is weakened, allowing the skin to lose its hydration. Applying ceramides directly to the skin helps to repair its barrier and retain moisture.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FCN1	rs11103631	GG+	G	- -	
IFNG	rs2069705	CT-	G	+ -	
IFNG	rs2430561	AT+	A	+ -	

Alcohol Erythema Reaction

LOW

Redness caused by drinking alcoholic beverages.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ALDH2	rs671	GG+	A	- -	

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Acne in Adolescence

UNDEFINED

The causes of acne have not yet been identified, but are believed to include genetic, environmental and individual skin factors. Oily skin is associated with acne, but all individuals with oily skin do not suffer from the condition.

Hereditary Chronic Mucocutaneous Candidiasis

UNDEFINED

Chronic mucocutaneous candidiasis, an inherited immunodeficiency disease, is a persistent or recurrent infection with Candida (a fungus) due to malfunctioning T cells (lymphocytes).

Erythema

UNDEFINED

Erythema is a dermatological lesion manifested by more or less intense skin redness that disappears when pressure is applied to it.

Jaundice

UNDEFINED

Syndrome of various diseases, characterized by the yellow coloration of tissues and organic secretions, resulting from the abnormal presence of bile pigments.

Poikiloderma

UNDEFINED

This disease is characterized by excessive skin pigmentation and apparent dilated vessels. Poikiloderma usually appears in patients over 40 years of age, who neglected sun protection during their lifetime. In addition to excessive exposure to ultraviolet rays, the disease may also be related to the skin's natural aging process, as well as genetic predisposition and/or hormonal factors.

Erythropoietic Protoporphyria

UNDEFINED

Erythropoietic protoporphyria (EPP) is due to an inherited deficiency in the activity of the enzyme ferrochelatase, and X-linked protoporphyria (XLPP) is due to an inherited increase in the activity of delta-aminolevulinic acid synthetase 2; both enzymes are in the heme biosynthesis pathway (Substrates and enzymes of the heme biosynthetic pathway and diseases associated with its deficiencies). Clinically, PPE and XLPP are almost identical. They typically manifest in childhood with itching and burning pain in the skin after brief exposure to sunlight. Gallstones are common later in life, and chronic liver disease occurs in about 10% of patients. Diagnosis is based on symptoms and increased levels of protoporphyrin in erythrocytes and plasma. Prevention is to avoid triggers (eg, sunlight, alcohol, and fasting) and perhaps the use of oral beta-carotene. Acute cutaneous symptoms can be relieved by cold baths, wet towels, analgesics and topical and/or oral corticosteroids. Patients with liver failure may need a liver transplant, but this type of transplant is not curative because the predominant source of overproduction of protoporphyria is the bone marrow.

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Tissue Remodeling (Healing)

UNDEFINED

Healing is the name given to the tissue repair process that replaces damaged tissue with new tissue. Repair involves the regeneration of specialized cells, formation of granulation tissue and tissue reconstruction. These events do not happen in isolation, but overlap and complete each other. A scar is the new tissue that forms during the healing process of a wound. Nature uses it as a means to close the body's injuries when perfect tissue regeneration is not possible.

Rosacea

UNDEFINED

A condition that causes redness and often small, red, pus-filled bumps on the face.

Skin Youthfulness

UNDEFINED

Genes that promote skin youthfulness, in the form of late aging, have been reported through research.

Special features

Rapid Healing

UNDEFINED

Ability to recover from injuries at an accelerated rate.

Extreme Cold Resistance

UNDEFINED

Ability to withstand cold temperatures without discomfort or risk of hypothermia.

Sunlight Resistance

UNDEFINED

Less damage from UV rays, lower risk of sunburn or skin cancer.

Regenerative Healing

UNDEFINED

Accelerated tissue repair, potentially including regeneration of certain tissues.

Vitamins

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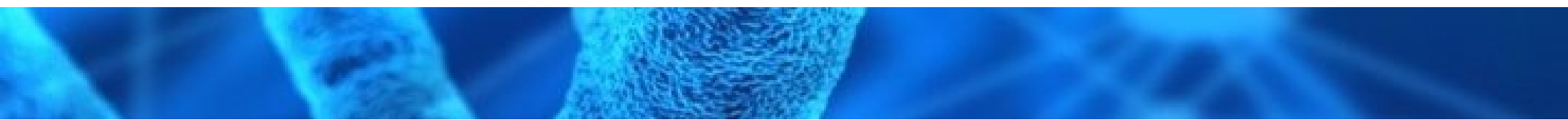
Vitamin B3 (Niacin)

HIGH

Niacin (nicotinic acid, vitamin B3) was considered a promising candidate to prevent cardiovascular disease because it is known to lower cholesterol in the blood, which is one of the main risk factors. Therefore, long-term therapy with niacin was assumed to reduce the risk of heart attack, and stroke. It's also important for maintaining the balance of the skin and nervous system. It is also relevant to the proper functioning of the digestive system.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BST1	rs4698412	AA+	A	+ +	
GAD1	rs2241165	AA-	T	- -	
NAMPT	rs61330082	CC-	A	- -	
SIRT1	rs12778366	TT+	C	+ +	
SIRT3	rs511744	CC+	T	+ +	
SIRT6	rs352493	TT+	C	+ +	



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Vitamin E

HIGH

Vitamin E is an important vitamin that helps regulate cholesterol, improve skin and hair health, and has an antioxidant action. Indication in orange or red indicates greater need.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
A2M	rs669	AA-	C	- -	
ABCA7	rs3764650	TT+	G	- -	
ABCA7	rs115550680	AA+	G	- -	
ACE	rs4343	AG+	A	+ -	
ADD1	rs4961	GT+	A,T	+ -	
ADRB2	rs1800888	CC+	T	- -	
AGAP2	rs12368653	GG+	A,T	- -	
APOA5	rs12272004	CC+	A	+ +	
APOC1	rs4420638	AA+	G	- -	
APOE	rs7412	TT+	T	+ +	
APOE4	rs429358	TT+	C	- -	
ARHGAP20	rs326946	TT-	C	- -	
ATP2B1	rs2681472	CT-	G	- -	
BAG3	rs2234962	CT+	C	- -	
BATF	rs2300603	TT+	C	- -	
BDNF	rs6265	GG-	T	- -	
BIN1	rs744373	CT-	G	+ -	
BRAP	rs3782886	AA-	C	- -	
BTD	rs104893686	TT+	G	- -	
BTD	rs104893687	CC+	T	- -	
C1ORF106	rs7522462	AG+	A	+ -	
CASC17	rs1859962	GG+	T	+ +	
CASC21	rs16902104	CC+	G,T	- -	
CASC8	rs1447295	CC+	C,T	- -	
CASC8	rs6983267	GG+	T	+ +	
CCL2	rs1024611	CT-	G	+ -	
CD2AP	rs9349407	GG+	C	- -	
CD2AP	rs10948363	AA+	G	- -	
CD58	rs12044852	CC+	A	- -	
CD86	rs1129055	GG+	A	- -	

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CD86	rs9282641	GG+	A	- -	🟢
CDH1	rs16260	AC+	A	+ -	🟡
CDH13	rs8055236	GG+	A,C,T	+ +	🟠
CDKN1B	rs34330	CT+	C	+ -	🟡
CDKN2B-AS1	rs1011970	GT+	T	- -	🟢
CDKN2B-AS1	rs1063192	TT-	A,T	- -	🔵
CDKN2B-AS1	rs10116277	TT+	T	- -	🔵
CDKN2B-AS1	rs10757272	TT+	T	+ +	🟠
CDKN2B-AS1	rs10757274	GG+	G	+ +	🟠
CETP	rs5882	AG+	A	+ -	🔵
CETP	rs2303790	AA+	G	- -	🟢
CFH	rs1061170	TT+	T	- -	🔵
CHAT	rs733722	CC+	A,G,T	- -	🟢
CHEK2	rs17879961	TT-	C,G	- -	🟢
CHST12	rs6952809	CC+	C	- -	🟢
CLSTN2	rs17411949	CC+	T	- -	🟢
CLU	rs11136000	CC+	C	- -	🟢
COMT	rs4680	AG+	A	+ -	🟢
CPS1	rs1047891	CC+	A	- -	🟢
CR1	rs3818361	CC-	G	- -	🟢
CR1	rs6656401	GG+	G,T	- -	🟢
CYP24A1	rs2296241	GG+	A	- -	🟢
CYP24A1	rs4809957	AG+	G	- -	🟢
CYP24A1	rs6068816	CT+	T	- -	🟢
CYP3A4	rs2740574	AA-	T	- -	🟢
CYP3A4	rs4646438	---		- -	🟢
CYP3A4	rs55785340	AA+	G	- -	🟢
CYP4F2	rs2108622	CC+	T	- -	🟢
DAB2IP	rs1571801	AC-	T	- -	🟢
DBC1	rs10984447	AA+	G	+ +	🟠
DKKL1	rs2303759	TT+	G	- -	🟢
DLEU1	rs2762051	CC+	T	- -	🟢
DMD	rs1800278	AA-	C	- -	🟢
DMD	rs1801187	GG-	T	- -	🟢
DMD	rs104894788	GG-	T	- -	🟢

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DNAJC5B	rs13279522	TT+	C	- -	🟢
DSG2	rs2230234	AA+	G,T	- -	🟢
ELAC2	rs4792311	GG+	A,C	- -	🔵
F12	rs1801020	CT-	G	+ -	🟡
F7	rs6046	CT-	A	- -	🟢
FGFR4	rs351855	CC-	A	- -	🟢
FLJ10986	rs1470407	CT+	C	- -	🟢
FMN2	rs17672135	TT+	C	+ +	🟠
FRMD4A	rs10906466	AG+	C,G	- -	🟢
GAB2	rs2373115	GG-	A	- -	🟢
GCKR	rs1260326	CT+	C	+ -	🟡
GOLM1	rs7019241	CT+	G,T	- -	🟢
GOLM1	rs10868366	GT+	T	- -	🟢
GPX1	rs1050450	CC-	A	- -	🟢
GSTP1	rs1695	GG+	G	+ +	🟠
HLA-DRA	rs3135391	CC-	A	- -	🟢
HLA-DRA	rs61731956	GG+	A	- -	🟢
HLA-DRB1	rs660895	GG+	G	+ +	🔴
HLA-DRB1	rs3135388	CC-	A	- -	🟢
HTRA1	rs11200638	AG+	A	+ -	🟠
IL-10	rs1800896	AG-	C	+ -	🟠
IL-2RA	rs2104286	AG-	C	- -	🟢
IL-2RA	rs12722489	AG-	T	- -	🟢
IL-4	rs2243250	CC+	T	- -	🟢
IL-6	rs1800795	CG+	G	- -	🟢
IL-7R	rs6897932	CC+	T	+ +	🟠
INTERGENIC	rs501120	AG-	C	+ -	🟡
INTERGENIC	rs1333049	CC+	C	+ +	🔴
INTERGENIC	rs2119704	CC+	A	- -	🟢
INTERGENIC	rs2383207	GG+	G	+ +	🟠
INTERGENIC	rs2836061	CT+	T	+ -	🟡
INTERGENIC	rs4728142	AG+	A	- -	🟢
INTERGENIC	rs5945572	AG+	G	+ -	🟡
INTERGENIC	rs7965399	TT+	A,C	- -	🟢
INTERGENIC	rs10260404	CT+	C	+ -	🟡

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INTERGENIC	rs10505483	AG-	T	+ -	🟠
INTERGENIC	rs10757278	GG+	G	+ +	🔴
INTERGENIC	rs10896449	AA+	G	- -	🟢
INTERGENIC	rs12473579	GG+	A	- -	🟢
INTERGENIC	rs13192841	GG+	A	- -	🟢
INTERGENIC	rs16901979	AC+	A	+ -	🔴
ITGA2	rs1126643	CC+	T	- -	🟢
ITGB3	rs5918	TT+	C	- -	🟢
JAZF1	rs10486567	AG+	A	+ -	🟡
KCNE1	rs1805127	AG-	A,C,G	+ -	🟡
KCNE2	rs2234916	AA+	G	- -	🟢
KCNE3	rs2270676	CT-	G	- -	🟢
KL	rs9536314	TT+	A,G	- -	🟢
KLF6	rs3750861	CC+	T	- -	🟢
LAG3	rs870849	CT+	C	- -	🟢
LDLR	rs688	CC+	T	- -	🟢
LRP6	rs2160525	AG+	G	- -	🟢
LRP8	rs5174	GG-	T	- -	🔵
LTA	rs1799724	CT+	T	+ -	🟠
MAOA	rs909525	GG-	T	+ +	🟠
MAOA	rs1137070	TT+	C	- -	🟢
MAOA	rs3027399	GG+	C	- -	🟢
MMP3	rs3025058	DI+	G	+ -	🟢
MPO	rs2333227	TT+	T	- -	🟢
MPV17L2	rs874628	TT-	G,T	- -	🟢
MRAS	rs2306374	TT+	C	- -	🟢
MS4A6A	rs610932	CC-	G	- -	🟢
MSMB	rs10993994	AA-	G	- -	🟢
MTHFD1	rs1076991	AG-	C,G	+ -	🟢
MTHFD1L	rs6922269	AG+	A	+ -	🟡
MTHFD1L	rs11754661	GG+	A,T	- -	🟢
MTHFD1L	rs17349743	TT+	C	- -	🟢
MYBPC3	rs11570112	CC-	A,C	- -	🟢
NAF1	rs7675998	GG+	G,T	- -	🟢
NKAIN3	rs7834588	CT+	T	+ -	🟢

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NOS3	rs1800779	AA+	G	- -	🟢
NPPA	rs5065	AA+	G	- -	🟢
PCAT19	rs11672691	AA+	T	- -	🟢
PCDH11X	rs5984894	AA+	G	- -	🟢
PCK1	rs8192708	AG+	G	+ -	🟡
PHACTR1	rs9349379	AA+	A	- -	🟢
PICALM	rs3851179	AG-	C	+ -	🔵
PICALM	rs10792832	AG+	G	+ -	🟡
PLAU	rs2227562	GG+	A	- -	🟢
PLAU	rs2227564	TT+	C	- -	🟢
PLD3	rs145999145	GG+	A	- -	🔵
PLPP3	rs17114036	AA+	G	- -	🟢
POLN	rs1923775	TT+	T	- -	🟢
PPP1R3B	rs3748140	GG-	T	- -	🟢
PPP1R3B	rs9987289	GG+	G	- -	🟢
PRRC2C	rs2421847	AA+	G	- -	🟢
PSEN1	rs63749824	CC+	G,T	- -	🟢
PSEN2	rs63750197	CC+	T	- -	🟢
PSMA6	rs1048990	CG+	G,T	- -	🟢
PSRC1	rs599839	AA+	A,C	- -	🟡
RNASEL	rs3738579	CT-	G	+ -	🔵
RPS6KB1	rs630923	CC+	A	- -	🟢
RYR2	rs34967813	AA+	G	- -	🟢
SCN5A	rs1805124	GG-	T	- -	🟢
SH2B3	rs3184504	CC+	A,C,G	- -	🟢
SLC30A7	rs11581062	AA+	G	- -	🟢
SMAD3	rs17228212	TT+	C	- -	🟢
SOD3	rs1799895	CC+	G	- -	🟢
SOD3	rs2855262	TT+	C	- -	🟢
TCF2	rs4430796	AG+	G	+ -	🟢
TCF7L2	rs12255372	GG+	T	- -	🟢
TLR4	rs4986790	AA+	G,T	- -	🟢
TLR4	rs4986791	CC+	T	- -	🟢
TMPO	rs17028450	CC+	T	- -	🟢
TMPRSS2	rs12329760	CT+	T	- -	🟢

PRECISION HEALTH PRECISYA

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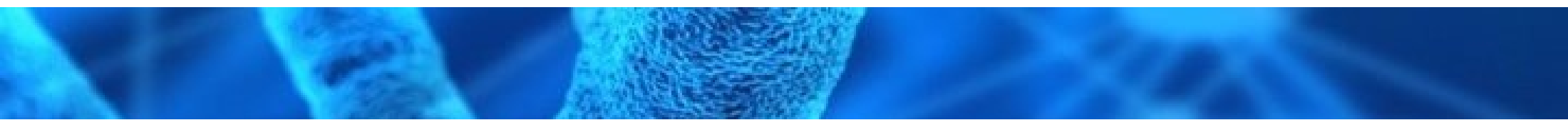
TNF	rs1800629	GG+	A	- -	●
TNFSF14	rs344560	GG-	C	- -	●
TNFSF14	rs2291667	CC-	A	- -	●
TNFSF4	rs1234313	AG+	G	+ -	●
TNFSF4	rs3861950	TT+	C	- -	●
TREM2	rs75932628	CC+	A,T	- -	●
TTN	rs2244492	GG-	T	- -	●
TTPA	rs121917850	GG-	T	- -	●
TTPA	rs121917851	CC-	A	- -	●
TXNRD2	rs5748469	AA+	A	- -	●
XRCC1	rs25489	GG-	G,T	- -	●
ZNF767P	rs354033	CC-	A	- -	●
ZNF827	rs13149290	TT+	T	- -	●
ZPR1	rs964184	CC+	C	- -	●



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Vitamin B7 (Biotin)

MEDIUM-HIGH

Vitamin B7 (Biotin) is one of the B-complex vitamins, responsible for breaking down fats and carbohydrates from food and transforming them into energy. It is important to produce keratin (stronger nails and hair). Promotes good skin health.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCA1	rs1883025	AG-	T	+ -	
ACP7	rs472265	AA+	G	- -	
AR	rs6152	GG+	A	- -	
C2	rs547154	CC-	T	- -	
C3	rs2230199	CG-	C,T	+ -	
C3	rs2230205	AG-	T	- -	
CAPN10	rs3792267	GG+	A	- -	
CDKAL1	rs4712523	AG+	G	- -	
CDKAL1	rs7756992	AG+	G,T	- -	
CDKN2A	rs10811661	TT+	T	+ +	
CDKN2A/B	rs2383208	AA+	G,T	- -	
CETP	rs5880	GG+	C	- -	
CETP	rs5882	AG+	A	+ -	
CETP	rs708272	CT-	A	+ -	
CETP	rs1532624	TT-	A	- -	
CETP	rs1864163	GG+	A	- -	
CETP	rs2303790	AA+	G	- -	
CFH	rs800292	CT-	A	+ -	
CFH	rs1061147	CC+	C	- -	
CFH	rs1061170	TT+	T	- -	
CFH	rs1065489	GT+	T	- -	
CFH	rs1329428	AG-	T	- -	
CFH	rs3753394	CT+	T	- -	
COL8A1	rs13081855	GG+	T	- -	
CX3CR1	rs3732378	AG+	A	+ -	
CX3CR1	rs3732379	CT+	T	- -	
DNER	rs1861612	AG+	A,C	- -	
EPHA2	rs3754334	CC-	A	- -	
FGFR3	rs121913482	CC+	T	- -	

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FGFR3	rs121913483	CC+	A,G,T	- -	🟢
FTO	rs1121980	CC-	A	- -	🟢
GAD1	rs701492	TT+	T	- -	🟡
GAD1	rs2241165	AA-	T	- -	🟢
GCKR	rs780094	AG-	C	- -	🟢
HHEX	rs5015480	TT+	T	- -	🟢
HTRA1	rs11200638	AG+	A	+ -	🟡
IL-6	rs1800795	CG+	G	+ -	🟠
INTERGENIC	rs2223841	AA-	C	- -	🟡
INTERGENIC	rs6625163	AA+	A	- -	🟡
INTERGENIC	rs12678919	AA+	G	- -	🟢
IRS1	rs2943641	CT+	C	+ -	🟡
JAZF1	rs1635852	CT+	C	- -	🟢
KCNJ11	rs5215	TT+	T	- -	🟢
KCNJ11	rs5219	CC+	T	- -	🟢
KCNQ1	rs2283228	AA+	C	- -	🟢
NOTCH4	rs422951	AG-	C	- -	🟢
PAX4	rs2233580	GG-	T	- -	🟢
PITX2	rs6533526	GG+	A	- -	🟢
PPARG	rs1801282	CG+	C	+ -	🟡
REST	rs1713985	AC-	T	- -	🟢
REST	rs2227902	GG+	T	- -	🟢
RHOU	rs6426514	GG+	A	- -	🟢
RPSAP52	rs1531343	CC+	C,T	- -	🟢
SKIV2L	rs429608	GG+	A	- -	🟢
SKIV2L	rs2734331	TT-	G	- -	🟢
SLC30A8	rs13266634	CC+	A,T	+ +	🔴
TCF7L2	rs12255372	GG+	T	- -	🟢
TGFBR3	rs1805110	CC-	A	- -	🟢
TLR3	rs3775291	GG-	G,T	+ +	🟠
TRIB3	rs2295490	AG+	G,T	+ -	🟡
VEGFA	rs3025039	CC+	T	- -	🟢
VPS26A	rs4812829	AG+	A	- -	🟢

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Sample

Vitamin B2

MEDIUM

This vitamin can be found in foods such as whole grains, milk, yogurt, soy, egg and wheat germ, and its deficiency can cause various symptoms in the body.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ETFDH	rs121964954	GG+	A	- -	
FMO3	rs909530	CC+	T	- -	
FMO3	rs1736557	AG+	A	+ -	
FMO3	rs2266780	AA+	G	- -	
FMO3	rs2266782	AG+	A	+ -	
FMO3	rs61753344	GG+	T	- -	
MTHFR	rs1476413	AG-	G,T	+ -	
MTHFR	rs1801133	CC-	A	- -	
SLC52A2	rs375088539	CC+	T	- -	
SLC52A2	rs782345472	CC+	T	- -	
SLC52A3	rs267606684	CC-	A	- -	

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Sample

Vitamin C

MEDIUM

Ascorbic acid or vitamin C is a molecule used in the hydroxylation of various biochemical reactions in cells. Indication in orange or red indicates greater need.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCA1	rs1883025	AG-	T	+ -	
ABCB1	rs1128503	CT-	G	- -	
ABCB1	rs3213619	TT-	G	- -	
ALDH2	rs671	GG+	A	+ +	
APOB	rs676210	AG+	A,T	- -	
APOB	rs1367117	GG+	A	- -	
ATM	rs664143	CC-	G,T	- -	
B3GALT1	rs13020412	AA+	G	- -	
BCO1	rs12934922	AA+	G,T	- -	
CDKN1A	rs1801270	CC+	A,T	- -	
CDKN2A	rs3731249	GG-	A,G,T	- -	
CETP	rs5882	AG+	A	+ -	
CFH	rs1061147	CC+	C	- -	
CFH	rs1061170	TT+	T	- -	
CLPTM1L	rs401681	CT+	T	+ -	
CYP1A1	rs1048943	AA-	A,C,G	- -	
CYP1A1	rs1800031	TT-	G	- -	
CYP1A1	rs41279188	CC-	A,T	- -	
CYP1A1	rs56313657	GG-	A,T	- -	
CYP1A1	rs72547509	TT-	G,T	- -	
CYP1B1	rs1056836	CC-	C	- -	
CYP2E1	rs2070673	TT+	T	- -	
CYP3A4	rs2740574	AA-	T	- -	
CYP3A5	rs776746	AG-	C	+ -	
DEF8	rs4268748	CT+	C	- -	
DIRC3	rs966423	CT+	G,T	+ -	
DPYD	rs1801266	CC-	A	- -	
DPYD	rs1801267	GG-	T	- -	
DPYD	rs1801268	GG-	A	- -	
E2F3	rs1570155	AG+	A,T	+ -	

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Sample

EPHX1	rs1051740	TT+	C	- -	
EPHX1	rs2234922	AG+	G,T	+ -	
ERI1	rs96621	CT+	C	- -	
FASLG	rs763110	TT+	T	- -	
G6PD	rs1050828	CC+	T	- -	
GPX1	rs1050450	CC-	A	- -	
GSTP1	rs1695	GG+	G	+ +	
HDAC4	rs3791406	CT+	C	+ -	
HLA-DRA	rs3135391	CC-	A	- -	
HLA-DRB1	rs660895	GG+	G	+ +	
HTRA1	rs11200638	AG+	A	+ -	
INTERGENIC	rs9287638	CC+	A	- -	
INTERGENIC	rs12661968	TT+	C	- -	
IRF4	rs12203592	CC+	T	- -	
KL	rs9536314	TT+	A,G	- -	
LMNA	rs553016	GG-	G,T	- -	
MC1R	rs1805005	GG+	T	- -	
MC1R	rs1805007	CC+	T	- -	
MC1R	rs1805008	CC+	T	- -	
MC1R	rs1805009	GG+	A,C	- -	
MTHFR	rs1476413	AG-	G,T	+ -	
MTHFR	rs1801131	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	
MTRR	rs1801394	AA+	G	- -	
PIK3CA	rs2699887	CT+	T	- -	
PIK3CA	rs104886003	GG+	A,C	- -	
PIK3CA	rs1057519942	GG+	A	- -	
PPARG	rs17036170	GG+	A	- -	
PTEN	rs121909229	GG+	A,C,T	- -	
PTEN	rs121909232	CC+	G	- -	
RNASEL	rs3738579	CT-	G	+ -	
SLC23A1	rs33972313	GG-	A,G,T	- -	
SLC39A6	rs1050631	CT-	A	+ -	
SLC45A2	rs185146	CT+	T	+ -	
SOD2	rs4880	CT-	G	+ -	

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Sample

SOD3	rs1799895	CC+	G	- -	
STAT4	rs10181656	CG+	C	+ -	
TERT	rs2736098	AG-	T	+ -	
TERT	rs2736100	TT-	A	+ +	
XPC	rs2228000	CC-	A	- -	
XPC	rs2228001	AA-	T	- -	
XRCC1	rs1799782	CT-	A	+ -	

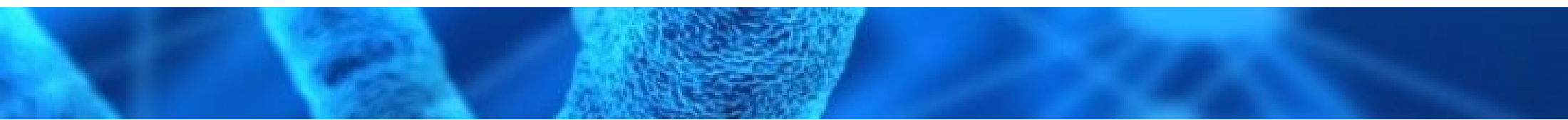
Vitamin A

NORMAL

Vitamin A or Retinol is a micronutrient belonging to the group of fat-soluble vitamins, which can be found in animal tissue in the form of retinoids or as a pro-vitamin in plant tissue, in the form of carotenoid. Indication in orange or red indicates greater need.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BCO1	rs6564851	TT+	G	- -	
BCO1	rs7501331	CC+	T	- -	
BCO1	rs12934922	AA+	G,T	- -	
BCO1	rs119478057	CC+	T	- -	

Weight



Name Sample

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Sample

Adiposity

MEDIUM-HIGH

Adiposity is associated with several secondary diseases such as type 2 diabetes, hypertension, cardiovascular disease, fatty liver and adipose tissue disorders. Essential to the risk of developing these secondary diseases is the body's fat distribution. Especially visceral fat in the belly region is associated with a higher risk, while fat deposits located in the buttocks and upper leg area are less harmful.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AATK	rs7220048	TT+	C	- -	
ADCYAP1	rs1893154	CC-	G	- -	
ADRB3	rs4994	TT-	G	- -	
ADSS	rs3102460	CT+	T	- -	
ANKAR	rs12053254	TT+	C	- -	
ANKK1	rs1800497	CT-	A	+ -	
ARMC4	rs587777047	AA+	C	- -	
ARMC4	rs587777049	GG+	T	- -	
BICC1	rs11006263	AA+	G	- -	
C2CD4C	rs12978500	AC+	A	- -	
C8ORF34	rs1517114	CC+	A,G,T	- -	
CCDC77	rs1048466	AG+	A,C	- -	
CD46	rs35366573	CC+	T	- -	
CDCA3	rs5443	CT+	T	+ -	
CDHR3	rs6967330	GG+	A	- -	
COL4A1	rs3742207	AC-	A,G	- -	
CYP2E1	rs2031920	CC+	T	- -	
CYP2E1	rs2070672	AA+	G	- -	
CYP2E1	rs72559710	GG+	A,C,T	- -	
DAPL1	rs16843372	TT+	C	- -	
DOCK8	rs192864327	GG+	C,T	- -	
ECT2	rs7646507	AG+	A	- -	
EEPD1	rs4302748	AG+	A	- -	
EHF	rs286913	CC-	G,T	- -	
EVA1A	rs17011455	TT+	C	- -	
FAM209B	rs6024938	CT+	T	- -	
FAM71F1	rs6971091	GG+	A	- -	
FARP1	rs688872	TT-	G	- -	

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Age

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Sample

FLJ33534	rs16857178	GG+	A	- -	🟢
FSIP1	rs10152640	AG+	G	- -	🟢
FTO	rs1121980	CC-	A	- -	🟢
FTO	rs1421085	TT+	C	- -	🟢
FTO	rs3751812	GG+	T	- -	🟢
FTO	rs9930506	AA+	G	- -	🟢
FTO	rs9939609	TT+	A	- -	🟢
FTO	rs17817449	TT+	A,G	- -	🟢
FTO	rs121918214	GG+	A	- -	🟢
GPC5	rs2352028	CT+	G,T	+ -	🟠
GSG1L	rs205391	CT+	C,G	- -	🟢
HDAC9	rs11984041	CC+	T	- -	🟢
IL-1A	rs1800587	CT-	A,C	+ -	🟡
IL-1B	rs1143634	CT-	A	- -	🟢
INSIG2	rs7566605	GG+	C	- -	🟢
INTERGENIC	rs2153299	AA-	C	- -	🟢
INTERGENIC	rs2575029	CC+	C	- -	🟢
INTERGENIC	rs5767992	CC+	C	- -	🟢
INTERGENIC	rs6486986	GT+	T	- -	🟢
INTERGENIC	rs10207060	GT+	A,G	- -	🟢
INTERGENIC	rs11070098	TT+	C	- -	🟢
INTERGENIC	rs17054265	CC+	G,T	- -	🟢
INTERGENIC	rs17468244	AA+	G	- -	🟢
KIF6	rs9380880	GG+	A	- -	🟢
LEPR	rs1137101	AA+	G	- -	🟢
LGALS17A	rs8103033	AG+	A	- -	🟢
LHPP	rs12773846	GG+	A,C	- -	🟢
LINC01299	rs6981992	GT+	A,T	- -	🟢
LINC01500	rs405460	AC-	A,T	- -	🟢
LIPC	rs261332	GG+	G	- -	🟢
LIPC	rs1800588	CC+	T	- -	🟡
LPP	rs1152846	AG-	C	+ -	🟢
LYPLAL1	rs2605100	GG+	G	+ +	🟠
MC4R	rs10871777	AG+	G	+ -	🟠
MC4R	rs12970134	AG+	A	- -	🟢

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Sample

MDFIC	rs7784447	GG+	A	- -	
MSRA	rs545854	CG-	G	+ -	
NAT2	rs1208	GG+	G	- -	
NAT2	rs1041983	CC+	T	- -	
NAT2	rs1799929	CT+	T	+ -	
NAT2	rs1801279	GG+	A	- -	
NAT2	rs1801280	CC+	C	- -	
NAT2	rs1805158	CC+	A,T	- -	
NDUFA8	rs3818638	AG-	C	- -	
NIPSNAP3B	rs2472476	AG-	T	- -	
NMNAT2	rs4652795	CT+	T	- -	
NPM2	rs11776272	GG+	G	- -	
NXPH1	rs765855	GG+	G	- -	
PCDH9	rs17081231	AA+	G	- -	
PCSK1	rs6232	AG-	C	+ -	
PFKP	rs6602024	GG+	A	- -	
PKNOX2	rs10893366	CT+	T	- -	
PLEKHG1	rs17427389	GG+	A,T	- -	
POC5	rs2112347	GT+	G	- -	
PPARG	rs3856806	CC+	T	- -	
PTPRD	rs1975197	CC-	A	- -	
PTPRN2	rs10274279	TT+	C	- -	
PVALB	rs2022068	AA+	G	- -	
RAB17	rs2292873	AG-	T	- -	
RASEF	rs10867921	AG+	A	- -	
RSU1	rs11254160	AG+	A	- -	
RYR2	rs1057517873	AA+	G	- -	
S100P	rs3822262	AA-	G	- -	
SLC22A2	rs316019	GG-	C	- -	
SLC22A2	rs8177504	CC-	A,T	- -	
SLC22A2	rs8177507	GG-	G,T	- -	
SLC22A2	rs8177516	CC-	A,T	- -	
SLC22A2	rs8177517	AA-	C,G	- -	
SLC29A3	rs1084004	CC+	C,G	- -	
SLC29A3	rs121912583	GG+	A	- -	

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Name Sample

Age

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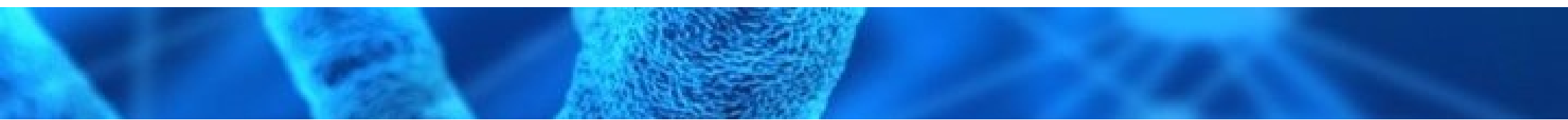
Health insurance

Sample

SMYD3	rs11800820	CC+	A,T	- -	🟢
SPAG16	rs16851771	AA+	G	- -	🟢
SPOCK3	rs9312517	AA+	G	- -	🟢
STON2	rs6574644	AA+	A	- -	🟢
TBC1D1	rs35859249	CC+	A,T	- -	🟢
TCF4	rs613872	TT+	T	- -	🟢
TCF4	rs9960767	CC+	C,G	- -	🟡
TFAP2B	rs987237	AA+	G	- -	🟢
TM9SF2	rs9513627	AA+	A	- -	🟢
TMEM18	rs6548238	CC+	C	- -	🟢
TMEM229B	rs1077989	AC+	C	- -	🟢
TMEM45B	rs10894147	CC+	T	- -	🟢
TMOD1	rs1475545	AG-	T	- -	🟢
TPTE2P1	rs2483374	AC+	C,T	- -	🟢
TRAPPC9	rs267607137	CC-	A	- -	🟢
TRIM66	rs4929923	CC+	C	- -	🟢
UGT2B7	rs12233719	GG+	A,C,T	- -	🟢
UNC13A	rs12608932	AC+	C	+ -	🟡
VSIG10	rs7957470	GG+	G	- -	🟢
WDR11-AS1	rs4783244	GG+	T	- -	🟢
ZBTB46	rs6062314	TT+	G,T	- -	🟢
ZNF536	rs3786800	AA-	C	- -	🟢



Bibliography

A2M: Laing EE, Möller-Levet CS, Poh N, Santhi N, Archer SN, Dijk D-J. Blood transcriptome based biomarkers for human circadian phase. Takahashi JS, ed. eLife. 2017;6:e20214. doi:10.7554/eLife.20214.

AATK: Tanja Haag, Christina E. Herkt, Sara K. Walesch, Antje M. Richter, Reinhard H. DammannGenes Cancer. 2014 Sep; 5(9-10): 365–374. doi: 10.18632/genesandcancer.28PMCID: PMC4209602 Cécile Jacovetti, Veronica Jimenez, Eduard Ayuso, Ross Laybutt, Marie-Line Peyot, Marc Prentki, Fatima Bosch, Romano RegazziMol Endocrinol. 2015 May; 29(5): 693–702. Published online 2015 Mar 9. doi: 10.1210/me.2014-1299PMCID: PMC5414744Aron Kos, Nikkie F. M. Olde Loohuis, Martha L. Wieczorek, Jeffrey C. Glennon

ABCA1: Ishigami M, Ogasawara F, Nagao K, et al. Temporary sequestration of cholesterol and phosphatidylcholine within extracellular domains of ABCA1 during nascent HDL generation. Scientific Reports. 2018;8:6170. doi:10.1038/s41598-018-24428-6.

ABCA2: Davis W. The ATP-binding cassette transporter-2 (ABCA2) regulates esterification of plasma membrane cholesterol by modulation of sphingolipid metabolism. Biochimica et biophysica acta. 2014;1841(1):168-179. doi:10.1016/j.bbalip.2013.10.019.

ABCA7: Aikawa T, Holm M-L, Kanekiyo T. ABCA7 and Pathogenic Pathways of Alzheimer's Disease. Brain Sciences. 2018;8(2):27. doi:10.3390/brainsci8020027.

ABCB1: Parvathaneni RK, DeLeo VL, Spiekerman JJ, Chakraborty D, Devos KM. Parallel loss of introns in the ABCB1 gene in angiosperms. BMC Evolutionary Biology. 2017;17:238. doi:10.1186/s12862-017-1077-x.

ABCC11: Toyoda Y, Takada T, Miyata H, Ishikawa T, Suzuki H. Regulation of the Axillary Osmidrosis-Associated ABCC11 Protein Stability by N-Linked Glycosylation: Effect of Glucose Condition. Brodsky JL, ed. PLoS ONE. 2016;11(6):e0157172. doi:10.1371/journal.pone.0

ABCC6: Pomozi V, Brampton C, van de Wetering K, et al. Pyrophosphate Supplementation Prevents Chronic and Acute Calcification in ABCC6-Deficient Mice. The American Journal of Pathology. 2017;187(6):1258-1272. doi:10.1016/j.ajpath.2017.02.009.

ABCG2: Shu-Ching Liang, Chih-Yung Yang, Ju-Yu Tseng, Hong-Ling Wang, Chien-Yi Tung, Hong-Wen Liu, Chin-Yau Chen, Yi-Chen Yeh, Teh-Ying Chou, Muh-Hwa Yang, Jacqueline Whang-Peng, Chi-Hung LinNeoplasia. 2015 Mar; 17(3): 265–278. Published online 2015 Mar 23. doi: 10.1016/j.neo.2015.01.004PMCID: PMC4372652 Boglárka Zámbó, Zsuzsa Bartos, Orsolya Mózner, Edit Szabó, György Várady, Gyula Poór, Márton Pálinkás, Hajnalka Andrikovics, Tamás Heged?s, László Homolya, Balázs SarkadiSci Rep. 2018;

ABCG8: Genetic variations at ABCG5/G8 genes modulate plasma lipids concentrations in patients with familial hypercholesterolemia. (PMID: 20172523) Garcia-Rios A … Perez-Jimenez F Atherosclerosis 2010 3 21 39 70 A genome-wide association scan identifies the hepatic cholesterol transporter ABCG8 as a susceptibility factor for human gallstone disease. (PMID: 17632509) Buch S … Hampe J Nature genetics 2007 3 4 39 70 Increased gallstone risk in humans conferred by common variant of hepatic ATP-binding cassette transporter for cholesterol. (PMID: 17626266) Grünhage F … Lammert F Hepatology (Baltimore, Md.) 2007 2 3 21 39 Two genes that map to the STSL locus cause sitosterolemia: genomic structure and spectrum of mutations involving sterolin-1 and sterolin-2, encoded by ABCG5 and ABCG8, respectively. (PMID: 11452359) Lu K … Patel SB American journal of human genetics 2001 3 4 21 70 Accumulation of dietary cholesterol in sitosterolemia caused by mutations in adjacent ABC transporters. (PMID: 11099417) Berge KE … Hobbs HH Science (New York, N.Y.) 2000

ABI3BP: Cloning and sequencing of a novel human gene that encodes a putative target protein of Nesh-SH3. (PMID: 11501947) Matsuda S … Hamaguchi M Journal of human genetics 2001 2 3 4 Genome-wide association study of suicide attempts in mood disorder patients. (PMID: 21041247) Perlis RH … Smoller JW The American journal of psychiatry 2010 3 39 Coeliac disease-associated risk variants in TNFAIP3 and REL implicate altered NF-kappaB signalling. (PMID: 19240061) Trynka G … Wijmenga C Gut 2009 3 39 Cancer-associated loss of TARSH gene expression in human primary lung cancer. (PMID: 16205947) Terauchi K … Fushiki S Journal of cancer research and clinical oncology 2006 3 21 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T … Sugano S Nature genetics 2004

ACE: Tikhomirova VE, Kost OA, Kryukova OV, et al. ACE phenotyping in human heart. Bader M, ed. PLoS ONE. 2017;12(8):e0181976. doi:10.1371/journal.pone.0181976. Meta-analysis of genetic studies in ischemic stroke: thirty-two genes involving approximately 18,000 cases and 58,000 controls. (PMID: 15534175) Casas JP ... Sharma P Archives of neurology 2004 3 4 39 70 Structural details on the binding of antihypertensive drugs captopril and enalaprilat to human testicular angiotensin I-converting enzyme. (PMID: 15236580) Natesh R ... Acharya KR Biochemistry 2004 3 4 21 24

ACHE: Joan Torrent, Alba Vilchez-Acosta, Diego Muñoz-Torrero, Marie Trovaslet, Florian Nachon, Arnaud Chatonnet, Katarina Grznarova, Isabelle Acquatella-Tran Van Ba, Ronan Le Goffic, Laetitia Herzog, Vincent Béringue, Human RezaeiActa Neuropathol Commun. 2015; 3: 18. Published online 2015 Apr 3. doi: 10.1186/s40478-015-0188-0 PMCID: PMC4383067 Ronit Heinrich, Rivka Hertz, Esther Zemel, Irit Mann, Liat Brenner, Amir Massarweh, Shai Berlin, Ido PerlmanFront Mol Neurosci. 2018; 11: 88.

ACMSD: Thirtamara-Rajamani K, Li P, Escobar Galvis ML, Labrie V, Brundin P, Brundin L. Is the Enzyme ACMSD a Novel Therapeutic Target in Parkinson's Disease? Journal of Parkinson's Disease. 2017;7(4):577-587. doi:10.3233/JPD-171240.

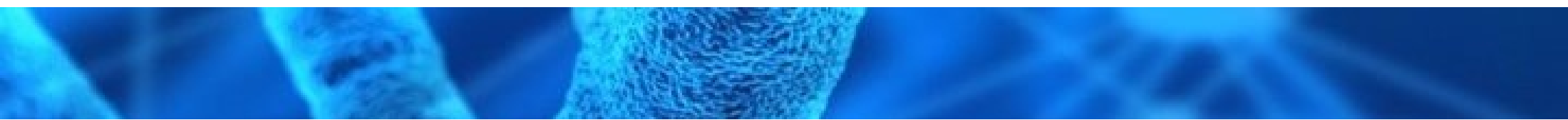
ACP7: David C Cantu, Michael J Forrester, Katherine Charov, Peter J ReillyProtein Sci. 2012 May; 21(5): 655–666. Published online 2012 Feb 28. doi: 10.1002/pro.2050PMCID: PMC3403463Till F. Schäberle, Mahsa Mir Mohseni, Friederike Lohr, Alexander Schmitz, Gabriele M. KönigAntimicrob Agents Chemother. 2014 Feb; 58(2): 950–956. doi: 10.1128/AAC.01894-13PMCID: PMC3910853Yi-Qiang Cheng, Jane M. Coughlin, Si-Kyu Lim, Ben ShenMethods Enzymol. Author manuscript; available in PMC 2015 Jun 25.Published in

ACYP2: Common variants in ACYP2 influence susceptibility to cisplatin-induced hearing loss. (PMID: 25665007) Xu H ... Yang JJ Nature genetics 2015 2 3 GWAS for discovery and replication of genetic loci associated with sudden cardiac arrest in patients with coronary artery disease. (PMID: 21658281) Aouizerat BE ... Tseng ZH BMC cardiovascular disorders 2011 3 40 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 40 Genetic variants in nuclear-encoded mitochondrial genes influence AIDS progression. (PMID: 20877624) Hendrickson SL ... O'Brien SJ PloS one 2010 3 40 Assessment of a polymorphism of SDK1 with hypertension in Japanese Individuals. (PMID: 19851296) Oguri M ... Yamada Y American journal of hypertension 2010

ADA: Predisposição genética de responsividade à terapia para hepatite C. crônica (PMID: 16886895) Hwang Y. Chen DS Farmacogenômica 2006 3 23 45 58 Catabolismo endotelial da adenosina extracelular durante hipóxia: o papel da adenosina desaminase de superfície e CD26. (PMID: 16670267) Eltzschig HK. Colgan SP Blood 2006 3 4 23 58 Uma variação genética funcional da adenosina desaminase afeta a duração e a intensidade do sono profundo em humanos. (PMID: 16221767) Rétey JV. Landolt HP Proceedings da A

ADAMTS12: Timothy J. Mead, Daniel R. McCulloch, Jason C. Ho, Yaoyao Du, Sheila M. Adams, David E. Birk, Suneel S. ApteJCI Insight. 2018 Apr 5; 3(7): e92941. Published online 2018 Apr 5. doi: 10.1172/jci.insight.92941PMCID: PMC5928868Angela Moncada-Pazos, Alvaro J. Obaya, María Llamazares, Ritva Heljasvaara, María F. Suárez, Enrique Colado, Agnès Noël, Santiago Cal, Carlos López-OtínJ Biol Chem. 2012 Nov 16; 287(47): 39554–39563. Published online 2012 Sep 27. doi: 10.1074/jbc.M112.408625Correction in:

ADAMTS2: Novel types of mutation responsible for the dermatosparactic type of Ehlers-Danlos syndrome (Type VIIC) and common polymorphisms in the ADAMTS2 gene. (PMID: 15373769) Colige A ... Nusgens BV The Journal of investigative dermatology 2004 2 3 21 70 Human Ehlers-Danlos syndrome type VII C and bovine dermatosparaxis are caused by mutations in the procollagen I N-proteinase gene. (PMID: 10417273) Colige A ... Nusgens BV American journal of human genetics 1999 3 4 21 70 Expanding the clinical and mutational spectrum of the Ehlers-Danlos syndrome, dermatosparaxis type. (PMID: 26765342) Van Damme T ... Malfait F Genetics in medicine : official journal of the American College of Medical Genetics 2016 3 70 Genome-wide association scan of the time to onset of attention deficit hyperactivity disorder. (PMID: 18937294) Lasky-Su J ... Faraone SV American journal of medical genetics. Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics 2008 3 39 Multiple congenital skull fractures as a presentation of Ehlers-Danlos syndrome type VIIC. (PMID: 18973246) Bar-Yosef O ... Kuint J American journal of medical genetics. Part A 2008 2



ADCY5: Cloning and sequence of partial cDNAs encoding the human type V and VI adenylyl cyclases and subsequent RNA-quantification in various tissues. (PMID: 10481931) Raimundo S . Wisser H Clinica chimica acta; international journal of clinical chemistry 1999 2 3 4 58 Gain-of-function ADCY5 mutations in familial dyskinesia with facial myokymia. (PMID: 24700542) Chen YZ . Torkamani A Annals of neurology 2014 3 4 58 ADCY5 couples glucose to insulin secretion in human islets. (PMID: 24740569) Hodson

ADCYAP1: Gaia Bazzi, Andrea Galimberti, Quentin R. Hays, Ilaria Bruni, Jacopo G. Cecere, Luca Gianfranceschi, Keith A. Hobson, Yolanda E. Morbey, Nicola Saino, Christopher G. Guglielmo, Diego RuboliniEcol Evol. 2016 May; 6(10): 3226–3239. Published online 2016 Apr 7. doi: 10.1002/ece3.2053PMCID: PMC4870208 Andrea Contina, Eli S. Bridge, Jeremy D. Ross, J. Ryan Shipley, Jeffrey F. KellyPLoS One. 2018; 13(1): e0190859. Published online 2018 Jan 11. doi: 10.1371/journal.pone.0190859PMCID: PMC5764313

ADD1: Hsu W, Wang W, Lin W, et al. Adducin-1 is essential for spindle pole integrity through its interaction with TPX2. EMBO Reports. 2018;19(8):e45607. doi:10.15252/embr.201745607.

ADD2: Costessi L, Porro F, Iaconcig A, Muro AF. TDP-43 regulates β-adducin (Add2) transcript stability. RNA Biology. 2014;11(10):1280-1290. doi:10.1080/15476286.2014.996081.

ADGRE2: Boyden SE, Desai A, Cruse G, et al. Vibratory Urticaria Associated with a Missense Variant in ADGRE2. The New England journal of medicine. 2016;374(7):656-663. doi:10.1056/NEJMoa1500611.

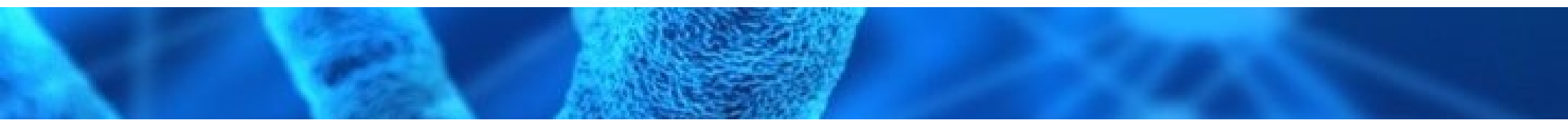
ADIPOQ: Impaired multimerization of human adiponectin mutants associated with diabetes. Molecular structure and multimer formation of adiponectin. (PMID: 12878598) Waki H ... Kadowaki T The Journal of biological chemistry 2003 3 4 21 69 Circulating high molecular weight adiponectin isoform is heritable and shares a common genetic background with insulin resistance in nondiabetic White Caucasians from Italy: evidence from a family-based study. (PMID: 19761474) Menzaghi C ... Trischitta V Journal of internal medicine 2010 3 21 39 Gene-nutrient interactions in the metabolic syndrome: single nucleotide polymorphisms in ADIPOQ and ADIPOR1 interact with plasma saturated fatty acids to modulate insulin resistance. (PMID: 20032495) Ferguson JF ... Roche HM The American journal of clinical nutrition 2010 3 21 39 Sialic acid modification of adiponectin is not required for multimerization or secretion but determines half-life in circulation. (PMID: 19855092) Richards AA ... Whitehead JP Molecular endocrinology (Baltimore, Md.) 2010 3 4 21 C-reactive protein -717C>T genetic polymorphism associates with esophagectomy-induced stress hyperglycemia. (PMID: 20145925) Motoyama S ... Ogawa J World journal of surgery 2010 FGF21/adiponectin ratio predicts deterioration in glycemia: a 4.6-year prospective study in China. (PMID: 34321008) Liu D ... Jia W Cardiovascular diabetology 2021 3 The correlation of salivary telomere length and single nucleotide polymorphisms of the ADIPOQ, SIRT1 and FOXO3A genes with lifestyle-related diseases in a Japanese population. (PMID: 33507936) Han X ... Murohashi I PloS one 2021 3 Adiponectin Gene Variant rs3774261, Effects on Lipid Profile and Adiponectin Levels after a High Polyunsaturated Fat Hypocaloric Diet with Mediterranean Pattern. (PMID: 34073587) de Luis Roman DA ... López JJ Nutrients 2021

ADORA2A: Genetic polymorphism of the adenosine A2A receptor is associated with habitual caffeine consumption. (PMID: 17616786) Cornelis MC . Campos H The American journal of clinical nutrition 2007 3 23 26 45 58 Association between ADORA2A and DRD2 polymorphisms and caffeine-induced anxiety. (PMID: 18305461) Childs E . de Wit H Neuropsychopharmacology : official publication of the American College of Neuropsychopharmacology 2008 The 2.6 angstrom crystal structure of a human A2A adenosine receptor bo

ADRA2A: Linnstaedt SD, Walker MG, Riker KD, et al. Genetic variant rs3750625 in the 3'UTR of ADRA2A affects stress-dependent acute pain severity after trauma and alters a microRNA-34a regulatory site. Pain. 2017;158(2):230-239. doi:10.1097/j.pain.0000000000000742

ADRB2: Slota C, Shi A, Chen G, Bevans M, Weng N. Norepinephrine preferentially modulates memory CD8 T cell function inducing inflammatory cytokine production and reducing proliferation in response to activation. Brain, behavior, and immunity. 2015;46:168-179. do

ADRB3: Li Y, Lu X, Wang H, et al. ADRB3 Gene Trp64Arg Polymorphism and Essential Hypertension: A Meta-Analysis Including 9,555 Subjects. Frontiers in Genetics. 2018;9:106. doi:10.3389/fgene.2018.00106.



ADSS: Ante Bubi?, Natalia Mrnjavac, Igor Stuparevi?, Marta ?yczek, Beata Wielgus-Kutrowska, Agnieszka Bzowska, AšlerJ Enzyme Inhib Med Chem. 2018; 33(1): 1405–1414. Published online 2018 Sep 7. doi: 10.1080/14756366.2018.1506773PMCID: PMC6136348 Jan M. Boitz, Rona Strasser, Phillip A. Yates, Armando Jardim, Buddy UllmanJ Biol Chem. 2013 Mar 29; 288(13): 8977–8990. Published online 2013 Feb 12. doi: 10.1074/jbc.M112.431486PMCID: PMC3610970Ahmed Elhady, Shimaa Adss

AGAP2: Bingqing Hui, Hao Ji, Yetao Xu, Juan Wang, Zhonghua Ma, Chongguo Zhang, Keming Wang, Yan ZhouCell Death Dis. 2019 Mar; 10(3): 207. Published online 2019 Feb 27. doi: 10.1038/s41419-019-1384-9PMCID: PMC6393474Yegor Doush, Arif A. Surani, Amaia Navarro-Corcuera, Stephanie McArdle, E. Ellen Billett, Cristina Montiel-DuarteSci Rep. 2019; 9: 390. Published online 2019 Jan 23. doi: 10.1038/s41598-018-36888-xPMCID: PMC6344547Huaying Dong, Wei Wang, Shaowei Mo, Ru Chen, Kejian Zou, Jing Han

AGER: Kumar V, Fleming T, Terjung S, et al. Homeostatic nuclear RAGE-ATM interaction is essential for efficient DNA repair. Nucleic Acids Research. 2017;45(18):10595-10613. doi:10.1093/nar/gkx705.

AGPAT2: Cloning and expression of two human lysophosphatidic acid acyltransferase cDNAs that enhance cytokine-induced signaling responses in cells. (PMID: 9212163) West J . Leung DW DNA and cell biology 1997 2 3 4 23 58 Enzymatic activity of naturally occurring 1-acylglycerol-3-phosphate-O-acyltransferase 2 mutants associated with congenital generalized lipodystrophy. (PMID: 15629135) Haque W . Agarwal AK Biochemical and biophysical research communications 2005 3 4 23 58 AGPAT2 is mutated in congen

AGT: Krisztián Fodor, Janina Wolf, Ralf Erdmann, Wolfgang Schliebs, Matthias WilmannsPLoS Biol. 2012 Apr; 10(4): e1001309. Published online 2012 Apr 17. doi: 10.1371/journal.pbio.1001309PMCID: PMC3328432Ryousuke Satou, Hiroyuki Kobori, Akemi Katsurada, Kayoko Miyata, L. Gabriel NavarAm J Physiol Renal Physiol. 2016 Dec 1; 311(6): F1211–F1216. Published online 2016 Aug 10. doi: 10.1152/ajprenal.00320.2016PMCID: PMC5210198 Xin-Ke Chen, Li-Juan Ouyang, Zheng-Qin Yin, Yuan-You Xia, Xiu-Rong Chen

AGTR1: Youguang Pu, Fangfang Zhao, Yinpeng Li, Mingda Cui, Haiyan Wang, Xianghui Meng, Shanbao CaiBMC Cancer. 2017; 17: 45. Published online 2017 Jan 10. doi: 10.1186/s12885-016-3002-xPMCID: PMC5223322Anukriti Singh, Nidhi Srivastava, Sonal Amit, S.N. Prasad, M.P. Misra, Bushra AteeqTransl Oncol. 2018 Apr; 11(2): 233–242. Published online 2018 Feb 3. doi: 10.1016/j.tranon.2017.12.007PMCID: PMC5884113 Suwattanee Kooptiwut, Keerati Wanchai, Namoiy Semprasert, Chatchawan Srisawat, Pa-thai Yenchitsoma

AHCY: Effect of genetic variation in the human S-adenosylhomocysteine hydrolase gene on total homocysteine concentrations and risk of recurrent venous thrombosis. (PMID: 15241484) Gellekink H . Blom HJ European journal of human genetics : EJHG 2004 3 4 23 45 58 S-adenosylhomocysteine hydrolase deficiency in a human: a genetic disorder of methionine metabolism. (PMID: 15024124) Baric I . Mudd SH Proceedings of the National Academy of Sciences of the United States of America 2004 3 4 23 58 Catalyti

AHI1: Karina Tuz, Yi-Chun Hsiao, Oscar Juárez, Bingxing Shi, Erin Y. Harmon, Ian G. Phelps, Michelle R. Lennartz, Ian A. Glass, Dan Doherty, Russell J. FerlandJ Biol Chem. 2013 May 10; 288(19): 13676–13694. Published online 2013 Mar 26. doi: 10.1074/jbc.M112.420786PMCID: PMC3650405 Gilly Wolf, Tzuri Lifschytz, Hagar Ben-Ari, Pavel Tatarskyy, Tirzah Kreisel Merzel, Amit Lotan, Bernard LererTransl Psychiatry. 2018; 8: 124. Published online 2018 Jul 2. doi: 10.1038/s41398-018-0171-1PMCID: PMC6028478

AHR: Genome-wide meta-analysis identifies regions on 7p21 (AHR) and 15q24 (CYP1A2) as determinants of habitual caffeine consumption. Cornelis MC1, Monda KL, Yu K, Paynter N, Azzato EM, Bennett SN, Berndt SI, Boerwinkle E, Chanock S, Chatterjee N, Couper D, Curhan G, Heiss G, Hu FB, Hunter DJ, Jacobs K, Jensen MK, Kraft P, Landi MT, Nettleton JA, Purdue MP, Rajaraman P, Rimm EB, Rose LM, Rothman N, Silverman D, Stolzenberg-Solomon R, Subar A, Yeager M, Chasman DI, van Dam RM, Caporaso NE. Seok S-

AK8: Mukai T, Crnkovi? A, Umehara T, Ivanova NN, Kyrpides NC, Söll D. RNA-Dependent Cysteine Biosynthesis in Bacteria and Archaea. Harwood CS, ed. mBio. 2017;8(3):e00561-17. doi:10.1128/mBio.00561-17.

AKT1: Mutational profile of advanced primary and metastatic radioactive iodine-refractory thyroid cancers reveals distinct pathogenetic roles for BRAF, PIK3CA, and AKT1. (PMID: 19487299) Ricarte-Filho JC ... Fagin JA Cancer research 2009 3 22 40 71 Detection of the transforming AKT1 mutation E17K in non-small cell lung cancer by high resolution melting. (PMID: 18611285) Do H ... Dobrovic A BMC research notes 2008 3 22 40 71 A transforming mutation in the pleckstrin homology domain of AKT1 in cancer. (PMID: 17611497) Carpten JD ... Thomas JE Nature 2007 3 4 22 71 Isolation and characterization of the human AKT1 gene, identification of 13 single nucleotide polymorphisms (SNPs), and their lack of association with Type II diabetes. (PMID: 11508278) Matsubara A ... Permutt MA Diabetologia 2001 3 4 22 40 Germline PIK3CA and AKT1 mutations in Cowden and Cowden-like syndromes. (PMID: 23246288) Orloff MS ... Eng C American journal of human genetics 2013

ALAS2: Systematic molecular genetic analysis of congenital sideroblastic anemia: evidence for genetic heterogeneity and identification of novel mutations. (PMID: 19731322) Bergmann AK . Neufeld EJ Pediatric blood & cancer 2010 3 4 23 58 C-terminal deletions in the ALAS2 gene lead to gain of function and cause X-linked dominant protoporphyria without anemia or iron overload. (PMID: 18760763) Whatley SD . Puy H American journal of human genetics 2008 3 4 23 58 The major splice variant of human 5-ami

ALDH2: Kuroda A, Hegab AE, Jingtao G, et al. Effects of the common polymorphism in the human aldehyde dehydrogenase 2 (ALDH2) gene on the lung. Respiratory Research. 2017;18:69. doi:10.1186/s12931-017-0554-5.

ALK: Sharma GG, Mota I, Mologni L, Patrucco E, Gambacorti-Passerini C, Chiarle R. Tumor Resistance against ALK Targeted Therapy-Where It Comes From and Where It Goes. Cancers. 2018;10(3):62. doi:10.3390/cancers10030062.

ALLC: Christina B. Cooley, Lisa M. Ryno, Lars Plate, Gareth J. Morgan, John D. Hulleman, Jeffery W. Kelly, R. Luke WisemanProc Natl Acad Sci U S A. 2014 Sep 9; 111(36): 13046–13051. Published online 2014 Aug 25. doi: 10.1073/pnas.1406050111PMCID: PMC4246986 Gareth J Morgan, Jeffery W KellyJ Mol Biol. Author manuscript; available in PMC 2017 Oct 23.Published in final edited form as: J Mol Biol. 2016 Oct 23; 428(21): 4280–4297. Published online 2016 Aug 26

ALOX12: William H. Witola, Susan Ruosu Liu, Alexandre Montpetit, Ruth Welti, Magali Hypolite, Mary Roth, Ying Zhou, Ernest Mui, Marie-France Cesbron-Delauw, Gilbert J. Fournie, Pierre Cavailles, Cordelia Bisanz, Kenneth Boyer, Shawn Withers, A. Gwendolyn Noble, Charles N. Swisher, Peter T. Heydemann, Peter Rabiah, Stephen P. Muench, Rima McLeodInfect Immun. 2014 Jul; 82(7): 2670–2679. doi: 10.1128/IAI.01505-13PMCID: PMC4097613Soeun Kim, Irene F. Choi, Jessica R. Quante, Lei Zhang, Dennis R. Roop, M

ALOX15: Yaoyu Chen, Cong Peng, Sheela A. Abraham, Yi Shan, Zhiru Guo, Ngoc Desouza, Giulia Cheloni, Dongguang Li, Tessa L. Holyoake, Shaoguang LiJ Clin Invest. 2014 Sep 2; 124(9): 3847–3862. Published online 2014 Aug 8. doi: 10.1172/JCI66129PMCID: PMC4151200Dmitry Namgaladze, Ryan G. Snodgrass, Carlo Angioni, Nina Grossmann, Nathalie Dehne, Gerd Geisslinger, Bernhard BrüneJ Biol Chem. 2015 Oct 2; 290(40): 24484–24494. Published online 2015 Aug 14. doi: 10.1074/jbc.M115.678243PMCID: PMC4591829Ryan G

ALOX5: 5-lipoxygenase pharmacogenetics in asthma: overlap with Cys-leukotriene receptor antagonist loci. (PMID: 19214143) Tantisira KG . Weiss ST Pharmacogenetics and genomics 2009 3 23 43 56 Coactosin-like protein functions as a stabilizing chaperone for 5-lipoxygenase: role of tryptophan 102. (PMID: 19807693) Esser J . Rådmark O The Biochemical journal 2009 3 4 23 56 Phosphorylation of serine 271 on 5-lipoxygenase and its role in nuclear export. (PMID: 18978352) Flamand N . Brock TG The Journal

ANKAR: Tijen Ceylan, Gökhan Kuran, Esra Bilgin, Fatih ÇelenkIran Red Crescent Med J. 2012 Aug; 14(8): 475–478. Published online 2012 Aug 30.PMCID: PMC3470841 Resmiye E. Tirali, Haluk Bodur, Gülden EceMed Oral Patol Oral Cir Bucal. 2012 May; 17(3): e517–e522. Published online 2011 Dec 6. doi: 10.4317/medoral.17566PMCID: PMC3476099 Francisco J. A. Nascimento, Agnes M. L. Karlson, Johan Näslund, Ragnar ElmgrenOecologia. 2011 Jun; 166-166(2): 337–347. Published online 2010 Dec

ANKK1: Rubio-Solsona E, Martí S, Vílchez JJ, Palau F, Hoenicka J. ANKK1 is found in myogenic precursors and muscle fibers subtypes with glycolytic metabolism. Alway SE, ed. PLoS ONE. 2018;13(5):e0197254. doi:10.1371/journal.pone.0197254.

ANKRD1: Jiménez AP, Traum A, Boettger T, Hackstein H, Richter AM, Dammann RH. The tumor suppressor RASSF1A induces the YAP1 target gene ANKRD1 that is epigenetically inactivated in human cancers and inhibits tumor growth. Oncotarget. 2017;8(51):88437-88452. doi:1

ANXA11: Hua K, Li Y, Zhao Q, Fan L, Tan B, Gu J. Downregulation of Annexin A11 (ANXA11) Inhibits Cell Proliferation, Invasion, and Migration via the AKT/GSK-3? Pathway in Gastric Cancer. Medical Science Monitor?: International Medical Journal of Experimental and

AOC1: Nguyen T-N, Tuan PA, Mukherjee S, Son S, Ayele BT. Hormonal regulation in adventitious roots and during their emergence under waterlogged conditions in wheat. Journal of Experimental Botany. 2018;69(16):4065-4082. doi:10.1093/jxb/ery190.

APH1B: Biundo F, Ishiwari K, Del Prete D, D'Adamio L. Deletion of the ?-secretase subunits Aph1B/C impairs memory and worsens the deficits of knock-in mice modeling the Alzheimer-like famílial Danish dementia. Oncotarget. 2016;7(11):11923-11944. doi:10.18632/onc

APOA1: Stela Z. Berisha, Greg Brubaker, Takhar Kasumov, Kimberly T. Hung, Patricia M. DiBello, Ying Huang, Ling Li, Belinda Willard, Katherine A. Pollard, Laura E. Nagy, Stanley L. Hazen, Jonathan D. SmithJ Lipid Res. 2015 Mar; 56(3): 653-664. doi: 10.1194/jlr.M056754PMCID: PMC4340312Ying Huang, Joseph A. DiDonato, Bruce S. Levison, Dave Schmitt, Lin Li, Yuping Wu, Jennifer Buffa, Timothy Kim, Gary Gerstenecker, Xiaodong Gu, Chandra Kadiyala, Zeneng Wang, Miranda K. Culley, Jennie E. Hazen, Anthon

APOA4: Wangshu Qin, Xinzhi Li, Liwei Xie, Sha Li, Jianan Liu, Linna Jia, Xue Dong, Xiaomeng Ren, Junjie Xiao, Changqing Yang, Yifa Zhou, Zheng ChenNucleic Acids Res. 2016 Jul 27; 44(13): 6423–6433. Published online 2016 Apr 29. doi: 10.1093/nar/gkw341PMCID: PMC5291254Yupeng Zhang, Jing He, Jing Zhao, Min Xu, Danwen Lou, Patrick Tso, Zongfang Li, Xiaoming LiBiochem Biophys Res Commun. Author manuscript; available in PMC 2018 May 27.Published in final edited form as: Biochem Biophys Res Commun. 2017

APOA5: Lin E, Kuo P-H, Liu Y-L, Yang AC, Kao C-F, Tsai S-J. Association and interaction of APOA5, BUD13, CETP, LIPA and health-related behavior with metabolic syndrome in a Taiwanese population. Scientific Reports. 2016;6:36830. doi:10.1038/srep36830.

APOB: Niu C, Luo Z, Yu L, et al. Associations of the APOB rs693 and rs17240441 polymorphisms with plasma APOB and lipid levels: a meta-analysis. Lipids in Health and Disease. 2017;16:166. doi:10.1186/s12944-017-0558-7.

APOC1: Association of apolipoproteins e4 and c1 with onset age and memory: a study of sporadic Alzheimer disease in Taiwan. (PMID: 20145290) Chuang WL . Huang CC Journal of geriatric psychiatry and neurology 2010 3 23 45 58 APOC1 T45S polymorphism is associated with reduced obesity indices and lower plasma concentrations of leptin and apolipoprotein C-I in aboriginal Canadians. (PMID: 19812053) Lahiry P . Hegele RA Journal of lipid research 2010 3 23 45 58 ApoE and ApoC-I polymorphisms: associatio

APOC3: Apolipoprotein C3 polymorphisms, cognitive function and diabetes in Caribbean origin Hispanics. (PMID: 19424489) Smith CE . Ordovás JM PloS one 2009 3 23 45 58 A null mutation in human APOC3 confers a favorable plasma lipid profile and apparent cardioprotection. (PMID: 19074352) Pollin TI . Shuldiner AR Science (New York, N.Y.) 2008 3 23 45 58 Structure and dynamics of human apolipoprotein CIII. (PMID: 18408013) Gangabadage CS . Wijmenga SS The Journal of biological chemistry 2008 3 4 23 58

APOE: Achariyar TM, Li B, Peng W, et al. Glymphatic distribution of CSF-derived apoE into brain is isoform specific and suppressed during sleep deprivation. Molecular Neurodegeneration. 2016;11:74. doi:10.1186/s13024-016-0138-8. A influência genética sobre a memória humana: uma revisão Fabiana Michelsen de Andrade1*, Vanessa Kappel da Silva 1 e Jaqueline Bohrer Schuch 2 The role of cigarette smoking and statins in the development of postmenopausal osteoporosis: a pilot study utilizing the Marshfi

APOE4: ApoE-Isoform-Dependent SARS-CoV-2 Neurotropism and Cellular Response. (PMID: 33450186) Wang C ... Shi Y Cell stem cell 2021 3 4 Cytokine polymorphisms and Alzheimer disease: possible associations. (PMID: 20213229) Ribizzi G ... Megna M Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology 2010 3 39 Cognitive function after major noncardiac surgery, apolipoprotein E4 genotype, and biomarkers of brain injury. (PMID: 20216394) McDonagh DL ... Neurologic Outcome Research Group Anesthesiology 2010 3 39 MTHFR C677T, FII G20210A, FV Leiden G1691A, NOS3 intron 4 VNTR, and APOE epsilon4 gene polymorphisms are not associated with spontaneous cervical artery dissection. (PMID: 20446941) Jara-Prado A ... Arauz A International journal of stroke : official journal of the International Stroke Society 2010 3 39 Association analysis of CbetaS 844ins68 and MTHFD1 G1958A polymorphisms with Alzheimer's disease in Chinese. (PMID: 20217437) Bi XH ... Zhang JW Journal of neural transmission (Vienna, Austria : 1996) 2010 3 39 Effect of Alzheimer disease genetic risk disclosure on dietary supplement use. (PMID: 20219963) Vernarelli JA ... Green RC The American journal of clinical nutrition 2010 3 39 Age-at-Onset and APOE-Related Heterogeneity in Pathologically Confirmed Sporadic Alzheimer Disease. (PMID: 33722993) Smirnov DS ... Salmon DP Neurology 2021 3 Human-lineage-specific genomic elements are associated with neurodegenerative disease and APOE transcript usage. (PMID: 33824317) Chen Z ... Ryten M Nature communications 2021 3 Low CD4+ cell count nadir exacerbates the impacts of APOE ε4 on functional connectivity and memory in adults with HIV. (PMID: 33587445) Yang FN ... Jiang X AIDS (London, England) 2021 3 Lipid-bound ApoE3 self-assemble into elliptical disc-shaped particles. (PMID: 33189719) Larsen AH ... Midtgaard SR Biochimica et biophysica acta. Biomembranes 2021 3 Testing influences of APOE and BDNF genes and heart failure on cognitive function. (PMID: 32703621) Jung M ... Pressler SJ Heart & lung : the journal of critical care 2021 3 Association of Subjective Hearing Loss and Apolipoprotein E ε4 Allele on Alzheimer's Disease Neurodegeneration. (PMID: 33301284) Neff RM ... McNulty B Otology & neurotology : official publication of the American Otological Society, American Neurotology Society [and] European Academy of Otology and Neurotology 2021 3 Association of APOE Genotype With Heterogeneity of Cognitive Decline Rate in Alzheimer Disease. (PMID: 33771840) Qian J ... Serrano-Pozo A Neurology 2021 3 Differences in Recycling of Apolipoprotein E3 and E4-LDL Receptor Complexes-A Mechanistic Hypothesis. (PMID: 34068576) Kim M ... Bezprozvanny I International journal of molecular sciences 2021 3 TOMM40 and APOE variants synergistically increase the risk of Alzheimer's disease in a Chinese population. (PMID: 32725468) Zhu Z ... Ding D Aging clinical and experimental research 2021 3 Association of Apolipoprotein E Polymorphisms and Risks of Ischemic Stroke in Chinese Patients with Type 2 Diabetes Mellitus. (PMID: 33490286) Wang N ... Ma L Journal of diabetes research 2021 3 Association of APOE4 and Clinical Variability in Alzheimer Disease With the Pattern of Tau- and Amyloid-PET. (PMID: 33262228) La Joie R ... Rabinovici GD Neurology 2021 3 The sex-specific effect of the apolipoprotein E allele and methylenetetrahydrofolate reductase gene polymorphism on the biochemical, anatomical, and cognitive profiles of patients clinically diagnosed with probable Alzheimer's disease. (PMID: 33166415) Kim HJ ... Park J International journal of geriatric psychiatry 2021 3 APOE and Alzheimer's disease: advances in genetics, pathophysiology, and therapeutic approaches. (PMID: 33340485) Serrano-Pozo A ... Hyman BT The Lancet. Neurology 2021 3 [Correlation of cerebrospinal fluid amyloid β-protein 42 and neurofilament light protein levels with postoperative neurocognitive dysfunction in elderly patients]. (PMID: 33963718) Zhang X ... Fu Q Nan fang yi ke da xue xue bao = Journal of Southern Medical University 2021 3 Association of Apolipoprotein E gene polymorphism with the risk of T2DM and obesity among Egyptian subjects. (PMID: 33059023) Galal AA ... Elshazli RM Gene 2021 3 Effects of APOE e4-allele and mental work demands on cognitive decline in old age: Results from the German Study on Ageing, Cognition, and Dementia in Primary Care Patients (AgeCoDe). (PMID: 32819031) Rodriguez FS ... Riedel-Heller SG International journal of geriatric psychiatry 2021 3 APOE ε4 and cognitive reserve effects on the functional network in the Alzheimer's disease spectrum. (PMID: 32314201) Li T ... Alzheimer’s Disease Neuroimaging Initiative Brain imaging and behavior 2021 3 High density lipoprotein structure-function and role in reverse cholesterol transport. (PMID: 20213545) Lund-Katz S ... Phillips MC Sub-cellular biochemistry 2010 21 Combination of apolipoprotein E2 and lipoprotein lipase heterozygosity causes severe hypertriglyceridemia during pregnancy. (PMID: 16142021) Hiéronimus S ... Fredenrich A Diabetes & metabolism 2005 3 A liver X receptor and retinoid X receptor heterodimer mediates apolipoprotein E expression, secretion and cholesterol homeostasis in astrocytes. (PMID: 14720212) Liang Y ... Paul SM Journal of neurochemistry 2004

APP: Hereditary cerebral hemorrhage with amyloidosis associated with the E693K mutation of APP. (PMID: 20697050) Bugiani O ... Tagliavini F Archives of neurology 2010 3 4 41 Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. (PMID: 18667258) Guerreiro RJ ... Clarimón J Neurobiology of aging 2010 3 23 41 APP and BACE1 miRNA genetic variability has no major role in risk for Alzheimer disease. (PMID: 19462468) Bettens K ... Van Broeckhoven C Human mutation 2009 3 23 41 High striatal amyloid beta-peptide deposition across different autosomal Alzheimer disease mutation types. (PMID: 20008660) Villemagne VL ... Rowe CC Archives of neurology 2009 3 23 41 Regulation of FE65 nuclear translocation and function by amyloid beta-protein precursor in osmotically stressed cells. (PMID: 18468999) Nakaya T ... Suzuki T The Journal of biological chemistry 2008

AQP3: Structure and chromosomal localization of a human water channel (AQP3) gene. (PMID: 7558005) Ishibashi K . Marumo F Genomics 1995 2 3 4 23 56 AQP3 deficiency in humans and the molecular basis of a novel blood group system, GIL. (PMID: 12239222) Roudier N . Bailly P The Journal of biological chemistry 2002 3 4 23 56 Human adipose glycerol flux is regulated by a pH gate in AQP10. (PMID: 30420639) Gotfryd K . Gourdon P Nature communications 2018 3 4 56 Variation at the NFATC2 locus increases t

AR: Sakkiah S, Kusko R, Pan B, et al. Structural Changes Due to Antagonist Binding in Ligand Binding Pocket of Androgen Receptor Elucidated Through Molecular Dynamics Simulations. Frontiers in Pharmacology. 2018;9:492. doi:10.3389/fphar.2018.00492.

ARHGAP11A: Bin Dai, Xuan Zhang, Runze Shang, Jianlin Wang, Xisheng Yang, Hong Zhang, Qi Liu, Desheng Wang, Lin Wang, Kefeng DouCell Commun Signal. 2018; 16: 99. Published online 2018 Dec 13. doi: 10.1186/s12964-018-0312-4PMCID: PMC6293628 Yoshinori Kagawa, Shinji Matsumoto, Yuji Kamioka, Koshi Mimori, Yoko Naito, Taeko Ishii, Daisuke Okuzaki, Naohiro Nishida, Sakae Maeda, Atsushi Naito, Junichi Kikuta, Keizo Nishikawa, Junichi Nishimura, Naotsugu Haraguchi, Ichiro Takemasa, Tsunekazu Mizushima

ARHGAP20: Bao H, Li F, Wang C, et al. Structural Basis for the Specific Recognition of RhoA by the Dual GTPase-activating Protein ARAP3. The Journal of Biological Chemistry. 2016;291(32):16709-16719. doi:10.1074/jbc.M116.736140.

ARHGAP24: Gaosi Xu, Xiongbing Lu, Tianlun Huang, Jie FanOncotarget. 2016 Aug 9; 7(32): 51829–51839. Published online 2016 Jul 2. doi: 10.18632/oncotarget.10386PMCID: PMC5239517Lei Wang, Saie Shen, Mingsong Wang, Fangbao Ding, Haibo Xiao, Guoqing Li, Fengqing HuMed Sci Monit. 2019; 25: 21–31. Published online 2019 Jan 1. doi: 10.12659/MSM.911503PMCID: PMC6327779 Xianping Dai, Feng Geng, Jiale Dai, Mengshun Li, Ming LiuMed Sci Monit. 2018; 24: 8669–8677. Published online 2018 Nov 30. doi: 10.12659/MSM.

ARHGAP30: Zhang L, Cao H, He T, et al. Overexpression of PRDM13 inhibits glioma cells via Rho and GTP enzyme activation protein. International Journal of Molecular Medicine. 2018;42(2):966-974. doi:10.3892/ijmm.2018.3679.

ARL15: Nuno Rocha, Felicity Payne, Isabel Huang-Doran, Alison Sleigh, Katherine Fawcett, Claire Adams, Anna Stears, Vladimir Saudek, Stephen O'Rahilly, Inês Barroso, Robert K. SempleSci Rep. 2017; 7: 17593. Published online 2017 Dec 14. doi: 10.1038/s41598-017-17746-8PMCID: PMC5730586 Tanguy Corre, Francisco J. Arjona, Caroline Hayward, Sonia Youhanna, Jeroen H.F. de Baaij, Hendrica Belge, Nadine Nägele, Huguette Debaix, Maxime G. Blanchard, Michela Traglia, Sarah E. Harris, Sheila Ulivi, Rico Rue

ARMC4: Rim Hjeij, Anna Lindstrand, Richard Francis, Maimoona A. Zariwala, Xiaoqin Liu, You Li, Rama Damerla, Gerard W. Dougherty, Marouan Abouhamed, Heike Olbrich, Niki T. Loges, Petra Pennekamp, Erica E. Davis, Claudia M.B. Carvalho, Davut Pehlivan, Claudius Werner, Johanna Raidt, Gabriele Köhler, Karsten Häffner, Miguel Reyes-Mugica, James R. Lupski, Margaret W. Leigh, Margaret Rosenfeld, Lucy C. Morgan, Michael R. Knowles, Cecilia W. Lo, Nicholas Katsanis, Heymut Omran

ARMS2: Micklisch S, Lin Y, Jacob S, et al. Age-related macular degeneration associated polymorphism rs10490924 in ARMS2 results in deficiency of a complement activator. Journal of Neuroinflammation. 2017;14:4. doi:10.1186/s12974-016-0776-3.

ARRB2: Wang H, Deng Q-W, Peng A-N, et al. ?-arrestin2 functions as a key regulator in the sympathetic-triggered immunodepression after stroke. Journal of Neuroinflammation. 2018;15:102. doi:10.1186/s12974-018-1142-4.

ARVCF: Rappe U, Schlechter T, Aschoff M, Hotz-Wagenblatt A, Hofmann I. Nuclear ARVCF Protein Binds Splicing Factors and Contributes to the Regulation of Alternative Splicing. The Journal of Biological Chemistry. 2014;289(18):12421-12434. doi:10.1074/jbc.M113.530

ASIC2: Kimberly P. Gannon, Susan E. McKey, David E. Stec, Heather A. DrummondAm J Physiol Renal Physiol. 2015 Feb 15; 308(4): F339–F348. Published online 2014 Dec 17. doi: 10.1152/ajprenal.00572.2014PMCID: PMC4329487Zhi-hang Zhou, Jin-wen Song, Wen Li, Xue Liu, Liu Cao, Lu-ming Wan, Ying-xia Tan, Shou-ping Ji, Yu-mei Liang, Feng GongJ Exp Clin Cancer Res. 2017; 36: 130. Published online 2017 Sep 19. doi: 10.1186/s13046-017-0599-9PMCID: PMC5606037 Margaret P. Price, Huiyu Gong, Meredith G. Parsons

ASIP: Liu Y, Albrecht E, Schering L, et al. Agouti Signaling Protein and Its Receptors as Potential Molecular Markers for Intramuscular and Body Fat Deposition in Cattle. Frontiers in Physiology. 2018;9:172. doi:10.3389/fphys.2018.00172.

ASL: Regulation of cellular metabolism by protein lysine acetylation. (PMID: 20167786) Zhao S . Guan KL Science (New York, N.Y.) 2010 3 4 23 58 Functional complementation in yeast allows molecular characterization of missense argininosuccinate lyase mutations. (PMID: 19703900) Trevisson E . Salviati L The Journal of biological chemistry 2009 3 4 23 58 Argininosuccinate lyase deficiency: mutational spectrum in Italian patients and identification of a novel ASL pseudogene. (PMID: 17326097) Treviss

ASTN2: Chang H, Smallwood PM, Williams J, Nathans J. Intramembrane Proteolysis of Astrotactins. The Journal of Biological Chemistry. 2017;292(8):3506-3516. doi:10.1074/jbc.M116.768077.

ATF1: Fusion of the EWSR1 and ATF1 genes without expression of the MITF-M transcript in angiomatoid fibrous histiocytoma. (PMID: 15884099) Hallor KH ... Panagopoulos I Genes, chromosomes & cancer 2005 3 4 23 EWS and ATF-1 gene fusion induced by t(12;22) translocation in malignant melanoma of soft parts. (PMID: 8401579) Zucman J ... Thomas G Nature genetics 1993 2 3 23 The cAMP-regulated enhancer-binding protein ATF-1 activates transcription in response to cAMP-dependent protein kinase A. (PMID: 1655749) Rehfuss RP ... Goodman RH The Journal of biological chemistry 1991 3 4 23 Multiple cDNA clones encoding nuclear proteins that bind to the tax-dependent enhancer of HTLV-1: all contain a leucine zipper structure and basic amino acid domain. (PMID: 2196176) Yoshimura T ... Yoshida M The EMBO journal 1990 3 4 23 GWAS for discovery and replication of genetic loci associated with sudden cardiac arrest in patients with coronary artery disease. (PMID: 21658281) Aouizerat BE ... Tseng ZH BMC cardiovascular disorders 2011

ATM: Qian M, Liu Z, Peng L, et al. Boosting ATM activity alleviates aging and extends lifespan in a mouse model of progeria. Kaeberlein M, ed. eLife. 2018;7:e34836. doi:10.7554/eLife.34836.

ATP2B1: Okuyama Y, Hirawa N, Fujita M, et al. The effects of anti-hypertensive drugs and the mechanism of hypertension in vascular smooth muscle cell-specific ATP2B1 knockout mice. Hypertension Research. 2018;41(2):80-87. doi:10.1038/hr.2017.92.

ATP6V1B1: Gueutin V, Vallet M, Jayat M, et al. Renal β-intercalated cells maintain body fluid and electrolyte balance. The Journal of Clinical Investigation. 2013;123(10):4219-4231. doi:10.1172/JCI63492.

ATP8B4: Gao L, Emond MJ, Louie T, et al. Whole-exome Sequencing Identifies Rare Variants in ATP8B4 as a Risk Factor for Systemic Sclerosis. Arthritis & rheumatology (Hoboken, NJ). 2016;68(1):191-200. doi:10.1002/art.39449.

AURKA: Wang J, Nikhil K, Viccaro K, Chang L, White J, Shah K. Phosphorylation-dependent regulation of ALDH1A1 by Aurora kinase A: insights on their synergistic relationship in pancreatic cancer. BMC Biology. 2017;15:10. doi:10.1186/s12915-016-0335-5.

AUTS2: Zhonghua Gao, Pedro Lee, James M. Stafford, Melanie von Schimmelmann, Anne Schaefer, Danny ReinbergNature. 2014 Dec 18; 516(7531): 349–354. doi: 10.1038/nature13921PMCID: PMC4323097 Olivia Engmann, Benoit Labonte, Amanda Mitchell, Pavel Bashtrykov, Erin S. Calipari, Chaggai Rosenbluh, Yong-Hwee E. Loh, Deena M. Walker, Dominika Burek, Peter J. Hamilton, Orna Issler, Rachael L. Neve, Gustavo Turecki, Yasmin Hurd, Andrew Chess, Li Shen, Isabelle Mansuy, Albert Jeltsch, Schahram Akbarian

B3GALT1: A family of human beta3-galactosyltransferases. Characterization of four members of a UDP-galactose:beta-N-acetyl-glucosamine/beta-nacetyl-galactosamine beta-1,3-galactosyltransferase family. (PMID: 9582303) Amado M . Clausen H The Journal of biological chemistry 1998 2 3 4 23 58 Generation and annotation of the DNA sequences of human chromosomes 2 and 4. (PMID: 15815621) Hillier LW . Wilson RK Nature 2005 3 4 58 The status, quality, and expansion of the NIH full-length cDNA project: the Ma

B4GALT7: Crystal structures of ?-1,4-galactosyltransferase 7 enzyme reveal conformational changes and substrate binding. (PMID: 24052259) Tsutsui Y . Qasba PK The Journal of biological chemistry 2013 2 3 4 56 A novel missense mutation in the galactosyltransferase-I (B4GALT7) gene in a family exhibiting facioskeletal anomalies and Ehlers-Danlos syndrome resembling the progeroid type. (PMID: 15211654) Faiyaz-Ul-Haque M . Teebi AS American journal of medical genetics. Part A 2004 2 3 23 56 Molecular ba

BACE1: GGA1 regulates signal-dependent sorting of BACE1 to recycling endosomes, which moderates A? production. Toh WH, Chia PZC, Hossain MI, Gleeson PA. Mol Biol Cell. 2018 Jan 15;29(2):191-208. doi: 10.1091/mbc.E17-05-0270. Epub 2017 Nov 15. PMID: 29142073 Increased BACE1-AS long noncoding RNA and ?-amyloid levels in heart failure. Greco S, Zaccagnini G, Fuschi P, Voellenkle C, Carrara M, Sadeghi I, Bearzi C, Maimone B, Castelvecchio S, Stellos K, Gaetano C, Menicanti L, Martelli F. Cardiovasc R

BAG3: Ilker Kudret Sariyer, Nana Merabova, Prem Kumer Patel, Tijana Knezevic, Alessandra Rosati, Maria C. Turco, Kamel KhaliliPLoS One. 2012; 7(9): e45000. Published online 2012 Sep 12. doi: 10.1371/journal.pone.0045000 PMCID: PMC3440322 Margit Fuchs, Carole Luthold, Solenn M. Guilbert, Alice Anaïs Varlet, Herman Lambert, Alexandra Jetté, Sabine Elowe, Jacques Landry, Josée N. LavoiePLoS Genet. 2015 Oct; 11(10): e1005582. Published online 2015 Oct 23 10.1371/journal.pgen.1005582PMCID: PMC4619

BAG6: Jee-Young Mock, Yue Xu, Yihong Ye, William M. Clemons, Jr.Proc Natl Acad Sci U S A. 2017 Oct 31; 114(44): 11679–11684. Published online 2017 Oct 17. doi: 10.1073/pnas.1702940114PMCID: PMC5676875 Giedre Krenciute, Shangfeng Liu, Nur Yucer, Yi Shi, Priscilla Ortiz, Qiongming Liu, Beom-Jun Kim, Abiola Ore Odejimi, Mei Leng, Jun Qin, Yi WangJ Biol Chem. 2013 Jul 12; 288(28): 20547–20557. Published online 2013 May 30. doi: 10.1074/jbc.M112.443416PMCID: PMC3711319 Aishwarya Payapilly

BAP1: Bononi A, Giorgi C, Patergnani S, et al. BAP1 regulates IP3R3-mediated Ca2+flux to mitochondria suppressing cell transformation. Nature. 2017;546(7659):549-553. doi:10.1038/nature22798.

BATF: Sang-Heon Park, Jinseol Rhee, Seul-Ki Kim, Jung-Ah Kang, Ji-Sun Kwak, Young-Ok Son, Wan-Su Choi, Sung-Gyoo Park, Jang-Soo ChunArthritis Res Ther. 2018; 20: 161. Published online 2018 Aug 2. doi: 10.1186/s13075-018-1658-0PMCID: PMC6090970 Makoto Kurachi, R. Anthony Barnitz, Nir Yosef, Pamela M. Odorizzi, Michael A. Dilorio, Madeleine E. Lemieux, Kathleen Yates, Jernej Godec, Martin G. Klatt, Aviv Regev, E. John Wherry, W. Nicholas Haining Nat Immunol. 2014 Apr; 15(4): 373–383. Published onli

BCAT1: Wang Z-Q, Faddaoui A, Bachvarova M, et al. BCAT1 expression associates with ovarian cancer progression: possible implications in altered disease metabolism. Oncotarget. 2015;6(31):31522-31543.

BCO1: Gong X, Marisiddaiah R, Rubin LP. Inhibition of pulmonary ?-carotene 15, 15′-oxygenase expression by glucocorticoid involves PPAR?. Lobaccaro J-MA, ed. PLoS ONE. 2017;12(7):e0181466. doi:10.1371/journal.pone.0181466.

BCR: Yuda J, Miyamoto T, Odawara J, et al. Persistent detection of alternatively spliced BCR-ABL variant results in a failure to achieve deep molecular response. Cancer Science. 2017;108(11):2204-2212. doi:10.1111/cas.13353.

BDNF: Brain-derived neurotrophic factor and obesity in the WAGR syndrome. (PMID: 18753648) Han JC . Yanovski JA The New England journal of medicine 2008 3 4 23 45 58 Brain-derived neurotrophic factor (BDNF) gene: no major impact on antidepressant treatment response. (PMID: 19236730) Domschke K . Baune BT The international journal of neuropsychopharmacology 2010 3 23 45 58 The effect of depression, BDNF gene val66met polymorphism and gender on serum BDNF levels. (PMID: 19589373) Ozan E . Akarsu N Variations in FKBP5 and BDNF genes are suggestively associated with depression in a Swedish population-based cohort. (PMID: 20226536) Lavebratt C ... Forsell Y Journal of affective disorders 2010 3 39 The brain-derived neurotrophic factor Val66Met polymorphism, hippocampal volume, and cognitive function in geriatric depression. (PMID: 20220593) Benjamin S ... Taylor WD The American journal of geriatric psychiatry : official journal of the American Association for Geriatric Psychiatry 2010 3 39 Assessment of peripheral biomarkers potentially involved in episodic and chronic migraine: a case-control study with a focus on NGF, BDNF, VEGF, and PGE2. (PMID: 34991456) Mozafarihashjin M ... Martami F The journal of headache and pain 2022 3 Female-specific effect of the BDNF gene on Alzheimer's disease. (PMID: 28202203) Li GD ... Yao YG Neurobiology of aging 2017 3 Activity-dependent release of endogenous BDNF from mossy fibers evokes a TRPC3 current and Ca2+ elevations in CA3 pyramidal neurons. (PMID: 20220070) Li Y ... Pozzo-Miller L Journal of neurophysiology 2010 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 Association of COMT, BDNF and 5-HTT functional polymorphisms with personality characteristics. (PMID: 34856753) Tommasi M ... Gatta V Frontiers in bioscience (Landmark edition) 2021 3 Genetic polymorphisms for BDNF, COMT, and APOE do not affect gait or ankle motor control in chronic stroke: A preliminary cross-sectional study. (PMID: 32378476) Aljuhni R ... Madhavan S Topics in stroke rehabilitation 2021 Polyacrylamide gel-based microarray: a novel method applied to the association Study between the polymorphisms of BDNF gene and autism. (PMID: 20201430) Cheng L ... Lu Z Journal of biomedical nanotechnology 2009 3 39 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 Experiencing community and domestic violence is associated with epigenetic changes in DNA methylation of BDNF and CLPX in adolescents. (PMID: 31059136) Serpeloni F ... Elbert T Psychophysiology 2020 3 Association between categorization of emotionally-charged and neutral visual scenes and parameters of event-related potentials in carriers of different COMT, HTR2A, BDNF gene genotypes. (PMID: 32983417) Vorobyeva EV ... Stoletniy AS F1000Research 2020 3 Catechol-O-Methyltransferase Gene Polymorphisms and the Risk of Chemotherapy-Induced Prospective Memory Impairment in Breast Cancer Patients with Varying Tumor Hormonal Receptor Expression. (PMID: 32985495) Li W ... Shen L Medical science monitor : international medical journal of experimental and clinical research 2020

BHMT: Rui Y-N, Xu Z, Chen Z, Zhang S. The GST-BHMT assay reveals a distinct mechanism underlying Proteínasome inhibition-induced macroautophagy in mammalian cells. Autophagy. 2015;11(5):812-832. doi:10.1080/15548627.2015.1034402.

BICC1: Benjamin Rothé, Lucia Leal-Esteban, Florian Bernet, Séverine Urfer, Nicholas Doerr, Thomas Weimbs, Justyna Iwaszkiewicz, Daniel B. ConstamMol Cell Biol. 2015 Oct; 35(19): 3339–3353. Prepublished online 2015 Jul 27. Published online 2015 Sep 4. doi: 10.1128/MCB.00341-15PMCID: PMC4561730 Benjamin Rothé, Catherine N. Leettola, Lucia Leal-Esteban, Duilio Cascio, Simon Fortier, Manuela Isenschmid, James U. Bowie, Daniel B. ConstamStructure. 2018 Feb 6; 26(2): 209–224.e6. Published online 2017 De

BICD1: Leal-Esteban LC, Rothé B, Fortier S, Isenschmid M, Constam DB. Role of Bicaudal C1 in renal gluconeogenesis and its novel interaction with the CTLH complex. Barsh GS, ed. PLoS Genetics. 2018;14(7):e1007487. doi:10.1371/journal.pgen.1007487.

BIN1: Mutations in amphiphysin 2 (BIN1) disrupt interaction with dynamin 2 and cause autosomal recessive centronuclear myopathy. (PMID: 17676042) Nicot AS ... Laporte J Nature genetics 2007 2 3 4 21 70 A Roma founder BIN1 mutation causes a novel phenotype of centronuclear myopathy with rigid spine. (PMID: 29950440) Cabrera-Serrano M ... Paradas C Neurology 2018 3 4 70 Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. (PMID: 25260562) Böhm J ... Laporte J Brain : a journal of neurology 2014 3 4 70 Phenotype of a patient with recessive centronuclear myopathy and a novel BIN1 mutation. (PMID: 20142620) Claeys KG ... Stojkovic T Neurology 2010 3 4 21 The SH3 binding motif of HCV [corrected] NS5A protein interacts with Bin1 and is important for apoptosis and infectivity. (PMID: 16530520) Nanda SK ... Liang TJ Gastroenterology 2006

BMPR1B: Katja Stange, Julie Désir, Naseebullah Kakar, Thomas D. Mueller, Birgit S. Budde, Christopher T. Gordon, Denise Horn, Petra Seemann, Guntram BorckOrphanet J Rare Dis. 2015; 10: 84. Published online 2015 Jun 24. doi: 10.1186/s13023-015-0299-5PMCID: PMC4482310 Cherry Yin-Yi Chang, Yi Chen, Ming-Tsung Lai, Hui-Wen Chang, Jack Cheng, Carmen Chan, Chih-Mei Chen, Shan-Chih Lee, Ying-Ju Lin, Lei Wan, Pei-Wen Tsai, Su-Han Yang, Ching Chung, Jim Jinn-Chyuan Sheu, Fuu-Jen TsaiPLoS One. 2013.

BMPR2: Myung-Jin Kim, Seon Young Park, Hae Ryung Chang, Eun Young Jung, Anudari Munkhjargal, Jong-Seok Lim, Myeong-Sok Lee, Yonghwan KimBMB Rep. 2017 Jun; 50(6): 308–317. Published online 2017 Jun 30. doi: 10.5483/BMBRep.2017.50.6.059PMCID: PMC5498141 Adam Andruska, Edda SpiekerkoetterInt J Mol Sci. 2018 Sep; 19(9): 2499. Published online 2018 Aug 24. doi: 10.3390/ijms19092499PMCID: PMC6165502 Alexi Crosby, Elaine Soon, Frances M. Jones, Mark R. Southwood, Leila Haghighat, Mark R. Toshner.

BNC2: Laura Cesaratto, Eleonora Grisard, Michela Coan, Luigi Zandonà, Elena De Mattia, Elena Poletto, Erika Cecchin, Fabio Puglisi, Vincenzo Canzonieri, Maria Teresa Mucignat, Antonella Zucchetto, Gabriele Stocco, Alfonso Colombatti, Milena S Nicoloso, Riccardo SpizzoCell Death Dis. 2016 Sep; 7(9): e2374. Published online 2016 Sep 22. doi: 10.1038/cddis.2016.278Correction in: Cell Death Dis. 2016 Dec; 7(12): e2526.PMCID: PMC5059877Egon Urgard, Anu Reigo, Eva Reinmaa, Ana Rebane, Andres MetspaluCa

BPESC1: Wu YY, Briollais L. Mixed-effects models for joint modeling of sequence data in longitudinal studies. BMC Proceedings. 2014;8(Suppl 1):S92. doi:10.1186/1753-6561-8-S1-S92.

BRAF: Andrea Marranci, Zhijie Jiang, Marianna Vitiello, Elena Guzzolino, Laura Comelli, Samanta Sarti, Simone Lubrano, Cinzia Franchin, Ileabett Echevarría-Vargas, Andrea Tuccoli, Alberto Mercatanti, Monica Evangelista, Paolo Sportoletti, Giorgio Cozza, Ettore Luzi, Enrico Capobianco, Jessie Villanueva, Giorgio Arrigoni, Giovanni Signore, Silvia Rocchiccioli, Letizia Pitto, Nicholas Tsinoremas, Laura PolisenoMol Cancer. 2017; 16: 85. Published online 2017 Apr 28. doi: 10.1186/s12943-017-0645-4PMC

BRAP: Shoji S, Hanada K, Ohsawa N, Shirouzu M. Central catalytic domain of BRAP (RNF52) recognizes the types of ubiquitin chains and utilizes oligo-ubiquitin for ubiquitylation. Biochemical Journal. 2017;474(18):3207-3226. doi:10.1042/BCJ20161104.

BRCA2: Fatemeh Karami, Parvin MehdipourBiomed Res Int. 2013; 2013: 928562. Published online 2013 Nov 7. doi: 10.1155/2013/928562PMCID: PMC3838820 Hermela Shimelis, Romy L.S. Mesman, Catharina Von Nicolai, Asa Ehlen, Lucia Guidugli, Charlotte Martin, Fabienne M.G.R. Calléja, Huong Meeks, Emily Hallberg, Jamie Hinton, Jenna Lilyquist, Chunling Hu, Cora M. Aalfs, Kristiina Aittomäki, Irene Andrulis, Hoda Anton-Culver, Volker Arndt, Matthias W. Beckmann, Javier Benitez, Natalia V. Bogdanova.

BSCL2: Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. (PMID: 14981520) Windpassinger C . Wagner K Nature genetics 2004 2 3 4 23 45 58 Seipin deficiency alters fatty acid Delta9 desaturation and lipid droplet formation in Berardinelli-Seip congenital lipodystrophy. (PMID: 19278620) Boutet E . Magré J Biochimie 2009 3 4 23 58 The human lipodystrophy gene BSCL2/seipin may be essential for normal adipocyte differentiation. (PMID: 18

BST1: Crystallographic studies on human BST-1/CD157 with ADP-ribosyl cyclase and NAD glycohydrolase activities. (PMID: 11866528) Yamamoto-Katayama S … Morikawa K Journal of molecular biology 2002 3 4 23 BST-1, a surface molecule of bone marrow stromal cell lines that facilitates pre-B-cell growth. (PMID: 8202488) Kaisho T … Hirano T Proceedings of the National Academy of Sciences of the United States of America 1994 2 3 4 Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. (PMID: 21292315) International Parkinson Disease Genomics Consortium … Wood NW Lancet (London, England) 2011 3 41 Common genetic variation in the HLA region is associated with late-onset sporadic Parkinson's disease. (PMID: 20711177) Hamza TH … Payami H Nature genetics 2010 3 41 Analysis of GWAS-linked loci in Parkinson disease reaffirms PARK16 as a susceptibility locus. (PMID: 20697102) Tan EK … Liu JJ Neurology 2010

BTD: Yonggang Xie, Xiaosu Li, Xian Zhang, Shaolin Mei, Hongyu Li, Andreacarola Urso, Sijun ZhueLife. 2014; 3: e03596. Published online 2014 Oct 6. doi: 10.7554/eLife.03596PMCID: PMC4221738Vera S. Hunnekuhl, Michael AkamEvoDevo. 2017; 8: 18. Published online 2017 Oct 23. doi: 10.1186/s13227-017-0082-xPMCID: PMC5654096 Chompunut Lumsangkul, Yang-Kwang Fan, Shen-Chang Chang, Jyh-Cherng Ju, Hsin-I. Chiang PLoS One. 2018; 13(5): e0196973. Published online 2018 May 9. doi: 10.1371/journal.pone.0196973

BTNL2: Xiang Tong, Yao Ma, Xundong Niu, Zhipeng Yan, Sitong Liu, Bo Peng, Shifeng Peng, Hong FanMedicine (Baltimore) 2016 Jul; 95(30): e4325. Published online 2016 Jul 29. doi: 10.1097/MD.0000000000004325PMCID: PMC5265849Krishanthi S. Subramaniam, Emily Spaulding, Emil Ivan, Eugene Mutimura, Ryung S. Kim, Xikui Liu, Chen Dong, Catherine M. Feintuch, Xingxing Zhang, Kathryn Anastos, Gregoire Lauvau, Johanna P. DailyJ Infect Dis. 2015 Oct 15; 212(8): 1322–1331. Published online 2015 Apr 15. doi: 10.

C11ORF30: Marenholz I, Grosche S, Kalb B, et al. Genome-wide association study identifies the SERPINB gene cluster as a susceptibility locus for food allergy. Nature Communications. 2017;8:1056. doi:10.1038/s41467-017-01220-0.

C11ORF49: Rachel B Keller, Thao T Tran, Shawna M Pyott, Melanie G Pepin, Ravi Savarirayan, George McGillivray, UW Center For Mendelian Genomics, Deborah A Nickerson, Michael J Bamshad, Peter H ByersGenet Med. 2018 Apr; 20(4): 411–419. Published online 2017 Aug 17. doi: 10.1038/gim.2017.115PMCID: PMC5816725Todd A Johnson, Yoshihito Niimura, Hiroshi Tanaka, Yusuke Nakamura, Tatsuhiko TsunodaGenome Biol. 2011; 12(3): R21. Published online 2011 Mar 11. doi: 10.1186/gb-2011-12-3-r21PMCID: PMC3129671C.-L.

C1ORF106: Mohanan V, Nakata T, Desch AN, et al. C1orf106 is a colitis risk gene that regulates stability of epithelial adherens junctions. Science (New York, NY). 2018;359(6380):1161-1166. doi:10.1126/science.aan0814.

C1ORF127: Liu Y-Y, Chen H-Y, Zhang M-L, Tian D, Li S, Lee J-Y. Loss of fragile histidine triad and amplification of 1p36.22 and 11p15.5 in primary gastric adenocarcinomas. World Journal of Gastroenterology: WJG. 2012;18(33):4522-4532. doi:10.3748/wjg.v18.i33.4522.

C1QTNF6: The secreted protein discovery initiative (SPDI), a large-scale effort to identify novel human secreted and transmembrane proteins: a bioinformatics assessment. (PMID: 12975309) Clark HF ... Gray A Genome research 2003 2 3 4 Novel associations for hypothyroidism include known autoimmune risk loci. (PMID: 22493691) Eriksson N ... Do CB PloS one 2012 3 39 Genetic susceptibility to type 2 diabetes is associated with reduced prostate cancer risk. (PMID: 20203524) Pierce BL ... Ahsan H Human heredity 2010 3 39 Variant of TYR and autoimmunity susceptibility loci in generalized vitiligo. (PMID: 20410501) Jin Y ... Spritz RA The New England journal of medicine 2010 3 39 Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. (PMID: 19430480) Barrett JC ... Type 1 Diabetes Genetics Consortium Nature genetics 2009

C2: Yun S-H, Sim E-H, Han S-H, et al. In vitro and in vivo anti-leukemic effects of cladoloside C2 are mediated by activation of Fas/ceramide synthase 6/p38 kinase/c-Jun NH2-terminal kinase/caspase-8. Oncotarget. 2018;9(1):495-511. doi:10.18632/oncotarget.230

C2CD4C: Han J, Perez JT, Chen C, et al. Genome-wide CRISPR/Cas9 Screen Identifies Host Factors Essential for Influenza Virus Replication. Cell reports. 2018;23(2):596-607. doi:10.1016/j.celrep.2018.03.045.

C3: Elvington M, Liszewski MK, Bertram P, Kulkarni HS, Atkinson JP. A C3(H20) recycling pathway is a component of the intracellular complement system. The Journal of Clinical Investigation. 2017;127(3):970-981. doi:10.1172/JCI89412.

C8ORF34: Hagit N Baris, Wai-Man Chan, Caroline Andrews, Doron M Behar, Diana J Donovan, Cynthia C Morton, Judith Ranells, Tuya Pal, Azra H Ligon, Elizabeth C EngleClin Case Rep. 2013 Oct; 1(1): 30–37. Published online 2013 Sep 30. doi: 10.1002/ccr3.11PMCID: PMC3885256 Jenna C. Carlson, Jennifer Standley, Aline Petrin, John R. Shaffer, Azeez Butali, Carmen J. Buxo, Eduardo Castilla, Kaare Christensen, Frederic W-D Deleyiannis, Jacqueline T. Hecht, L. Leigh Field, Ariuntuul Garidkhuu, Lina M. Moreno

CA8: Fu ES, Erasso DM, Zhuang GZ, et al. Impact of human CA8 on thermal antinociception in relation to morphine equivalence in mice. Neuroreport. 2017;28(18):1215-1220. doi:10.1097/WNR.0000000000000872.

CACNB2: Liu A, Wang Y, Sahana G, et al. Genome-wide Association Studies for Female Fertility Traits in Chinese and Nordic Holsteins. Scientific Reports. 2017;7:8487. doi:10.1038/s41598-017-09170-9.

CADM1: Richard Hunte, Patricia Alonso, Remy Thomas, Cassandra Alexandria Bazile, Juan Carlos Ramos, Louise van der Weyden, Juan Dominguez-Bendala, Wasif Noor Khan, Noula ShembadePLoS Pathog. 2018 Apr; 14(4): e1006968. Published online 2018 Apr 26. doi: 10.1371/journal.ppat.1006968PMCID: PMC5919438 Rajeshree Pujari, Richard Hunte, Remy Thomas, Louise van der Weyden, Dan Rauch, Lee Ratner, Jennifer K. Nyborg, Juan Carlos Ramos, Yoshimi Takai, Noula ShembadePLoS Pathog. 2015 Mar; 11(3): e1004721.

CALCA: Aydin-Yüce T, Kurscheid G, Bachmann HS, et al. No Association of CALCA Polymorphisms and Aseptic Loosening after Primary Total Hip Arthroplasty. BioMed Research International. 2018;2018:3687415. doi:10.1155/2018/3687415.

CALHM1: Vingtdeux V, Chang EH, Frattini SA, et al. CALHM1 deficiency impairs cerebral neuron activity and memory flexibility in mice. Scientific Reports. 2016;6:24250. doi:10.1038/srep24250.

CALN1: Kazuhiro Kobuke, Kenji Oki, Celso E. Gomez-Sanchez, Elise P. Gomez-Sanchez, Haruya Ohno, Kiyotaka Itcho, Yoko Yoshii, Masayasu Yoneda, Noboru HattoriHypertension. Author manuscript; available in PMC 2019 Jan 1.Published in final edited form as: Hypertension. 2018 Jan; 71(1): 125–133. Published online 2017 Nov 6. doi: 10.1161/HYPERTENSIONAHA.117.10205PMCID: PMC5730498 Olivia Engmann, Benoit Labonte, Amanda Mitchell, Pavel Bashtrykov, Erin S. Calipari, Chaggai Rosenbluh, Yong-Hwee E. Loh

CAMK2A: Sébastien Küry, Geeske M. van Woerden, Thomas Besnard, Martina Proietti Onori, Xénia Latypova, Meghan C. Towne, Megan T. Cho, Trine E. Prescott, Melissa A. Ploeg, Stephan Sanders, Holly A.F. Stessman, Aurora Pujol, Ben Distel, Laurie A. Robak, Jonathan A. Bernstein, Anne-Sophie Denommé-Pichon, Gaëtan Lesca, Elizabeth A. Sellars, Jonathan Berg, Wilfrid Carré, Øyvind Løvold Busk, Bregje W.M. van Bon, Jeff L. Waugh, Matthew Deardorff, George E. Hoganson, Katherine B. Bosanko, Diana S. Johnson

CAPN10: Tomohisa Hatta, Shun-ichiro Iemura, Tomokazu Ohishi, Hiroshi Nakayama, Hiroyuki Seimiya, Takao Yasuda, Katsumi Iizuka, Mitsunori Fukuda, Jun Takeda, Tohru Natsume, Yukio HorikawaSci Rep. 2018; 8: 16756. Published online 2018 Nov 13. doi: 10.1038/s41598-018-35204-xPMCID: PMC6233169Nattachet Plengvidhya, Kanjana Chanprasert, Watip Tangjittipokin, Wanna Thongnoppakhun, Pa-thai YenchitsomanusJ Diabetes Investig. 2015 Nov; 6(6): 632–639. Published online 2015 Mar 24. doi: 10.1111/jdi.12341PMCID:

CARD14: Liu H, Bao F, Irwanto A, et al. An association study of TOLL and CARD with leprosy susceptibility in Chinese population. Human Molecular Genetics. 2013;22(21):4430-4437. doi:10.1093/hmg/ddt286.

CARD9: Zhong X, Chen B, Yang L, Yang Z. Molecular and physiological roles of the adaptor protein CARD9 in immunity. Cell Death & Disease. 2018;9(2):52. doi:10.1038/s41419-017-0084-6.

CASC17: Jackson VE, Latourelle JC, Wain LV, et al. Meta-analysis of exome array data identifies six novel genetic loci for lung function. Wellcome Open Research. 2018;3:4. doi:10.12688/wellcomeopenres.12583.3.

CASC21: Li W, Qi Y, Cui X, et al. Characteristic of HPV Integration in the Genome and Transcriptome of Cervical Cancer Tissues. BioMed Research International. 2018;2018:6242173. doi:10.1155/2018/6242173.

CASC8: Hu L, Chen S-H, Lv Q-L, et al. Clinical Significance of Long Non-Coding RNA CASC8 rs10505477 Polymorphism in Lung Cancer Susceptibility, Platinum-Based Chemotherapy Response, and Toxicity. Tchounwou PB, ed. International Journal of Environmental Research and Public Health. 2016;13(6):545. doi:10.3390/ijerph13060545.

CASZ1: Zhihui Liu, Wenling Li, Xuefei Ma, Nancy Ding, Francesco Spallotta, Eileen Southon, Lino Tessarollo, Carlo Gaetano, Yoh-suke Mukouyama, Carol J. ThieleJ Biol Chem. 2014 Oct 24; 289(43): 29801–29816. Published online 2014 Sep 4. doi: 10.1074/jbc.M114.570416 PMCID: PMC4207993 Natarajan Bhaskaran, Zhihui Liu, Senthil S. Saravanamuthu, Chunhua Yan, Ying Hu, Lijin Dong, Peggy Zelenka, Lixin Zheng, Vassili Bletsos, Rachel Harris, Brenna Harrington, Aaron Weinberg, Carol J. Thiele, Fengchun Ye

CAT: Gene polymorphisms in association with emerging cardiovascular risk markers in adult women. (PMID: 20078877) Fan AZ ... Mokdad AH BMC medical genetics 2010 3 21 39 Polymorphisms in antioxidant defence genes and susceptibility to hepatocellular carcinoma in a Moroccan population. (PMID: 19929244) Ezzikouri S ... Benjelloun S Free radical research 2010 3 21 39 [Lack of association between metabolic and antioxidant gene polymorphisms (GSTM1, GSTT1, CAT, MnSOD, GPX1) and maternal quitting of smoking in pregnancy--preliminary results]. (PMID: 20301895) Hozyasz KK ... Jagodziński PP Przeglad lekarski 2009 3 21 39 [Association of genetic factors with clinical peculiarities of hypertensive disease in patients with burdened familial anamnesis]. (PMID: 19254215) Minushkina LO ... Zateĭshchikov DA Kardiologiia 2009 3 21 39 Functional variants in the catalase and myeloperoxidase genes, ambient air pollution, and respiratory-related school absences: an example of epistasis in gene-environment interactions. (PMID: 19897513) Wenten M ... Gilliland FD American journal of epidemiology 2009

CAV3: Irano N, de Camargo GMF, Costa RB, et al. Genome-Wide Association Study for Indicator Traits of Sexual Precocity in Nellore Cattle. Zhang Q, ed. PLoS ONE. 2016;11(8):e0159502. doi:10.1371/journal.pone.0159502.

CBLB: Xiao Y, Tang J, Guo H, et al. Targeting CBLB as a Potential Therapeutic Approach for Disseminated Candidiasis. Nature medicine. 2016;22(8):906-914. doi:10.1038/nm.4141.

CBS: The cystathionine beta-synthase (CBS) mutation c.1224-2A>C in Central Europe: Vitamin B6 nonresponsiveness and a common ancestral haplotype. (PMID: 15365998) Linnebank M ... Koch HG Human mutation 2004 3 4 21 39 69 A pathogenic linked mutation in the catalytic core of human cystathionine beta-synthase disrupts allosteric regulation and allows kinetic characterization of a full-length dimer. (PMID: 17352495) Sen S ... Banerjee R Biochemistry 2007 3 21 24 69 Cystathionine beta-synthase T833C/844INS68 polymorphism: a family-based study on mentally retarded children. (PMID: 16375773) Dutta S ... Mukhopadhyay K Behavioral and brain functions : BBF 2005 3 21 39 69 High homocysteine and thrombosis without connective tissue disorders are associated with a novel class of cystathionine beta-synthase (CBS) mutations. (PMID: 12007221) Maclean KN ... Kraus JP Human mutation 2002 3 4 21 69 High prevalence of the I278T mutation of the human cystathionine beta-synthase detected by a novel screening application. (PMID: 11434706) Linnebank M ... Koch HG Thrombosis and haemostasis 2001 N-Terminal Acetyltransferase Naa40p Whereabouts Put into N-Terminal Proteoform Perspective. (PMID: 33916271) Jonckheere V ... Van Damme P International journal of molecular sciences 2021 3 Reprogrammed transsulfuration promotes basal-like breast tumor progression via realigning cellular cysteine persulfidation. (PMID: 34737229) Erdélyi K ... Nagy P Proceedings of the National Academy of Sciences of the United States of America 2021 3 Association of maternal dietary intakes and CBS gene polymorphisms with congenital heart disease in offspring. (PMID: 32800907) Li Y ... Qin J International journal of cardiology 2021 3 Derangement of hepatic polyamine, folate, and methionine cycle metabolism in cystathionine beta-synthase-deficient homocystinuria in the presence and absence of treatment: Possible implications for pathogenesis. (PMID: 33483253) Maclean KN ... Stabler SP Molecular genetics and metabolism 2021 3 Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. (PMID: 33223529) Mor-Shaked H ... Harel T European journal of human genetics : EJHG 2021 Systems analysis of RhoGEF and RhoGAP regulatory proteins reveals spatially organized RAC1 signalling from integrin adhesions. (PMID: 32203420) Müller PM ... Rocks O Nature cell biology 2020 3 Wnt regulation: exploring Axin-Disheveled interactions and defining mechanisms by which the SCF E3 ubiquitin ligase is recruited to the destruction complex. (PMID: 32129710) Schaefer KN ... Peifer M Molecular biology of the cell 2020 The midbody interactome reveals unexpected roles for PP1 phosphatases in cytokinesis. (PMID: 31586073) Capalbo L ... D'Avino PP Nature communications 2019 3 Gain of Additional BIRC3 Protein Functions through 3'-UTR-Mediated Protein Complex Formation. (PMID: 30948266) Lee SH ... Mayr C Molecular cell 2019 3 Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. (PMID: 31515488) Fragoza R ... Yu H Nature communications 2019 3 PLEKHA4/kramer Attenuates Dishevelled Ubiquitination to Modulate Wnt and Planar Cell Polarity Signaling. (PMID: 31091453) Shami Shah A ... Baskin JM Cell reports 2019 3 WWP2 ubiquitylates RNA polymerase II for DNA-PK-dependent transcription arrest and repair at DNA breaks. (PMID: 31048545) Caron P ... van Attikum H Genes & development 2019

CCDC180: Prediction of the coding sequences of unidentified human genes. XVII. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. (PMID: 10819331) Nagase T ... Ohara O DNA research : an international journal for rapid publication of reports on genes and genomes 2000 2 3 4 The full-ORF clone resource of the German cDNA Consortium. (PMID: 17974005) Bechtel S ... Schupp I BMC genomics 2007 3 4 DNA sequence and analysis of human chromosome 9. (PMID: 15164053) Humphray SJ ... Dunham I Nature 2004 3 4 The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). (PMID: 15489334) Gerhard DS ... MGC Project Team Genome research 2004 3 4 Identification of Coiled-Coil Domain-Containing Protein 180 and Leucine-Rich Repeat-Containing Protein 4 as Potential Immunohistochemical Markers for Liposarcoma Based on Proteomic Analysis Using Formalin-Fixed, Paraffin-Embedded Tissue. (PMID: 30790560) Aoyama T ... Sawada N The American journal of pathology 2019

CCDC33: Paula L. Hyland, Neal D. Freedman, Nan Hu, Ze-Zhong Tang, Lemin Wang, Chaoyu Wang, Ti Ding, Jin-Hu Fan, You-Lin Qiao, Asieh Golozar, William Wheeler, Kai Yu, Jeff Yuenger, Laurie Burdett, Stephen J. Chanock, Sanford M. Dawsey, Margaret A. Tucker, Alisa M. Goldstein, Christian C. Abnet, Philip R. TaylorCarcinogenesis. 2013 May; 34(5): 1062–1068. Published online 2013 Jan 28. doi: 10.1093/carcin/bgt030PMCID: PMC3643422 Arnon Mazza, Irit Gat-Viks, Hesso Farhan, Roded Sharan Algorithms Mol Biol

CCDC77: Maria Barandalla, Hui Shi, Hui Xiao, Silvia Colleoni, Cesare Galli, Pietro Lio, Matthew Trotter, Giovanna LazzariStem Cell Res Ther. 2017; 8: 160. Published online 2017 Jul 5. doi: 10.1186/s13287-017-0602-6PMCID: PMC5497375 Lei Wei, Song Liu, Jeffrey Conroy, Jianmin Wang, Antonios Papanicolau-Sengos, Sean T. Glenn, Mitsuko Murakami, Lu Liu, Qiang Hu, Jacob Conroy, Kiersten Marie Miles, David E. Nowak, Biao Liu, Maochun Qin, Wiam Bshara, Angela R. Omilian, Karen Head, Michael Bianchi.

CCHCR1: Tervaniemi MH, Katayama S, Skoog T, et al. Intracellular signalling pathways and cytoskeletal functions converge on the psoriasis candidate gene CCHCR1 expressed at P-bodies and centrosomes. BMC Genomics. 2018;19:432. doi:10.1186/s12864-018-4810-y.

CCL2: Yaeli Lebel-Haziv, Tsipi Meshel, Gali Soria, Adva Yeheskel, Elad Mamon, Adit Ben-BaruchNeoplasia. 2014 Sep; 16(9): 723–740. Published online 2014 Sep 20. doi: 10.1016/j.neo.2014.08.004PMCID: PMC4234876Nicole Lavender, Jinming Yang, Sheau-Chiann Chen, Jiqing Sai, C. Andrew Johnson, Philip Owens, Gregory D. Ayers, Ann RichmondBMC Cancer. 2017; 17: 88. Published online 2017 Jan 31. doi: 10.1186/s12885-017-3074-2PMCID: PMC5286656Vincent R. Parillaud, Guillaume Lornet, Yann Monnet, Anne-Laure Pr

CCR7: Kobayashi D, Endo M, Ochi H, Hojo H, Miyasaka M, Hayasaka H. Regulation of CCR7-dependent cell migration through CCR7 homodimer formation. Scientific Reports. 2017;7:8536. doi:10.1038/s41598-017-09113-4.

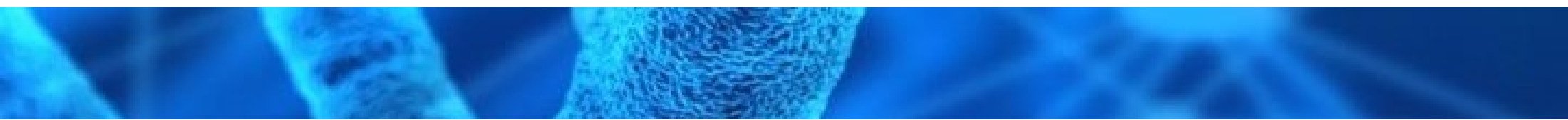
CD2AP: CD2AP mutations are associated with sporadic nephrotic syndrome and focal segmental glomerulosclerosis (FSGS). (PMID: 19131354) Gigante M . Gesualdo L Nephrology, dialysis, transplantation : official publication of the European Dialysis and Transplant Association - European Renal Association 2009 3 23 45 58 CD2AP and Cbl-3/Cbl-c constitute a critical checkpoint in the regulation of ret signal transduction. (PMID: 18753381) Tsui CC . Pierchala BA The Journal of neuroscience : the official jo

CD38: A missense mutation in the CD38 gene, a novel factor for insulin secretion: association with Type II diabetes mellitus in Japanese subjects and evidence of abnormal function when expressed in vitro. (PMID: 9754820) Yagui K ... Kanatsuka A Diabetologia 1998 3 4 23 41 Human gene encoding CD38 (ADP-ribosyl cyclase/cyclic ADP-ribose hydrolase): organization, nucleotide sequence and alternative splicing. (PMID: 9074508) Nata K ... Okamoto H Gene 1997 2 3 4 23 Isolation of a cDNA encoding the human CD38 (T10) molecule, a cell surface glycoprotein with an unusual discontinuous pattern of expression during lymphocyte differentiation. (PMID: 2319135) Jackson DG ... Bell JI Journal of immunology (Baltimore, Md. : 1950) 1990 2 3 4 23 CD38 gene polymorphisms contribute to genetic susceptibility to B-cell chronic lymphocytic leukemia: evidence from two case-control studies in Polish Caucasians. (PMID: 19240243) Jamroziak K ... Robak T Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 23 41 CD38 gene polymorphism and chronic lymphocytic leukemia: a role in transformation to Richter syndrome? (PMID: 18424664) Aydin S ... Deaglio S Blood 2008

CD46: Mutations in alternative pathway complement proteins in American patients with atypical hemolytic uremic syndrome. (PMID: 20513133) Maga TK . Smith RJ Human mutation 2010 3 4 45 58 Genetics of HUS: the impact of MCP, CFH, and IF mutations on clinical presentation, response to treatment, and outcome. (PMID: 16621965) Caprioli J . International Registry of Recurrent and Familial HUS/TTP Blood 2006 3 4 45 58 Localization of regions in CD46 that interact with adenovirus. (PMID: 15919905) Gaggar

CD58: Sable R, Durek T, Taneja V, et al. Constrained Cyclic Peptides as Immunomodulatory Inhibitors of the CD2:CD58 Protein–Protein Interaction. ACS chemical biology. 2016;11(8):2366-2374. doi:10.1021/acschembio.6b00486.

CD86: Levy R, Rotfogel Z, Hillman D, et al. Superantigens hyperinduce inflammatory cytokines by enhancing the B7-2/CD28 costimulatory receptor interaction. Proceedings of the National Academy of Sciences of the United States of America. 2016;113(42):E6437-E6446



CDCA3: Drug target discovery by gene expression analysis: cell cycle genes. (PMID: 12188893) Walker MG Current cancer drug targets 2001 Large-scale sequencing in human chromosome 12p13: experimental and computational gene structure determination. (PMID: 9074930) Ansari-Lari MA ... Gibbs RA Genome research 1997 2 3 4 Identification of type 2 diabetes-associated combination of SNPs using support vector machine. (PMID: 20416077) Ban HJ ... Park KJ BMC genetics 2010 3 40 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004 3 4 Comprehensive interactome profiling of the human Hsp70 network highlights functional differentiation of J domains. (PMID: 33957083) Piette BL ... Taipale M Molecular

CDH1: Van der Post RS, Vogelaar IP, Carneiro F, et al. Hereditary diffuse gastric cancer: updated clinical guidelines with an emphasis on germline CDH1mutation carriers. Journal of Medical Genetics. 2015;52(6):361-374. doi:10.1136/jmedgenet-2015-103094.

CDH13: Abigail C. Killen, Melissa Barber, Joshua J. W. Paulin, Barbara Ranscht, John G. Parnavelas, William D. AndrewsBrain Struct Funct. 2017; 222(8): 3567–3585. Published online 2017 Apr 6. doi: 10.1007/s00429-017-1418-yPMCID: PMC5676827Andrea Forero, Olga Rivero, Sina Wäldchen, Hsing-Ping Ku, Dominik P. Kiser, Yvonne Gärtner, Laura S. Pennington, Jonas Waider, Patricia Gaspar, Charline Jansch, Frank Edenhofer, Thérèse J. Resink, Robert Blum, Markus Sauer, Klaus-Peter LeschFront Cell Neurosci. 2

CDHR3: Kelly Watters, Ann C. PalmenbergPLoS Pathog. 2018 Dec; 14(12): e1007477. Published online 2018 Dec 10. doi: 10.1371/journal.ppat.1007477PMCID: PMC6301718Yury A. Bochkov, Kelly Watters, Shamaila Ashraf, Theodor F. Griggs, Mark K. Devries, Daniel J. Jackson, Ann C. Palmenberg, James E. GernProc Natl Acad Sci U S A. 2015 Apr 28; 112(17): 5485–5490. Published online 2015 Apr 6. doi: 10.1073/pnas.1421178112PMCID: PMC4418890 Klaus Bønnelykke, Amaziah T. Coleman, Michael D. Evans, Jonathan Thorsen

CDK4: Tarrado-Castellarnau M, de Atauri P, Tarragó-Celada J, et al. De novo MYC addiction as an adaptive response of cancer cells to CDK4/6 inhibition. Molecular Systems Biology. 2017;13(10):940. doi:10.15252/msb.20167321.

CDKAL1: Palmer CJ, Bruckner RJ, Paulo JA, et al. Cdkal1, a type 2 diabetes susceptibility gene, regulates mitochondrial function in adipose tissue. Molecular Metabolism. 2017;6(10):1212-1225. doi:10.1016/j.molmet.2017.07.013.

CDKN1A: Kreis N-N, Friemel A, Zimmer B, et al. Mitotic p21Cip1/CDKN1A is regulated by cyclin-dependent kinase 1 phosphorylation. Oncotarget. 2016;7(31):50215-50228. doi:10.18632/oncotarget.10330.

CDKN1B: Peschel I, Podmirseg SR, Taschler M, et al. FLT3 and FLT3-ITD phosphorylate and inactivate the cyclin-dependent kinase inhibitor p27Kip1 in acute myeloid leukemia. Haematologica. 2017;102(8):1378-1389. doi:10.3324/haematol.2016.160101.

CDKN2A: Gan Y, Ma W, Wang X, et al. Coordinated transcription of ANRIL and P16genes is silenced by P16 DNA methylation . Chinese Journal of Cancer Research. 2018;30(1):93-103. doi:10.21147/j.issn.1000-9604.2018.01.10.

CDKN2A/B: Kong Y, Sharma RB, Nwosu BU, Alonso LC. Islet biology, the CDKN2A/B locus and type 2 diabetes risk. Diabetologia. 2016;59(8):1579-1593. doi:10.1007/s00125-016-3967-7.

CDKN2B-AS1: ?ehime Gülsün Temel, Mahmut Çerkez ErgörenAnatol J Cardiol. 2019 Jan; 21(1): 31–38. Published online 2018 Dec 7. doi: 10.14744/AnatolJCardiol.2018.90907PMCID: PMC6382903Louis R. Pasquale, Stephanie J. Loomis, Jae H. Kang, Brian L. Yaspan, Wael Abdrabou, Donald L. Budenz, Teresa C. Chen, Elizabeth DelBono, David S. Friedman, Douglas Gaasterland, Terry Gaasterland, Cynthia L. Grosskreutz, Richard K. Lee, Paul R. Lichter, Yutao Liu, Catherine A. McCarty, Sayoko E. Moroi, Lana M. Olson.

CELF2: Sandya Ajith, Matthew R. Gazzara, Brian S. Cole, Ganesh Shankarling, Nicole M. Martinez, Michael J. Mallory, Kristen W. LynchRNA Biol. 2016 Jun; 13(6): 569–581. Published online 2016 Apr 20. doi: 10.1080/15476286.2016.1176663PMCID: PMC4962813 Michael J. Mallory, Samuel J. Allon, Jinsong Qiu, Matthew R. Gazzara, Iulia Tapescu, Nicole M. Martinez, Xiang-Dong Fu, Kristen W. LynchProc Natl Acad Sci U S A. 2015 Apr 28; 112(17): E2139–E2148. Published online 2015 Apr 13. doi: 10.1073/pnas.1423695

CELSR2: Chen Z, Lei Y, Cao X, et al. Genetic analysis of Wnt/PCP genes in neural tube defects. BMC Medical Genomics. 2018;11:38. doi:10.1186/s12920-018-0355-9.

CETP: Deguchi H, Banerjee Y, Elias DJ, Griffin JH. Elevated CETP Lipid Transfer Activity is Associated with the Risk of Venous Thromboembolism. Journal of Atherosclerosis and Thrombosis. 2016;23(10):1159-1167. doi:10.5551/jat.32201.

CFB: Wolfram G Brenner, Jan Erik Leuendorf, Anne Cortleven, Laetitia B B Martin, Hubert Schaller, Thomas SchmüllingJ Exp Bot. 2017 May 17; 68(11): 2769–2785. Published online 2017 May 12. doi: 10.1093/jxb/erx146PMCID: PMC5853388 Gloriane Schnabolk, Beth Coughlin, Kusumam Joseph, Kannan Kunchithapautham, Mausumi Bandyopadhyay, Elizabeth C. O'Quinn, Tamara Nowling, Bärbel RohrerInvest Ophthalmol Vis Sci. 2015 Mar; 56(3): 1850–1863. Published online 2015 Mar 17. doi: 10.1167/iovs.14-15910

CFH: Kerr H, Wong E, Makou E, et al. Disease-linked mutations in factor H reveal pivotal role of cofactor activity in self-surface–selective regulation of complement activation. The Journal of Biological Chemistry. 2017;292(32):13345-13360. doi:10.1074/jbc.M11

CHAT: Chen E, Lallai V, Sherafat Y, et al. Altered Baseline and Nicotine-Mediated Behavioral and Cholinergic Profiles in ChAT-Cre Mouse Lines. The Journal of Neuroscience. 2018;38(9):2177-2188. doi:10.1523/JNEUROSCI.1433-17.2018.

CHEK2: Luo L, Gao W, Wang J, et al. Study on the Mechanism of Cell Cycle Checkpoint Kinase 2 (CHEK2) Gene Dysfunction in Chemotherapeutic Drug Resistance of Triple Negative Breast Cancer Cells. Medical Science Monitor?: International Medical Journal of Experimen

CHRDL1: Pei Y, Zhang Y, Lei Y, Wu D, Ma T, Liu X. Hypermethylation of the CHRDL1 promoter induces proliferation and metastasis by activating Akt and Erk in gastric cancer. Oncotarget. 2017;8(14):23155-23166. doi:10.18632/oncotarget.15513.

CHRM2: Sakata K, Overacre AE. Promoter IV-BDNF deficiency disturbs cholinergic gene expression of CHRNA5, CHRM2, and CHRM5: effects of drug and environmental treatments. Journal of neurochemistry. 2017;143(1):49-64. doi:10.1111/jnc.14129.

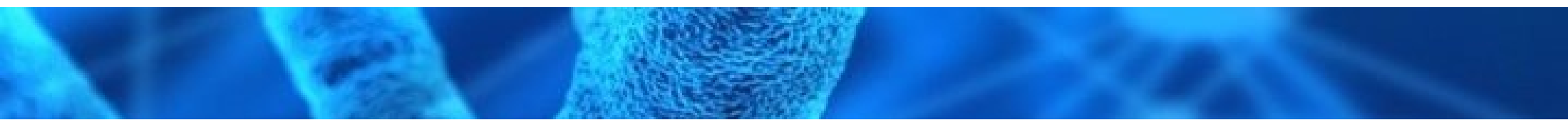
CHST12: Sasha Z. Prisco, Jeremy W. Prokop, Allison B. Sarkis, Nan Cher Yeo, Matthew J. Hoffman, Colin C. Hansen, Howard J. Jacob, Michael J. Flister, Jozef LazarHypertension. Author manuscript; available in PMC 2015 Oct 1.Published in final edited form as: Hypertension. 2014 Oct; 64(4): 883–890. Published online 2014 Jul 7. doi: 10.1161/HYPERTENSIONAHA.114.03550PMCID: PMC4162822Sasha Z. Prisco, Jessica R. C. Priestley, Brian D. Weinberg, Anthony R. Prisco, Matthew J. Hoffman, Howard J. Jacob.

CHUK: Chuk MK, Mulugeta Y, Roth-Cline M, Mehrotra N, Reaman GH. Enrolling adolescents in disease/target-appropriate adult oncology clinical trials of investigational agents. Clinical cancer research?: an official journal of the American Association for Cancer R

CLCN6: Yamamoto T, Shimojima K, Sangu N, et al. Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. Ishii R, ed. PLoS ONE. 2015;10(3):e0118946. doi:10.1371/journal.pone.0118946.

CLEC16A: Redmann V, Lamb CA, Hwang S, et al. Clec16a is Critical for Autolysosome Function and Purkinje Cell Survival. Scientific Reports. 2016;6:23326. doi:10.1038/srep23326.

CLEC7A: Kalia N, Kaur M, Sharma S, Singh J. A Comprehensive in Silico Analysis of Regulatory SNPs of Human CLEC7A Gene and Its Validation as Genotypic and Phenotypic Disease Marker in Recurrent Vulvovaginal Infections. Frontiers in Cellular and Infection Microbiology. 2018;8:65. doi:10.3389/fcimb.2018.00065.



CLOCK: Molecular cloning and characterization of the human CLOCK gene: expression in the suprachiasmatic nuclei. (PMID: 10198158) Steeves TD ... Takahashi JS Genomics 1999 2 3 4 21 Genetic variants in human CLOCK associate with total energy intake and cytokine sleep factors in overweight subjects (GOLDN population). (PMID: 19888304) Garaulet M ... Ordovas JM European journal of human genetics : EJHG 2010 3 21 39 Differential association of circadian genes with mood disorders: CRY1 and NPAS2 are associated with unipolar major depression and CLOCK and VIP with bipolar disorder. (PMID: 20072116) Soria V ... Urretavizcaya M Neuropsychopharmacology : official publication of the American College of Neuropsychopharmacology 2010 3 21 39 CLOCK gene is implicated in weight reduction in obese patients participating in a dietary programme based on the Mediterranean diet. (PMID: 20065968) Garaulet M ... Ordovas JM International journal of obesity (2005) 2010 3 21 39 Testing the circadian gene hypothesis in prostate cancer: a population-based case-control study. (PMID: 19934327) Zhu Y ... Stanford JL Cancer research 2009 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 For whom the circadian clock ticks? Investigation of PERIOD and CLOCK gene variants in bipolar disorder. (PMID: 34112033) Yegin Z ... Koc H Chronobiology international 2021 3 Daily Rhythm of Fractal Cardiac Dynamics Links to Weight Loss Resistance: Interaction with CLOCK 3111T/C Genetic Variant. (PMID: 34371977) Yang HW ... Hu K Nutrients 2021 3 Stabilization of heterochromatin by CLOCK promotes stem cell rejuvenation and cartilage regeneration. (PMID: 32737416) Liang C ... Liu GH Cell research 2021 3 Variants in clock genes could be associated with lower risk of type 2 diabetes in an elderly Greek population. (PMID: 34674804) Tsekmekidou X ... Kotsa K Maturitas 2021

CLPTM1L: Puskás LG, Mán I, Szebeni G, Tiszlavicz L, Tsai S, James MA. Novel Anti-CRR9/CLPTM1L Antibodies with Antitumorigenic Activity Inhibit Cell Surface Accumulation, PI3K Interaction, and Survival Signaling. Molecular cancer therapeutics. 2016;15(5):985-997. d

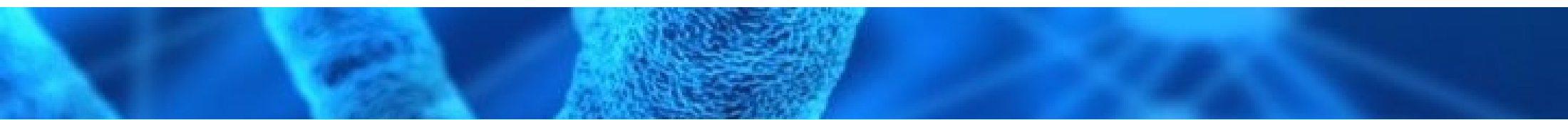
CLSTN2: Tatiana V Lipina, Tuhina Prasad, Daisaku Yokomaku, Lin Luo, Steven A Connor, Hiroshi Kawabe, Yu Tian Wang, Nils Brose, John C Roder, Ann Marie CraigNeuropsychopharmacology. 2016 Feb; 41(3): 802–810. Prepublished online 2015 Jul 14. Published online 2015 Aug 12. doi: 10.1038/npp.2015.206 PMCID: PMC4707826 Gemma de Ramon Francàs, Tobias Alther, Esther T. StoeckliFront Neuroanat. 2017; 11: 76. Published online 2017 Aug 30. doi: 10.3389/fnana.2017.00076 PMCID: PMC5582071 Leslie K. Jacobsen.

CLU: Pseudoexfoliation and Alzheimer's associated CLU risk variant, rs2279590, lies within an enhancer element and regulates CLU, EPHX2 and PTK2B gene expression. Padhy B, Hayat B, Nanda GG, Mohanty PP, Alone DP. Hum Mol Genet. 2017 Nov 15;26(22):4519-4529. doi: 10.1093/hmg/ddx329. PMID: 28973302

CNTNAP2: Zhuoyang Lu, M. V. V. V. Sekhar Reddy, Jianfang Liu, Ana Kalichava, Jiankang Liu, Lei Zhang, Fang Chen, Yun Wang, Luis Marcelo F. Holthauzen, Mark A. White, Suchithra Seshadrinathan, Xiaoying Zhong, Gang Ren, Gabby RudenkoJ Biol Chem. 2016 Nov 11; 291(46): 24133–24147. Published online 2016 Sep 12. doi: 10.1074/jbc.M116.748236 PMCID: PMC5104938 Ruoqi Gao, Nicolas H. Piguel, Alexandria E. Melendez-Zaidi, Maria Dolores Martin-de-Saavedra, Sehyoun Yoon, Marc P. Forrest, Kristoffer Myczek.

COL11A1: Jia D, Liu Z, Deng N, et al. A COL11A1-correlated pan-cancer gene signature of activated fibroblasts for the prioritization of therapeutic targets. Cancer letters. 2016;382(2):203-214. doi:10.1016/j.canlet.2016.09.001.

COL11A2: Dominant and recessive forms of fibrochondrogenesis resulting from mutations at a second locus, COL11A2. (PMID: 22246659) Tompson SW ... Cohn DH American journal of medical genetics. Part A 2012 2 3 4 70 Mutation of COL11A2 causes autosomal recessive non-syndromic hearing loss at the DFNB53 locus. (PMID: 16033917) Chen W ... Smith RJ Journal of medical genetics 2005 3 4 21 70 Autosomal recessive disorder otospondylomegaepiphyseal dysplasia is associated with loss-of-function mutations in the COL11A2 gene. (PMID: 10677296) Melkoniemi M ... Ala-Kokko L American journal of human genetics 2000 3 4 21 70 Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13). (PMID: 10581026) McGuirt WT ... Smith RJ Nature genetics 1999 2 3 4 70 Heterozygous glycine substitution in the COL11A2 gene in the original patient with the Weissenbacher-Zweymüller syndrome demonstrates its identity with heterozygous OSMED (nonocular Stickler syndrome). (PMID: 9805126) Pihlajamaa T ... Ala-Kokko L American journal of medical genetics 1998



COL17A1: Xiaoyan Yan, Chuanbao Zhang, Tingyu Liang, Fan Yang, Haoyuan Wang, Fan Wu, Wen Wang, Zheng Wang, Wen Cheng, Jiangnan Xu, Tao Jiang, Jing Chen, Yaozhong DingOncotarget. 2017 Oct 17; 8(49): 85794–85803. Published online 2017 Aug 24. doi: 10.18632/oncotarget.20526PMCID: PMC5689647Varalee Yodsurang, Chizu Tanikawa, Takafumi Miyamoto, Paulisally Hau Yi Lo, Makoto Hirata, Koichi MatsudaOncotarget. 2017 Aug 22; 8(34): 55790–55803. Published online 2017 Jun 9. doi: 10.18632/oncotarget.18433PMCID: P

COL1A1: Zhang Z, Wang Y, Zhang J, Zhong J, Yang R. COL1A1 promotes metastasis in colorectal cancer by regulating the WNT/PCP pathway. Molecular Medicine Reports. 2018;17(4):5037-5042. doi:10.3892/mmr.2018.8533.

COL1A2: Schäfer G, Hitchcock JK, Shaw TM, Katz AA, Parker MI. A Novel Role of Annexin A2 in Human Type I Collagen Gene Expression. Journal of Cellular Biochemistry. 2015;116(3):408-417. doi:10.1002/jcb.24989.

COL27A1: Belbin GM, Odgis J, Sorokin EP, et al. Genetic identification of a common collagen disease in Puerto Ricans via identity-by-descent mapping in a health system. McCarthy M, ed. eLife. 2017;6:e25060. doi:10.7554/eLife.25060.

COL2A1: Sakagami N, Ono W, Ono N. Diverse contribution of Col2a1-expressing cells to the craniofacial skeletal cell lineages. Orthodontics & craniofacial research. 2017;20(Suppl 1):44-49. doi:10.1111/ocr.12168.

COL3A1: Wang X-Q, Tang Z-X, Yu D, et al. Epithelial but not stromal expression of collagen alpha-1(III) is a diagnostic and prognostic indicator of colorectal carcinoma. Oncotarget. 2016;7(8):8823-8838. doi:10.18632/oncotarget.6815.

COL4A1: Debbie S. Kuo, Cassandre Labelle-Dumais, Mao Mao, Marion Jeanne, William B. Kauffman, Jennifer Allen, Jack Favor, Douglas B. GouldHum Mol Genet. 2014 Apr 1; 23(7): 1709–1722. Published online 2013 Nov 7. doi: 10.1093/hmg/ddt560 PMCID: PMC3943517 Frances E. Jones, Matthew A. Bailey, Lydia S. Murray, Yinhui Lu, Sarah McNeilly, Ursula Schlötzer-Schrehardt, Rachel Lennon, Yoshikazu Sado, David G. Brownstein, John J. Mullins, Karl E. Kadler, Tom Van AgtmaelDis Model Mech. 2016 Feb 1; 9(2): 165–1

COL4A3: Choquet H, Thai KK, Yin J, et al. A large multi-ethnic genome-wide association study identifies novel genetic loci for intraocular pressure. Nature Communications. 2017;8:2108. doi:10.1038/s41467-017-01913-6.

COL4A4: Mutations in the COL4A4 gene in thin basement membrane disease. (PMID: 12631110) Buzza M ... Savige J Kidney international 2003 3 4 21 39 Determination of the genomic structure of the COL4A4 gene and of novel mutations causing autosomal recessive Alport syndrome. (PMID: 9792860) Boye E ... Antignac C American journal of human genetics 1998 3 4 21 70 The clinical spectrum of type IV collagen mutations. (PMID: 9195222) Lemmink HH ... Smeets HJ Human mutation 1997 3 4 21 70 Benign familial hematuria due to mutation of the type IV collagen alpha4 gene. (PMID: 8787673) Lemmink HH ... Smeets HJ The Journal of clinical investigation 1996 3 4 21 70 Identification of novel variants in the COL4A4 gene in Korean patients with thin basement membrane nephropathy. (PMID: 19675380) Baek JI ... Kim UK The Indian journal of medical research 2009

COL4A5: Efficient detection of Alport syndrome COL4A5 mutations with multiplex genomic PCR-SSCP. (PMID: 11223851) Barker DF ... Gregory MC American journal of medical genetics 2001 3 4 21 70 Mutational analysis of COL4A5 gene in Korean Alport syndrome. (PMID: 10684360) Cheong HI ... Choi Y Pediatric nephrology (Berlin, Germany) 2000 3 4 21 70 Spectrum of COL4A5 mutations in Finnish Alport syndrome patients. (PMID: 10862091) Martin P ... Tryggvason K Human mutation 2000 3 4 21 70 Detection of mutations in COL4A5 in patients with Alport syndrome. (PMID: 10094548) Plant KE ... Flinter FA Human mutation 1999 3 4 21 70 Detection of mutations in the COL4A5 gene in over 90% of male patients with X-linked Alport's syndrome by RT-PCR and direct sequencing. (PMID: 10561141) Inoue Y ... Yoshikawa N American journal of kidney diseases : the official journal of the National Kidney Foundation 1999

COL4A6: Deletion of the paired alpha 5(IV) and alpha 6(IV) collagen genes in inherited smooth muscle tumors. (PMID: 8356449) Zhou J ... Reeders ST Science (New York, N.Y.) 1993 2 3 4 21 Novel form of X-linked nonsyndromic hearing loss with cochlear malformation caused by a mutation in the type IV collagen gene COL4A6. (PMID: 23714752) Rost S ... Kunstmann E European journal of human genetics : EJHG 2014 3 4 70 Structure of the human type IV collagen COL4A6 gene, which is mutated in Alport syndrome-associated leiomyomatosis. (PMID: 8661006) Zhang X ... Tryggvason K Genomics 1996 3 4 21 Clonal overgrowth of esophageal smooth muscle cells in diffuse leiomyomatosis-Alport syndrome caused by partial deletion in COL4A5 and COL4A6 genes. (PMID: 20951201) Oohashi T ... Ninomiya Y Matrix biology : journal of the International Society for Matrix Biology 2011 3 70 Variation at the NFATC2 locus increases the risk of thiazolidinedione-induced edema in the Diabetes REduction Assessment with ramipril and rosiglitazone Medication (DREAM) study. (PMID: 20628086) Bailey SD ... DREAM investigators Diabetes care 2010

COL5A1: DeNigris J, Yao Q, Birk EK, Birk DE. Altered dermal fibroblast behavior in a collagen V haploinsufficient murine model of classic Ehlers-Danlos syndrome. Connective tissue research. 2016;57(1):1-9. doi:10.3109/03008207.2015.1081901.

COL5A2: Mutations of the alpha2(V) chain of type V collagen impair matrix assembly and produce ehlers-danlos syndrome type I. (PMID: 9425231) Michalickova K ... Cole WG Human molecular genetics 1998 3 4 21 70 A single base mutation in COL5A2 causes Ehlers-Danlos syndrome type II. (PMID: 9783710) Richards AJ ... Burrows NP Journal of medical genetics 1998 3 4 21 70 Sequence analysis of the COL5A2 gene in patients with spontaneous cervical artery dissections. (PMID: 11940702) Grond-Ginsbach C ... Brandt T Neurology 2002 3 4 21 Genomic organization of the human COL3A1 and COL5A2 genes: COL5A2 has evolved differently than the other minor fibrillar collagen genes. (PMID: 11566270) Välkkilä M ... Ala-Kokko L Matrix biology : journal of the International Society for Matrix Biology 2001 3 4 21 Clinical and molecular characterization of 40 patients with classic Ehlers-Danlos syndrome: identification of 18 COL5A1 and 2 COL5A2 novel mutations. (PMID: 23587214) Ritelli M ... Colombi M Orphanet journal of rare diseases 2013

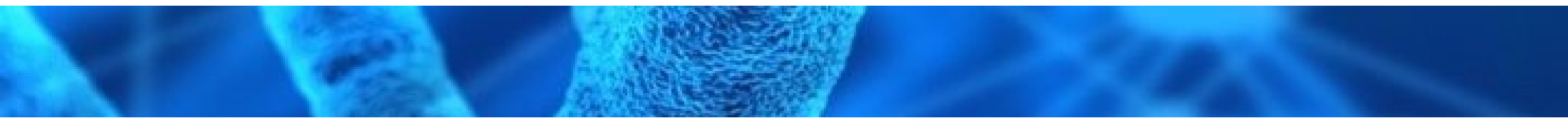
COL7A1: Tao Y, Gross N, Fan X, et al. Identification of novel enriched recurrent chimeric COL7A1-UCN2 in human laryngeal cancer samples using deep sequencing. BMC Cancer. 2018;18:248. doi:10.1186/s12885-018-4161-8.

COL8A1: Corominas J, Colijn JM, Geerlings MJ, et al. Whole-Exome Sequencing in Age-Related Macular Degeneration Identifies Rare Variants in COL8A1, a Component of Bruch's Membrane. Ophthalmology. 2018;125(9):1433-1443. doi:10.1016/j.ophtha.2018.03.040.

COLEC12: Lin-Li Chang, Wen-Hung Hsu, Mou-Chieh Kao, Chih-Chung Chou, Chung-Cheng Lin, Chung-Jung Liu, Bi-Chuang Weng, Fu-Chen Kuo, Chao-Hung Kuo, Ming-Hong Lin, Chun-Jen Wang, Chun-Hung Lin, Deng-Chyang Wu, Shau-Ku HuangSci Rep. 2018; 8: 3821. Published online 2018 Feb 28. doi: 10.1038/s41598-018-20957-2PMCID: PMC5830506 Eugene Lin, Po-Hsiu Kuo, Yu-Li Liu, Albert C. Yang, Shih-Jen Tsai Oncotarget. 2017 Nov 7; 8(55): 93349–93359. Published online 2017 Sep 16. doi: 10.18632/oncotarget.

COMT: Is there an association of genetic polymorphisms of the catechol-O-methyltransferase gene (rs165656 and rs174675) and the 5-hydroxytryptamine receptor 2A gene (rs4941573 and rs6313) with sleep bruxism in individuals with obstructive sleep apnea? (PMID: 34808513) Duarte J ... De Luca Canto G Archives of oral biology 2022 3 COMTVal158Met polymorphism is associated with ecstasy (MDMA)-induced psychotic symptoms in the Turkish population. (PMID: 35017287) Aytac HM ... Pehlivan S Neurosciences (Riyadh, Saudi Arabia) 2022 Warriors versus worriers: the role of COMT gene variants. Stein DJ1, Newman TK, Savitz J, Ramesar R. COMT genotype increases risk for bipolar I disorder and influences neurocognitive performance. Burdick KE1, Funke B, Goldberg JF, Bates JA, Jaeger J, Kucherlapati R, Malhotra AK. Genetic variants in COMT and neurocognitive impairment in families of patients with schizophrenia. Liao SY1, Lin SH, Liu CM, Hsieh MH, Hwang TJ, Liu SK, Guo SC, Hwu HG, Chen WJ. SARS-CoV-2-host proteome interactions for antiviral drug discovery. (PMID: 34709727) Liu X ... Varjosalo M Molecular systems biology 2021 3 Multilevel proteomics reveals host perturbations by SARS-CoV-2 and SARS-CoV. (PMID: 33845483) Stukalov A ... Pichlmair A Nature 2021 3 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 USP11 mediates repair of DNA-protein cross-links by deubiquitinating SPRTN metalloprotease. (PMID: 33567341) Perry M ... Ghosal G The Journal of biological chemistry 2021 3 BPA modulates the WDR5/TET2 complex to regulate ERβ expression in eutopic endometrium and drives the development of endometriosis. (PMID: 33022573) Xue W ... Yuanzhen Z Environmental pollution (Barking, Essex : 1987) 2021 3 Epistatic effect of Ankyrin repeat and kinase domain containing 1 - Dopamine receptor D2 and catechol-o-methyltransferase single nucleotide polymorphisms on the risk for hazardous use of alcohol in Lithuanian population. (PMID: 32889058) Kaminskaite M ... Bunevicius A Gene 2021 3 Effects of traumatic life events, cognitive biases and variation in dopaminergic genes on psychosis proneness. (PMID: 31889426) Kotowicz K ... Misiak B Early intervention in psychiatry

2021 3 Catechol-O-methyltransferase and dopamine receptor D4 gene variants: Possible association with substance abuse in Bangladeshi male. (PMID: 33544778) Sonia JA ... Kabir Y PloS one 2021 3 No links between genetic variation and developing theory of mind: A preregistered replication attempt of candidate gene studies. (PMID: 33666309) Opitz T ... Sodian B Developmental science 2021 3 Brain predictive coding processes are associated to COMT gene Val158Met polymorphism. (PMID: 33716157) Bonetti L ... Brattico E NeuroImage 2021 3 Genome-wide association study identifies RNF123 locus as associated with chronic widespread musculoskeletal pain. (PMID: 33926923) Rahman MS ... Williams FM Annals of the rheumatic diseases 2021 3 Association of COMT, BDNF and 5-HTT functional polymorphisms with personality characteristics. (PMID: 34856753) Tommasi M ... Gatta V Frontiers in bioscience (Landmark edition) 2021 3 Suppressor effect of catechol-O-methyltransferase gene in prostate cancer. (PMID: 34587154) Hashimoto Y ... Tanaka Y PloS one 2021 3 Risks of Macrosomia Associated with Catechol-O-Methyltransferase Genotypes and Genetic-Epigenetic Interactions among Children with and without Gestational Diabetes Exposure. (PMID: 33826421) Aguayo L ... Hou L Childhood obesity (Print) 2021 3 Impact of COMT val158met on tDCS-induced cognitive enhancement in older adults. (PMID: 33359367) Hayek D ... Flöel A Behavioural brain research 2021 3 COMT rs4818, pain sensitivity and duration, and alveolar bone grafting of oral clefts. (PMID: 32989615) Silva EMVM ... Vieira AR Oral and maxillofacial surgery 2021 3 COMT Genotype and Efficacy of Propranolol for TMD Pain: A Randomized Trial. (PMID: 33030089) Slade GD ... Tchivileva IE Journal of dental research 2021 3 Polymorphisms in COMT, ADRB2 and HTR1A genes are associated with temporomandibular disorders in individuals with other arthralgias. (PMID: 31264537) Bonato LL ... Casado PL Cranio : the journal of craniomandibular practice 2021 3 The Associations between COMT and MAO-B Genetic Variants with Negative Symptoms in Patients with Schizophrenia. (PMID: 34287249) Madzarac Z ... Pivac N Current issues in molecular biology 2021 3 Mutations and structural variations in Catechol-O-methyltransferase gene of patients exhibiting chronic persistent surgical pain. (PMID: 33478750) Dharaniprasad G ... Bhan V Revista espanola de anestesiologia y reanimacion 2021 3 Catechol-O-methyltransferase (COMT) polymorphism predicts rapid gait speed changes in healthy older adults. (PMID: 34231207) Sprague BN ... Rosano C Journal of the American Geriatrics Society 2021 3 Influence of Catechol-O-Methyltransferase Gene Polymorphism on the Correlation between Alexithymia and Hypervigilance to Pain. (PMID: 34948872) Ikarashi H ... Onishi H International journal of environmental research and public health 2021 3 Genetic polymorphisms for BDNF, COMT, and APOE do not affect gait or ankle motor control in chronic stroke: A preliminary cross-sectional study. (PMID: 32378476) Aljuhni R ... Madhavan S Topics in stroke rehabilitation 2021 3 Influence of COMT polymorphism in cognitive performance on dementia in community-dwelling elderly Mexican (SADEM study). (PMID: 33900525) Juárez-Cedillo T ... Vargas-Alarcón G Metabolic brain disease 2021 3 Enhancing dopamine tone modulates global and local cortical perfusion as a function of COMT val158met genotype. (PMID: 34390874) Furman DJ ... D'Esposito M NeuroImage 2021 3 Fear of pain moderates the relationship between self-reported fatigue and methionine allele of catechol-O-methyltransferase gene in patients with fibromyalgia. (PMID: 33909692) Ferrera D ... Gómez-Esquer F PloS one 2021 3 Association of COMT Polymorphisms with Multiple Physical Activity-Related Injuries among University Students in China. (PMID: 34682575) Chen S ... Li L International journal of environmental research and public health 2021 3 Association Between the COMT Val158Met Polymorphism and Antipsychotic Efficacy in Schizophrenia: An Updated Meta-Analysis. (PMID: 33100205) Ma J ... Qin S Current neuropharmacology 2021 3 The roles of borderline personality disorder symptoms and dispositional capability for suicide in suicidal ideation and suicide attempts: Examination of the COMT Val158Met polymorphism. (PMID: 34051678) Tull MT ... Gratz KL Psychiatry research 2021 3 Catechol-O-Methyltransferase Genotype, Frailty, and Gait Speed in a Biracial Cohort of Older Adults. (PMID: 33043988) Mance S ... Rosano C Journal of the American Geriatrics Society 2021 3 Aberrant Brain Signal Variability and COMT Genotype in Chronic TMD Patients. (PMID: 33622085) Lim M ... DaSilva AF Journal of dental research 2021 3 Association between polymorphisms in catechol-O-methyl transferase, opioid receptor Mu 1 and serotonin receptor genes with postoperative pain following root canal treatment. (PMID: 33559241) Karataş E ... Akbıyık N International endodontic journal 2021 3 The association between selected genetic variants and individual differences in experimental pain. (PMID: 33108341) Lie MU ... Nilsen KB Scandinavian journal of pain 2021 3 Sex moderates the association between the COMT Val158Met single-nucleotide polymorphism and disorderliness facet of novelty seeking. (PMID: 33352204) Scacchia P ... De Pascalis V Neuroscience research 2021 3 Computational analysis of deleterious single nucleotide polymorphisms in catechol O-Methyltransferase conferring risk to post-traumatic stress disorder. (PMID: 33865170) Chitrala KN ... Nagarkatti M Journal of psychiatric research 2021 3 The interactive effect of genetic polymorphisms of IL-10 and COMT on cognitive function in schizophrenia. (PMID: 33127070) Wang J ... Zhang X Journal of psychiatric research 2021 3 Inhibitory Control Mediates the Associations Between Parenting Practices and Depressive Symptoms in Adolescents: The Moderating Role of Catechol-O-Methyltransferase (COMT) Gene. (PMID: 34259955) Cao Y ... Zhang W Journal of youth and adolescence 2021 3 Investigating interactive effects of worry and the catechol-o-methyltransferase gene (COMT) on working memory performance. (PMID: 34173216) Louis CC ... Moser JS Cognitive, affective & behavioral neuroscience 2021 3 Do oral contraceptives affect young women's



memory? Dopamine-dependent working memory is influenced by COMT genotype, but not time of pill ingestion. (PMID: 34111174) Gravelsins L ... Einstein G PloS one 2021 3 SLC6A3 (DAT1) as a Novel Candidate Biomarker Gene for Suicidal Behavior. (PMID: 34199792) Rafikova E ... Vasilyev V Genes 2021 3 The COMT gene rs4680 polymorphism moderates the relationship between adult ADHD symptoms and executive dysfunction. (PMID: 33450700) Liu J ... Gong J Asian journal of psychiatry 2021 3 Influence of ADRB1, ADRB2, and COMT Genetic Polymorphisms on Postoperative Outcomes of Patients Undergoing Cardiac Valve Surgery. (PMID: 33451866) Dai S ... Wang E Clinical therapeutics 2021 3 The Effects of Childhood Maltreatment on Non-Suicidal Self-Injury in Male Adolescents: The Moderating Roles of the Monoamine Oxidase A (MAOA) Gene and the Catechol-O-Methyltransferase (COMT) Gene. (PMID: 33807669) Gao Y ... Wang H International journal of environmental research and public health 2021 3 Association study of Catechol-o-methyltransferase and Alpha-1-adrenergic receptor gene polymorphisms with multiple phenotypes of heroin use disorder. (PMID: 33577997) Deji C ... Wei S Neuroscience letters 2021 Comparative host-coronavirus protein interaction networks reveal pan-viral disease mechanisms. (PMID: 33060197) Gordon DE ... Krogan NJ Science (New York, N.Y.) 2020 3 RIG-I regulates myeloid differentiation by promoting TRIM25-mediated ISGylation. (PMID: 32513696) Wu SF ... Chen S Proceedings of the National Academy of Sciences of the United States of America 2020 3 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 Interactome Mapping Provides a Network of Neurodegenerative Disease Proteins and Uncovers Widespread Protein Aggregation in Affected Brains. (PMID: 32814053) Haenig C ... Wanker EE Cell reports 2020 3 Extensive rewiring of the EGFR network in colorectal cancer cells expressing transforming levels of KRASG13D. (PMID: 31980649) Kennedy SA ... Kolch W Nature communications 2020 3 A SARS-CoV-2 protein interaction map reveals targets for drug repurposing. (PMID: 32353859) Gordon DE ... Krogan NJ Nature 2020 3 Synthetic Lethal and Resistance Interactions with BET Bromodomain Inhibitors in Triple-Negative Breast Cancer. (PMID: 32416067) Shu S ... Polyak K Molecular cell 2020 3 The influence of dopaminergic polymorphisms on selective stopping. (PMID: 31863847) Rincón-Pérez I ... Albert J Behavioural brain research 2020 3 The effect of rs1076560 (DRD2) and rs4680 (COMT) on tardive dyskinesia and cognition in schizophrenia subjects. (PMID: 32931693) Punchaichira TJ ... Thelma BK Psychiatric genetics 2020 3 Genetic susceptibility to parenting style: DRD2 and COMT influence creativity. (PMID: 32119983) Si S ... Zhang J NeuroImage 2020 3 Genetic Biomarkers of Panic Disorder: A Systematic Review. (PMID: 33158196) Tretiakov A ... Klimov E Genes 2020 3 Association of COMT Gene Polymorphisms with Response to Methadone Maintenance Treatment Among Chinese Opioid-Dependent Patients. (PMID: 32407152) Duan L ... Li X Genetic testing and molecular biomarkers 2020 3 COMT val158met genotype alters the effects of methamphetamine dependence on dopamine and dopamine-related executive function: preliminary findings. (PMID: 32739643) Saloner R ... Ellis RJ Psychiatry research 2020 3 Interaction between catechol-O-methyltransferase polymorphism and childhood trauma in suicidal ideation of patients with post-traumatic stress disorder. (PMID: 32618128) Kwon A ... Lee SH Brain and behavior 2020 3 Association between categorization of emotionally-charged and neutral visual scenes and parameters of event-related potentials in carriers of different COMT, HTR2A, BDNF gene genotypes. (PMID: 32983417) Vorobyeva EV ... Stoletniy AS F1000Research 2020 3 Effects of childhood trauma experience and COMT Val158Met polymorphism on brain connectivity in a multimodal MRI study. (PMID: 32997444) Tian T ... Zhu W Brain and behavior 2020 3 Evaluation of COMT (rs4680), CNR2 (rs2501432), CNR2 (rs2229579), UCP2 (rs659366), and IL-17 (rs763780) gene variants in synthetic cannabinoid use disorder patients. (PMID: 32662357) Pehlivan S ... Cetinay Aydin P Journal of addictive diseases 2020 3 The role of COMT polymorphism in modulation of prefrontal activity during verbal fluency in bipolar disorder. (PMID: 32822765) Devrimci-Ozguven H ... Baskak B Neuroscience letters 2020

COQ2: Jun-yi Zhu, Yulong Fu, Adam Richman, Zhanzheng Zhao, Patricio E. Ray, Zhe HanJ Am Soc Nephrol. 2017 Sep; 28(9): 2607–2617. Published online 2017 Apr 20. doi: 10.1681/ASN.2016060626PMCID: PMC5576924Diran Herebian, Annette Seibt, Sander H. J. Smits, Richard J. Rodenburg, Ertan Mayatepek, Felix DistelmaierAnn Clin Transl Neurol. 2017 Dec; 4(12): 902–908. Published online 2017 Oct 17. doi: 10.1002/acn3.486PMCID: PMC5740244Kotaro Ogaki, Shinsuke Fujioka, Michael G Heckman, Sruti Rayaprolu, Alexa

COQ4: He CH, Xie LX, Allan CM, Tran UC, Clarke CF. Coenzyme Q supplementation or over-expression of the yeast Coq8 putative kinase stabilizes multi-subunit Coq polypeptide complexes in yeast coq null mutants. Biochimica et biophysica acta. 2014;1841(4):630-644. doi:10.1016/j.bbalip.2013.12.017.

COQ6: Ozeir M, Pelosi L, Ismail A, Mellot-Draznieks C, Fontecave M, Pierrel F. Coq6 Is Responsible for the C4-deamination Reaction in Coenzyme Q Biosynthesis in Saccharomyces cerevisiae. The Journal of Biological Chemistry. 2015;290(40):24140-24151. doi:10.1074/jbc.M115.675744.

COQ7: Wang Y, Smith C, Parboosingh JS, Khan A, Innes M, Hekimi S. Pathogenicity of two COQ7 mutations and responses to 2,4-dihydroxybenzoate bypass treatment. Journal of Cellular and Molecular Medicine. 2017;21(10):2329-2343. doi:10.1111/jcmm.13154.

COQ8A: Stefely JA, Licitra F, Laredj L, et al. Cerebellar Ataxia and Coenzyme Q Deficiency Through Loss of Unorthodox Kinase Activity. Molecular cell. 2016;63(4):608-620. doi:10.1016/j.molcel.2016.06.030.

COQ9: Luna-Sánchez M, Díaz-Casado E, Barca E, et al. The clinical heterogeneity of coenzyme Q10 deficiency results from genotypic differences in the Coq9 gene. EMBO Molecular Medicine. 2015;7(5):670-687. doi:10.15252/emmm.201404632.

CORO2A: cDNA cloning of a novel WD repeat protein mapping to the 9q22.3 chromosomal region. (PMID: 8985118) Zaphiropoulos PG . Toftgård R DNA and cell biology 1996 2 3 4 23 56 Common variants in FOXP1 are associated with generalized vitiligo. (PMID: 20526340) Jin Y . Spritz RA Nature genetics 2010 3 43 56 Coronin 2A regulates a subset of focal-adhesion-turnover events through the cofilin pathway. (PMID: 19654210) Marshall TW . Bear JE Journal of cell science 2009 3 23 56 The status, quality, and ex

CPS1: The T1405N carbamoyl phosphate synthetase polymorphism does not affect plasma arginine concentrations in preterm infants. (PMID: 20520828) Moonen RM . Villamor E PloS one 2010 3 4 23 45 58 Structural insight on the control of urea synthesis: identification of the binding site for N-acetyl-L-glutamate, the essential allosteric activator of mitochondrial carbamoyl phosphate synthetase. (PMID: 19754428) Pekkala S . Cervera J The Biochemical journal 2009 3 4 23 58 Genetic variation in the mitoc

CR1: Fonseca MI, Chu S, Pierce AL, et al. Analysis of the Putative Role of CR1 in Alzheimer's Disease: Genetic Association, Expression and Function. Zabel MD, ed. PLoS ONE. 2016;11(2):e0149792. doi:10.1371/journal.pone.0149792.

CR1L: Complement receptor 1 gene variants are associated with erythrocyte sedimentation rate. (PMID: 21700265) Kullo IJ . Chute CG American journal of human genetics 2011 3 45 58 Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. (PMID: 19060910) Sabatti C . Peltonen L Nature genetics 2009 3 45 58 A human CR1-like transcript containing sequence for a binding protein for iC4 is expressed in hematopoietic and fetal lymphoid tissue. (PMID: 14687939) Log

CRP: Sudhakar M, Silambanan S, Chandran AS, Prabhakaran AA, Ramakrishnan R. C-Reactive Protein (CRP) and Leptin Receptor in Obesity: Binding of Monomeric CRP to Leptin Receptor. Frontiers in Immunology. 2018;9:1167. doi:10.3389/fimmu.2018.01167.

CRYBB2: A Novel CRYBB2 Stopgain Mutation Causing Congenital Autosomal Dominant Cataract in a Chinese Family Yu Zhou,1,2,3 Yaru Zhai,1 Lulin Huang,1,2,3 Bo Gong,1,2,3 Jie Li,4 Fang Hao,1,2,3 Zhengzheng Wu,4 Yi Shi,1,2,3 and Yin Yang1,3,4

CRYBB3: A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts. Jiao X1, Kabir F2, Irum B2,3, Khan AO4, Wang Q1, Li D1, Khan AA3, Husnain T3, Akram J5,6, Riazuddin S3,5,6, Hejtmancik JF1, Riazuddin SA2. Different evolution rates within the lens-specific beta-crystallin gene family. Aarts HJ1, Jacobs EH, van Willigen G, Lubsen NH, Schoenmakers JG.

CRYGD: Novel mutations in CRYGD are associated with congenital cataracts in Chinese families Guoxing Yang, Zhimin Chen, Wulin Zhang, Zhiqiang Liu & Jialiang Zhao

CSMD1: Escudero-Esparza A, Bartoschek M, Gialeli C, et al. Complement inhibitor CSMD1 acts as tumor suppressor in human breast cancer. Oncotarget. 2016;7(47):76920-76933. doi:10.18632/oncotarget.12729.

CTC1: Wang Y, Chai W. Pathogenic CTC1 mutations cause global genome instabilities under replication stress. Nucleic Acids Research. 2018;46(8):3981-3992. doi:10.1093/nar/gky114.

CTLA4: Mackroth MS, Abel A, Steeg C, Schulze zur Wiesch J, Jacobs T. Acute Malaria Induces PD1+CTLA4+ Effector T Cells with Cell-Extrinsic Suppressor Function. Engwerda CR, ed. PLoS Pathogens. 2016;12(11):e1005909. doi:10.1371/journal.ppat.1005909.

CTNNA3: Bing He, Ting Li, Lei Guan, Fang-E Liu, Xue-Mei Chen, Jing Zhao, Song Lin, Zhi-Zhen Liu, Hu-Qin ZhangOncotarget. 2016 Feb 16; 7(7): 8078–8089. Published online 2016 Jan 22. doi: 10.18632/oncotarget.6978PMCID: PMC4884977Elena Bacchelli, Fabiola Ceroni, Dalila Pinto, Silvia Lomartire, Maila Giannandrea, Patrizia D'Adamo, Elena Bonora, Piero Parchi, Raffaella Tancredi, Agatino Battaglia, Elena MaestriniJ Neurodev Disord. 2014; 6(1): 17. Published online 2014 Jul 10. doi: 10.1186/1866-1955-6-17

CTNNBL1: Van Maldegem F, Maslen S, Johnson CM, et al. CTNNBL1 facilitates the association of CWC15 with CDC5L and is required to maintain the abundance of the Prp19 spliceosomal complex. Nucleic Acids Research. 2015;43(14):7058-7069. doi:10.1093/nar/gkv643.

CX3CR1: Wang J, Gan Y, Han P, et al. Ischemia-induced Neuronal Cell Death Is Mediated by Chemokine Receptor CX3CR1. Scientific Reports. 2018;8:556. doi:10.1038/s41598-017-18774-0.

CYP17A1: Petrunak EM, Rogers SA, Aubé J, Scott EE. Structural and Functional Evaluation of Clinically Relevant Inhibitors of Steroidogenic Cytochrome P450 17A1. Drug Metabolism and Disposition. 2017;45(6):635-645. doi:10.1124/dmd.117.075317.

CYP19A1: Chen Z, Wang O, Nie M, et al. Aromatase deficiency in a Chinese adult man caused by novel compound heterozygous CYP19A1 mutations: Effects of estrogen replacement therapy on the bone, lipid, liver and glucose metabolism. Molecular and cellular endocrinolo

CYP1A1: Peddireddy V, Badabagni SP, Gundimeda SD, Mamidipudi V, Penagaluru PR, Mundluru HP. Association of CYP1A1, GSTM1 and GSTT1 gene polymorphisms with risk of non-small cell lung cancer in Andhra Pradesh region of South India. European Journal of Medical Rese

CYP1B1: Banerjee A, Chakraborty S, Chakraborty A, Chakrabarti S, Ray K. Functional and Structural Analyses of CYP1B1 Variants Linked to Congenital and Adult-Onset Glaucoma to Investigate the Molecular Basis of These Diseases. Anderson MG, ed. PLoS ONE. 2016;11(5)

CYP24A1: Julia Höbaus, Samawansha Tennakoon, Petra Heffeter, Charlotte Groeschel, Abhishek Aggarwal, Doris M. Hummel, Ursula Thiem, Rodrig Marculescu, Walter Berger, Enikö KállayInt J Cancer. 2016 Jan 15; 138(2): 440–450. Published online 2015 Aug 17. doi: 10.1002/ijc.29717 PMCID: PMC4832261 Peter J. Tebben, Ravinder J. Singh, Rajiv KumarEndocr Rev. 2016 Oct; 37(5): 521–547. Published online 2016 Sep 2. doi: 10.1210/er.2016-1070 PMCID: PMC5045493 Elaine W. Tieu, Wei Li, Jianjun Chen, Tae-Kang Kim.

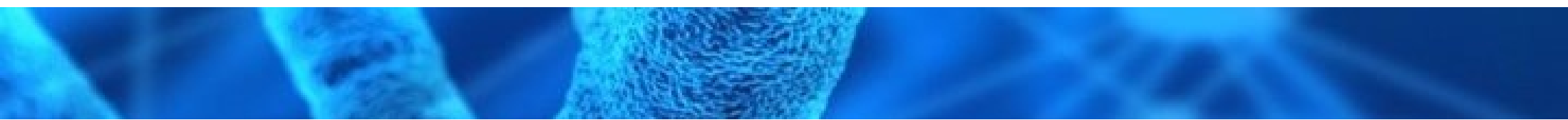
CYP2E1: W. A. García-Suástegui, L. A. Ramos-Chávez, M. Rubio-Osornio, M. Calvillo-Velasco, J. A. Atzin-Méndez, J. Guevara, D. Silva-Adaya Oxid Med Cell Longev. 2017; 2017: 4680732. Published online 2017 Jan 10. doi: 10.1155/2017/4680732 PMCID: PMC5259652 Cesar Kenaan, Erin V. Shea, Hsia-lien Lin, Haoming Zhang, Matthew J. Pratt-Hyatt, Paul F. Hollenberg Drug Metab Dispos. 2013 Jan; 41(1): 101–110. Published online 2013 Jan. doi: 10.1124/dmd.112.046094PMCID: PMC3533429 Hongming Liu, Guiyu Lou, Chong

CYP2R1: Thanapirom K, Suksawatamnuay S, Sukeepaisarnjareon W, et al. Genetic variation in the vitamin D pathway CYP2R1 gene predicts sustained HBeAg seroconversion in chronic hepatitis B patients treated with pegylated interferon: A multicenter study. Huang J-F,

CYP3A4: Liu J-E, Ren B, Tang L, et al. The independent contribution of miRNAs to the missing heritability in CYP3A4/5 functionality and the metabolism of atorvastatin. Scientific Reports. 2016;6:26544. doi:10.1038/srep26544.

CYP3A5: Yoshiyuki Shirasaka, Shu-Ying Chang, Mary F. Grubb, Chi-Chi Peng, Kenneth E. Thummel, Nina Isoherranen, A. David RodriguesDrug Metab Dispos. 2013 Aug; 41(8): 1566–1574. Published online 2013 Aug. doi: 10.1124/dmd.112.049940PMCID: PMC3716306Carrie A. Vyhlidal, Robin E. Pearce, Roger Gaedigk, Justina C. Calamia, Diana L. Shuster, Kenneth E. Thummel, J. Steven LeederDrug Metab Dispos. 2015 Aug; 43(8): 1286–1293. Published online 2015 Aug. doi: 10.1124/dmd.115.064998Correction in: Drug Metab Di

CYP4A11: Sirotina S, Ponomarenko I, Kharchenko A, et al. A Novel Polymorphism in the Promoter of the CYP4A11 Gene Is Associated with Susceptibility to Coronary Artery Disease. Disease Markers. 2018;2018:5812802. doi:10.1155/2018/5812802.



CYP4F2: CYP4F2 is a vitamin K1 oxidase: An explanation for altered warfarin dose in carriers of the V433M variant. (PMID: 19297519) McDonald MG ... Rettie AE Molecular pharmacology 2009 3 4 21 39 CYP4F2 gene V433M polymorphism is associated with ischemic stroke in the male Northern Chinese Han population. (PMID: 20227456) Deng S ... Zhiyi H Progress in neuro-psychopharmacology & biological psychiatry 2010 3 21 39 Pharmacogenetic relevance of CYP4F2 V433M polymorphism on acenocoumarol therapy. (PMID: 19270263) Pérez-Andreu V ... González-Conejero R Blood 2009 3 4 39 CYP4F2 genetic variant (rs2108622) significantly contributes to warfarin dosing variability in the Italian population. (PMID: 19207028) Borgiani P ... Novelli G Pharmacogenomics 2009 3 4 39 A genome-wide association study of acenocoumarol maintenance dosage. (PMID: 19578179) Teichert M ... Stricker BH Human molecular genetics 2009

CYP51A1: Režen T, Ogris I, Sever M, Merzel F, Golic Grdadolnik S, Rozman D. Evaluation of Selected CYP51A1 Polymorphisms in View of Interactions with Substrate and Redox Partner. Frontiers in Pharmacology. 2017;8:417. doi:10.3389/fphar.2017.00417.

DAB2IP: Bellazzo A, Di Minin G, Collavin L. Block one, unleash a hundred. Mechanisms of DAB2IP inactivation in cancer. Cell Death and Differentiation. 2017;24(1):15-25. doi:10.1038/cdd.2016.134.

DAPK1: Dayton J. Goodell, Vincent Zaegel, Steven J. Coultrap, Johannes W. Hell, K. Ulrich BayerCell Rep. Author manuscript; available in PMC 2017 Aug 9.Published in final edited form as: Cell Rep. 2017 Jun 13; 19(11): 2231–2243. doi: 10.1016/j.celrep.2017.05.068PMCID: PMC5549467Lei Pei, Shan Wang, Huijuan Jin, Linlin Bi, Na Wei, Honglin Yan, Xin Yang, Chengye Yao, Mengmeng Xu, Shu Shu, Yu Guo, Huanhuan Yan, Jianhua Wu, Hao Li, Pei Pang, Tian Tian, Qing Tian, Ling-Qiang Zhu, You Shang, Youming Lu.

DAPL1: Felix Grassmann, Ulrike Friedrich, Sascha Fauser, Tina Schick, Andrea Milenkovic, Heidi L. Schulz, Claudia N. von Strachwitz, Thomas Bettecken, Peter Lichtner, Thomas Meitinger, Nicole Arend, Armin Wolf, Christos Haritoglou, Guenther Rudolph, Usha Chakravarthy, Giuliana Silvestri, Gareth J. McKay, Sandra Freitag-Wolf, Michael Krawczak, R. Theodore Smith, John C. Merriam, Joanna E. Merriam, Rando Allikmets, Iris M. Heid, Bernhard H. F. Weber Neuromolecular Med. 2015; 17(2): 111–120.

DAT: Sorkina T, Ma S, Larsen MB, Watkins SC, Sorkin A. Small molecule induced oligomerization, clustering and clathrin-independent endocytosis of the dopamine transporter. Gether U, ed. eLife. 2018;7:e32293. doi:10.7554/eLife.32293.

DBC1: Qin B, Minter-Dykhouse K, Yu J, et al. DBC1 Functions as a Tumor Suppressor by Regulating p53 Stability. Cell reports. 2015;10(8):1324-1334. doi:10.1016/j.celrep.2015.01.066.

DBH: Huang J, Ren T, Cao S, et al. HBx-related long non-coding RNA DBH-AS1 promotes cell proliferation and survival by activating MAPK signaling in hepatocellular carcinoma. Oncotarget. 2015;6(32):33791-33804.

DDC: Yi Z, Ouyang S, Zhou C, et al. Andrographolide Inhibits Mechanical and Thermal Hyperalgesia in a Rat Model of HIV-Induced Neuropathic Pain. Frontiers in Pharmacology. 2018;9:593. doi:10.3389/fphar.2018.00593.

DDX58: Chang Y-C, Chi L-H, Chang W-M, et al. Glucose transporter 4 promotes head and neck squamous cell carcinoma metastasis through the TRIM24-DDX58 axis. Journal of Hematology & Oncology. 2017;10:11. doi:10.1186/s13045-016-0372-0.

DDX60L: Oliver Grünvogel, Katharina Esser-Nobis, Anna Reustle, Philipp Schult, Birthe Müller, Philippe Metz, Martin Trippler, Marc P. Windisch, Michael Frese, Marco Binder, Oliver Fackler, Ralf Bartenschlager, Alessia Ruggieri, Volker LohmannJ Virol. 2015 Oct 15; 89(20): 10548–10568. Published online 2015 Aug 12. doi: 10.1128/JVI.01297-15PMCID: PMC4580188 Rana Khsheibun, Tamar Paperna, Anat Volkowich, Izabella Lejbkowicz, Nili Avidan, Ariel MillerPLoS One. 2014; 9(7): e102331.

DEF8: PLEKHM1/DEF8/RAB7 complex regulates lysosome positioning and bone homeostasis. (PMID: 27777970) Fujiwara T . Zhao H JCI insight 2016 2 3 58 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T . Sugano S Nature genetics 2004 3 4 58 Architecture of the human interactome defines protein communities and disease networks. (PMID: 28514442) Huttlin EL . Harper JW Nature 2017 3 58 RNA-binding activity of TRIM25 is mediated by its PRY/SPRY domain and is

DENND5A: Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. (PMID: 27866705) Han C ... McPherson PS American journal of human genetics 2016 3 4 70 Family-wide characterization of the DENN domain Rab GDP-GTP exchange factors. (PMID: 20937701) Yoshimura S ... Barr FA The Journal of cell biology 2010 2 3 4 Prediction of the coding sequences of unidentified human genes. XIV. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. (PMID: 10470851) Kikuno R ... Ohara O DNA research : an international journal for rapid publication of reports on genes and genomes 1999 2 3 4 Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. (PMID: 27431290) Anazi S ... Alkuraya FS Molecular psychiatry 2017 4 70 The retromer coat complex coordinates endosomal sorting and dynein-mediated transport, with carrier recognition by the trans-Golgi network. (PMID: 19619496) Wassmer T ... Cullen PJ Developmental cell 2009

DERA: DERA is the human deoxyribose phosphate aldolase and is involved in stress response. (PMID: 25229427) Salleron L ... Lane L Biochimica et biophysica acta 2014 2 3 4 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010

DHFR: Identification and characterization of an inborn error of metabolism caused by dihydrofolate reductase deficiency. (PMID: 21310276) Banka S ... Newman WG American journal of human genetics 2011 3 4 70 Dihydrofolate reductase deficiency due to a homozygous DHFR mutation causes megaloblastic anemia and cerebral folate deficiency leading to severe neurologic disease. (PMID: 21310277) Cario H ... Schwarz K American journal of human genetics 2011 3 4 70 Correlations of inhibitor kinetics for Pneumocystis jirovecii and human dihydrofolate reductase with structural data for human active site mutant enzyme complexes. (PMID: 19196009) Cody V ... Rosowsky A Biochemistry 2009 3 4 21 Genetic association study of putative functional single nucleotide polymorphisms of genes in folate metabolism and spina bifida. (PMID: 19683694) Martinez CA ... Au KS American journal of obstetrics and gynecology 2009 3 21 39 A 19-base pair deletion polymorphism in dihydrofolate reductase is associated with increased unmetabolized folic acid in plasma and decreased red blood cell folate. (PMID: 19022952) Kalmbach RD ... Selhub J The Journal of nutrition 2008

DIAPH2: A human homologue of the Drosophila melanogaster diaphanous gene is disrupted in a patient with premature ovarian failure: evidence for conserved function in oogenesis and implications for human sterility. (PMID: 9497258) Bione S . Toniolo D American journal of human genetics 1998 3 4 23 58 A large-scale candidate gene association study of age at menarche and age at natural menopause. (PMID: 20734064) He C . Hunter DJ Human genetics 2010 3 45 58 The DNA sequence of the human X chromosome. (

DIRC3: Shen Z, Ren W, Bai Y, et al. DIRC3 and near NABP1 genetic polymorphisms are associated laryngeal squamous cell carcinoma patient survival. Oncotarget. 2016;7(48):79596-79604. doi:10.18632/oncotarget.12865.

DKK1: Lucia D’Amico, Sahil Mahajan, Aude-Hélène Capietto, Zhengfeng Yang, Ali Zamani, Biancamaria Ricci, David B. Bumpass, Melissa Meyer, Xinming Su, Andrea Wang-Gillam, Katherine Weilbaecher, Sheila A. Stewart, David G. DeNardo, Roberta FaccioJ Exp Med. 2016 May 2; 213(5): 827–840. doi: 10.1084/jem.20150950PMCID: PMC4854727Phillip C. Witcher, Sara E. Miner, Daniel J. Horan, Whitney A. Bullock, Kyung-Eun Lim, Kyung Shin Kang, Alison L. Adaniya, Ryan D. Ross, Gabriela G. Loots, Alexander G. Roblin

DKKL1: Yan Q, Wu X, Chen C, et al. Developmental expression and function of DKKL1/Dkkl1 in humans and mice. Reproductive Biology and Endocrinology: RB&E. 2012;10:51. doi:10.1186/1477-7827-10-51.

DLC1: Brajendra K. Tripathi, Tiera Grant, Xiaolan Qian, Ming Zhou, Philipp Mertins, Dunrui Wang, Alex G. Papageorge, Sergey G. Tarasov, Kent W. Hunter, Steven A. Carr, Douglas R. LowyJ Cell Biol. 2017 Dec 4; 216(12): 4255–4270. doi: 10.1083/jcb.201703105 PMCID: PMC5716279 Brajendra K. Tripathi, Xiaolan Qian, Philipp Mertins, Dunrui Wang, Alex G. Papageorge, Steven A. Carr, Douglas R. LowyJ Cell Biol. 2014 Dec 8; 207(5): 627–642. doi: 10.1083/jcb.201405105PMCID: PMC4259810 Choon Kiat Sim, Sun-Yee

DLEU1: Tianyou Liu, Zhiyang Han, Huanyu Li, Yuekun Zhu, Ziquan Sun, Anlong ZhuMol Cancer. 2018; 17: 118. Published online 2018 Aug 11. doi: 10.1186/s12943-018-0873-2PMCID: PMC6087004Chang Liu, Xing Tian, Jing Zhang, Lifeng JiangFront Genet. 2018; 9: 629. Published online 2018 Dec 7. doi: 10.3389/fgene.2018.00629PMCID: PMC6292861 Sanghoon Lee, Wen Luo, Tishi Shah, Changhong Yin, Timmy O’Connell, Tae-Hoon Chung, Sherrie L Perkins, Rodney R Miles, Janet Ayello, Erin Morris, Lauren Harrison.

DLG2: Claudio Reggiani, Sandra Coppens, Tayeb Sekhara, Ivan Dimov, Bruno Pichon, Nicolas Lufin, Marie-Claude Addor, Elga Fabia Belligni, Maria Cristina Digilio, Flavio Faletra, Giovanni Battista Ferrero, Marion Gerard, Bertrand Isidor, Shelagh Joss, Florence Niel-Bütschi, Maria Dolores Perrone, Florence Petit, Alessandra Renieri, Serge Romana, Alexandra Topa, Joris Robert Vermeesch, Tom Lenaerts, Georges Casimir, Marc Abramowicz, Gianluca Bontempi, Catheline Vilain, Nicolas Deconinck.

DLGAP1: GKAP, a novel synaptic protein that interacts with the guanylate kinase-like domain of the PSD-95/SAP90 family of channel clustering molecules. (PMID: 9024696) Kim E . Sheng M The Journal of cell biology 1997 2 3 4 23 56 DAP-1, a novel protein that interacts with the guanylate kinase-like domains of hDLG and PSD-95. (PMID: 9286858) Satoh K . Akiyama T Genes to cells : devoted to molecular & cellular mechanisms 1997 2 3 4 56 Variation at the NFATC2 locus increases the risk of thiazolidinedio

DMD: Ignazio Maggio, Luca Stefanucci, Josephine M. Janssen, Jin Liu, Xiaoyu Chen, Vincent Mouly, Manuel A.F.V. Gonçalves Nucleic Acids Res. 2016 Feb 18; 44(3): 1449–1470. Published online 2016 Jan 13. doi: 10.1093/nar/gkv1540 PMCID: PMC4756843 Ignazio Maggio, Jin Liu, Josephine M. Janssen, Xiaoyu Chen, Manuel A. F. V. Gonçalves Sci Rep. 2016; 6: 37051. Published online 2016 Nov 15. doi: 10.1038/srep37051 PMCID: PMC5109245 Frederique Ruf-Zamojski, Vikas Trivedi, Scott E. Fraser, Le A. TrinhPLoS O

DMRT1: Valentine A. Agbor, Shixin Tao, Ning Lei, Leslie L. HeckertBiol Reprod. 2013 Feb; 88(2): 51. Published online 2012 Dec 19. doi: 10.1095/biolreprod.112.103135PMCID: PMC3589237 Wei Sun, Han Cai, Gloria Zhang, Haiyan Zhang, Haisheng Bao, Li Wang, Jian Ye, Guoying Qian, Chutian GeSci Rep. 2017; 7: 4433. Published online 2017 Jun 30. doi: 10.1038/s41598-017-04938-5 Correction in: Sci Rep. 2018; 8: 6322.PMCID: PMC5493664 Kaitlyn A. Webster, Ursula Schach, Angel Ordaz, Jocelyn S. Steinfeld.

DNAH11: Mutations in dynein genes in patients affected by isolated non-syndromic asthenozoospermia. (PMID: 18492703) Zuccarello D . Foresta C Human reproduction (Oxford, England) 2008 3 23 45 58 Mutations in the DNAH11 (axonemal heavy chain dynein type 11) gene cause one form of situs inversus totalis and most likely primary ciliary dyskinesia. (PMID: 12142464) Bartoloni L . Antonarakis SE Proceedings of the National Academy of Sciences of the United States of America 2002 3 4 23 58 Common variatio

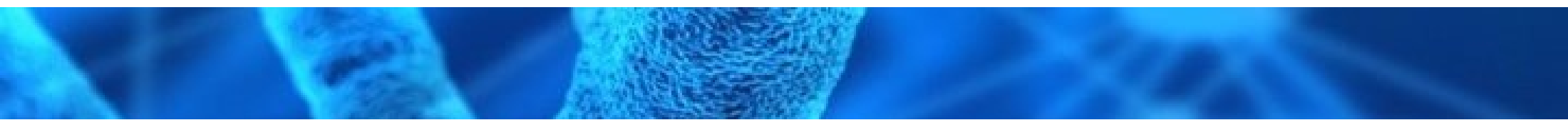
DNAJC5B: Ana Claudia Silva Braga, Bruno Moreira Carneiro, Mariana Nogueira Batista, Mônica Mayumi Akinaga, Cíntia Bittar, Paula RahalPLoS One. 2017; 12(11): e0188467. Published online 2017 Nov 28. doi: 10.1371/journal.pone.0188467PMCID: PMC5705118 Dov Shiffman, Stella Trompet, Judy Z. Louie, Charles M. Rowland, Joseph J. Catanese, Olga A. Iakoubova, Todd G. Kirchgessner, Rudi G. J. Westendorp, Anton J. M. de Craen, P. Eline Slagboom, Brendan M. Buckley, David J. Stott, Naveed Sattar, James J. Devlin

DNASE1L3: Caucasian-specific allele in non-synonymous single nucleotide polymorphisms of the gene encoding deoxyribonuclease I-like 3, potentially relevant to autoimmunity, produces an inactive enzyme. (PMID: 19559017) Ueki M … Yasuda T Clinica chimica acta; international journal of clinical chemistry 2009 3 4 23 41 cDNA cloning of human DNase gamma: chromosomal localization of its gene and enzymatic properties of recombinant protein. (PMID: 14646506) Shiokawa D … Tanuma S Apoptosis : an international journal on programmed cell death 1998 2 3 4 23 Cloning and characterization of an actin-resistant DNase I-like endonuclease secreted by macrophages. (PMID: 9714828) Baron WF … Baker KP Gene 1998 2 3 4 23 Characterization of human DNase I family endonucleases and activation of DNase gamma during apoptosis. (PMID: 11141064) Shiokawa D … Tanuma S Biochemistry 2001 3 4 23 Identification, localization, and expression of two novel human genes similar to deoxyribonuclease I. (PMID: 9205125) Rodriguez AM … Schneider MC Genomics 1997

DNER: Du J, Wang X, Zhang X, Zhang X, Jiang H. DNER modulates the length, polarity and synaptogenesis of spiral ganglion neurons via the Notch signaling pathway. Molecular Medicine Reports. 2018;17(2):2357-2365. doi:10.3892/mmr.2017.8115.

DOCK7: Shinichi Nakamuta, Yu-Ting Yang, Chia-Lin Wang, Nicholas B. Gallo, Jia-Ray Yu, Yilin Tai, Linda Van AelstJ Cell Biol. 2017 Dec 4; 216(12): 4313–4330. doi: 10.1083/jcb.201704157PMCID: PMC5716287Yu-Ting Yang, Chia-Lin Wang, Linda Van AelstNat Neurosci. 2012 Sep; 15(9): 1201–1210. Published online 2012 Jul 29. doi: 10.1038/nn.3171PMCID: PMC3431462Yilin Tai, Justyna A. Janas, Chia-Lin Wang, Linda Van AelstCell Rep. Author manuscript; available in PMC 2015 Jan 30.Published in final edited form a

DOCK8: Shiraishi A, Uruno T, Sanematsu F, et al. DOCK8 Protein Regulates Macrophage Migration through Cdc42 Protein Activation and LRAP35a Protein Interaction. The Journal of Biological Chemistry. 2017;292(6):2191-2202. doi:10.1074/jbc.M116.736306.



DPAGT1: Podralska M, Zió?kowska-Suchanek I, ?urawek M, et al. Genetic variants in ATM, H2AFX and MRE11 genes and susceptibility to breast cancer in the polish population. BMC Cancer. 2018;18:452. doi:10.1186/s12885-018-4360-3.

DPP6: Lin L, Murphy JG, Karlsson R-M, et al. DPP6 Loss Impacts Hippocampal Synaptic Development and Induces Behavioral Impairments in Recognition, Learning and Memory. Frontiers in Cellular Neuroscience. 2018;12:84. doi:10.3389/fncel.2018.00084.

DPYD: Khushman M, Patel GK, Hosein PJ, et al. Germline pharmacogenomics of DPYD*9A (c.85T>C) variant in patients with gastrointestinal malignancies treated with fluoropyrimidines. Journal of Gastrointestinal Oncology. 2018;9(3):416-424. doi:10.21037/jgo.2018.02

DRD4: Dopamine receptor D4 polymorphism predicts the effect of L-DOPA on gambling behavior. (PMID: 19914604) Eisenegger C . Fehr E Biological psychiatry 2010 Association of VNTR polymorphisms in the MAOA promoter and DRD4 exon 3 with heroin dependence in male Chinese addicts. (PMID: 20218801) Chien CC . Lung FW The world journal of biological psychiatry : the official journal of the World Federation of Societies of Biological Psychiatry 2010 3 23 45 58 Candidate gene studies of ADHD: a meta-analy

DSG2: Overmiller AM, McGuinn KP, Roberts BJ, et al. c-Src/Cav1-dependent activation of the EGFR by Dsg2. Oncotarget. 2016;7(25):37536-37555. doi:10.18632/oncotarget.7675.

DTNBP1: Zhang W, Daly KM, Liang B, et al. BDNF rescues prefrontal dysfunction elicited by pyramidal neuron-specific DTNBP1 deletion in vivo. Journal of Molecular Cell Biology. 2017;9(2):117-131. doi:10.1093/jmcb/mjw029.

E2F3: Gamper I, Burkhart DL, Bywater MJ, et al. Determination of the physiological and pathological roles of E2F3 in adult tissues. Scientific Reports. 2017;7:9932. doi:10.1038/s41598-017-09494-6.

ECT2: Lauren P. Huff, Molly J. DeCristo, Dimitri Trembath, Pei Fen Kuan, Margaret Yim, Jinsong Liu, Danielle R. Cook, C. Ryan Miller, Channing J. Der, Adrienne D. CoxGenes Cancer. 2013 Nov; 4(11-12): 460–475. doi: 10.1177/1947601913514851PMCID: PMC3877668 Verline Justilien, Syed A. Ali, Lee Jamieson, Ning Yin, Adrienne D. Cox, Channing J. Der, Nicole R. Murray, Alan P. Fields Cancer Cell. Author manuscript; available in PMC 2018 Feb 13. Published in final edited form as: Cancer Cell. 2017 Feb 13;

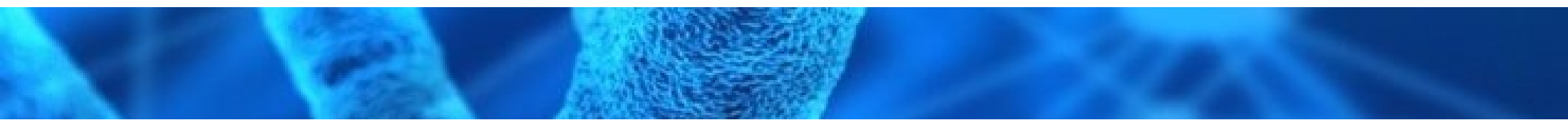
EDA2R: Zheng J, Wang J, Pouliot M, et al. Gene expression profiling in non-human primate jejunum, ileum and colon after total-body irradiation: a comparative study of segment-specific molecular and cellular responses. BMC Genomics. 2015;16:984. doi:10.1186/s1286

EDAR: Endo C, Johnson TA, Morino R, et al. Genome-wide association study in Japanese females identifies fifteen novel skin-related trait associations. Scientific Reports. 2018;8:8974. doi:10.1038/s41598-018-27145-2.

EDEM1: A dual role for EDEM1 in the processing of rod opsin. (PMID: 19934218) Kosmaoglou M . Cheetham ME Journal of cell science 2009 3 4 23 58 Role of EDEM in the release of misfolded glycoproteins from the calnexin cycle. (PMID: 12610306) Molinari M . Paganetti P Science (New York, N.Y.) 2003 2 3 4 58 EDEM2 initiates mammalian glycoprotein ERAD by catalyzing the first mannose trimming step. (PMID: 25092655) Ninagawa S . Mori K The Journal of cell biology 2014 3 4 58 EDEM1 recognition and deliver

EEPD1: Yuehan Wu, Suk-Hee Lee, Elizabeth A. Williamson, Brian L. Reinert, Ju Hwan Cho, Fen Xia, Aruna Shanker Jaiswal, Gayathri Srinivasan, Bhavita Patel, Alexis Brantley, Daohong Zhou, Lijian Shao, Rupak Pathak, Martin Hauer-Jensen, Sudha Singh, Kimi Kong, Xaiohua Wu, Hyun-Suk Kim, Timothy Beissbarth, Jochen Gaedcke, Sandeep Burma, Jac A. Nickoloff, Robert A. HromasPLoS Genet. 2015 Dec; 11(12): e1005675. Published online 2015 Dec 18. doi: 10.1371/journal.pgen.1005675 PMCID: PMC4684289Hyun-Suk Kim

EHBP1: Li Z, Schulze RJ, Weller SG, et al. A novel Rab10-EHBP1-EHD2 complex essential for the autophagic engulfment of lipid droplets. Science Advances. 2016;2(12):e1601470. doi:10.1126/sciadv.1601470.



EHF: Jing Shi, Yiping Qu, Xinru Li, Fang Sui, Demao Yao, Qi Yang, Bingyin Shi, Meiju Ji, Peng HouCell Death Dis. 2016 Oct; 7(10): e2442. Published online 2016 Oct 27. doi: 10.1038/cddis.2016.346 PMCID: PMC5134001 Sara L. Fossum, Michael J. Mutolo, Antonio Tugores, Sujana Ghosh, Scott H. Randell, Lisa C. Jones, Shih-Hsing Leir, Ann HarrisJ Biol Chem. 2017 Jun 30; 292(26): 10938–10949. Published online 2017 May 1. doi: 10.1074/jbc.M117.775304PMCID: PMC5491778 Yanyan Lv, Fang Sui, Jingjing Ma, Xiao

EIF2AK2: Xie M, Yu Y, Kang R, et al. PKM2-dependent glycolysis promotes NLRP3 and AIM2 inflammasome activation. Nature Communications. 2016;7:13280. doi:10.1038/ncomms13280.

ELAC2: Reinhard L, Sridhara S, Hällberg BM. The MRPP1/MRPP2 complex is a tRNA-maturation platform in human mitochondria. Nucleic Acids Research. 2017;45(21):12469-12480. doi:10.1093/nar/gkx902.

ELN: Expression of elastolytic cathepsins in human skin and their involvement in age-dependent elastin degradation. (PMID: 32007579) Panwar P ... Brömme D Biochimica et biophysica acta. General subjects 2020 3 Elastin in the Tumor Microenvironment. (PMID: 32845499) Wang Y ... Resnick MB Advances in experimental medicine and biology 2020 3 Elastin is a key factor of tumor development in colorectal cancer. (PMID: 32171282) Li J ... Liu G BMC cancer 2020 3 Remodeling Matrix Synthesis in a Rat Model of Aortocaval Fistula and the Cyclic Stretch: Impaction in Pulmonary Arterial Hypertension-Congenital Heart Disease. (PMID: 32630068) Chang CJ ... Lai YJ International journal of molecular sciences 2020 3 Transglutaminase-Mediated Cross-Linking of Tropoelastin to Fibrillin Stabilises the Elastin Precursor Prior to Elastic Fibre Assembly. (PMID: 32898582) Lockhart-Cairns MP ... Tarakanova A Journal of molecular biology 2020 Identification and characterization of seven novel mutations of elastin gene in a cohort of patients affected by supravalvular aortic stenosis. (PMID: 19844261) Micale L ... Merla G European journal of human genetics : EJHG 2010 3 21 70 Differential regulation of elastic fiber formation by fibulin-4 and -5. (PMID: 19570982) Choudhury R ... Kielty CM The Journal of biological chemistry 2009 3 4 21 An autosomal-recessive form of cutis laxa is due to homozygous elastin mutations, and the phenotype may be modified by a heterozygous fibulin 5 polymorphism. (PMID: 19194475) Mégarbané H ... Fischer J The Journal of investigative dermatology 2009 3 4 21 Fibrillin-1 interactions with fibulins depend on the first hybrid domain and provide an adaptor function to tropoelastin. (PMID: 17255108) El-Hallous E ... Reinhardt DP The Journal of biological chemistry 2007 3 4 21 Domains 16 and 17 of tropoelastin in elastic fibre formation. (PMID: 17037986) Wachi H ... Seyama Y The Biochemical journal

EMX2OS: Antisense transcripts at the EMX2 locus in human and mouse. (PMID: 12573261) Noonan FC . Simon TC Genomics 2003 2 3 23 58 Genome wide association study (GWAS) of Chagas cardiomyopathy in Trypanosoma cruzi seropositive subjects. (PMID: 24324551) Deng X . REDSII Chagas Study Group from the NHLBI Retrovirus Epidemiology Donor Study-II Component International PloS one 2013 3 58 lncRNAdb: a reference database for long noncoding RNAs. (PMID: 21112873) Amaral PP . Mattick JS Nucleic acids research

ENPP1: Wang H, Gonzalez-Garcia I, Traba J, et al. ATP-degrading ENPP1 is required for survival (or persistence) of long-lived plasma cells. Scientific Reports. 2017;7:17867. doi:10.1038/s41598-017-18028-z.

ENTPD7: Tordella L, Khan S, Hohmeyer A, et al. SWI/SNF regulates a transcriptional program that induces senescence to prevent liver cancer. Genes & Development. 2016;30(19):2187-2198. doi:10.1101/gad.286112.116.

EPHA2: The EPHA2 gene is associated with cataracts linked to chromosome 1p Alan Shiels,corresponding author1,2 Thomas M. Bennett,1 Harry L.S. Knopf,1 Giovanni Maraini,3 Anren Li,4 Xiaodong Jiao,4 and J. Fielding Hejtmancik4 EPHA2 MUTATIONS CONTRIBUTE TO CONGENITAL CATARACT THROUGH DIVERSE MECHANISMS Alpana Dave,1 Sarah Martin,1 Raman Kumar,2,3 Jamie E. Craig,1 Kathryn P. Burdon,1 and Shiwani Sharmacorresponding author1

EPHX1: Václavíková R, Hughes DJ, Souček P. Microsomal Epoxide Hydrolase 1 (EPHX1): Gene, Structure, Function, and Role in Human Disease. Gene. 2015;571(1):1-8. doi:10.1016/j.gene.2015.07.071.

ERCC2: Li L, Bai H, Yang J, Cao D, Shen K. Genome-wide DNA copy number analysis in clonally expanded human ovarian cancer cells with distinct invasive/migratory capacities. Oncotarget. 2017;8(9):15136-15148. doi:10.18632/oncotarget.14767.

ERG: Neil P. Dufton, Claire R. Peghaire, Lourdes Osuna-Almagro, Claudio Raimondi, Viktoria Kalna, Abhishek Chuahan, Gwilym Webb, Youwen Yang, Graeme M. Birdsey, Patricia Lalor, Justin C. Mason, David H. Adams, Anna M. RandiNat Commun. 2017; 8: 895. Published online 2017 Oct 12. doi: 10.1038/s41467-017-01169-0PMCID: PMC5638819 Ashley P. Ng, Yifang Hu, Donald Metcalf, Craig D. Hyland, Helen Ierino, Belinda Phipson, Di Wu, Tracey M. Baldwin, Maria Kauppi, Hiu Kiu, Ladina Di Rago, Douglas J. Hilton.

ERI1: Characterization of 3'hExo, a 3' exonuclease specifically interacting with the 3' end of histone mRNA. (PMID: 16912046) Yang XC . Dominski Z The Journal of biological chemistry 2006 3 4 23 58 Crystallographic structure of the nuclease domain of 3'hExo, a DEDDh family member, bound to rAMP. (PMID: 15451662) Cheng Y . Patel DJ Journal of molecular biology 2004 3 4 23 58 A 3' exonuclease that specifically interacts with the 3' end of histone mRNA. (PMID: 14536070) Dominski Z . Marzluff WF Mole

ERRFI1: Junmei Cairns, Brooke L Fridley, Gregory D Jenkins, Yongxian Zhuang, Jia Yu, Liewei WangEMBO Rep. 2018 Mar; 19(3): e44767. Published online 2018 Jan 15. doi: 10.15252/embr.201744767PMCID: PMC5835844Cristina Migliore, Elena Morando, Elena Ghiso, Sergio Anastasi, Vera P Leoni, Maria Apicella, Davide Cora', Anna Sapino, Filippo Pietrantonio, Filippo De Braud, Amedeo Columbano, Oreste Segatto, Silvia GiordanoEMBO Mol Med. 2018 Sep; 10(9): e8746. Published online 2018 Jul 24. doi: 10.15252/emmm.

ESR1: Takeshita T, Yamamoto Y, Yamamoto-Ibusuki M, et al. Clinical significance of monitoring ESR1 mutations in circulating cell-free DNA in estrogen receptor positive breast cancer patients. Oncotarget. 2016;7(22):32504-32518. doi:10.18632/oncotarget.8839.

ESR2: Rumi MAK, Singh P, Roby KF, et al. Defining the Role of Estrogen Receptor ?in the Regulation of Female Fertility. Endocrinology. 2017;158(7):2330-2343. doi:10.1210/en.2016-1916.

ETFDH: High frequency of ETFDH c.250G>A mutation in Taiwanese patients with late-onset lipid storage myopathy. (PMID: 20370797) Lan MY . Chen SS Clinical genetics 2010 3 4 43 56 ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. (PMID: 19249206) Liang WC . Nishino I Neuromuscular disorders : NMD 2009 3 4 23 56 The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferr

EVA1A: Mengtao Li, Guang Lu, Jia Hu, Xue Shen, Jiabao Ju, Yuanxu Gao, Liujing Qu, Yan Xia, Yingyu Chen, Yun BaiStem Cell Reports. 2016 Mar 8; 6(3): 396–410. Published online 2016 Feb 18. doi: 10.1016/j.stemcr.2016.01.011PMCID: PMC4788774Xin Lin, Ming Cui, Dong Xu, Dubeiqi Hong, Yan Xia, Chentong Xu, Riyong Li, Xuan Zhang, Yaxin Lou, Qihua He, Ping Lv, Yingyu ChenCell Death Dis. 2018 Jul; 9(7): 768. Published online 2018 Jul 10. doi: 10.1038/s41419-018-0800-x PMCID: PMC6039435 Shu Zhang, Xin Lin.

EVI5: Didonna A, Isobe N, Caillier SJ, et al. A non-synonymous single-nucleotide polymorphism associated with multiple sclerosis risk affects the EVI5 interactome. Human Molecular Genetics. 2015;24(24):7151-7158. doi:10.1093/hmg/ddv412.

F12: Gao WND, Carpentier DCJ, Ewles HA, Lee S, Smith GL. Vaccinia virus proteins A36 and F12/E2 show strong preferences for different kinesin light chain isoforms. Traffic (Copenhagen, Denmark). 2017;18(8):505-518. doi:10.1111/tra.12494.

F13A1: Peltier J, Roperch J-P, Audebert S, Borg J-P, Camoin L. Activation peptide of the coagulation factor XIII (AP-F13A1) as a new biomarker for the screening of colorectal cancer. Clinical Proteomics. 2018;15:15. doi:10.1186/s12014-018-9191-3.

F13B: Guo Z-P, Hou H-T, Jing R, Song Z-G, Liu X-C, He G-W. Plasma protein profiling in patients undergoing coronary artery bypass grafting surgery and clinical significance. Oncotarget. 2017;8(36):60528-60538. doi:10.18632/oncotarget.16366.

F2: Kamal RP, Alymova IV, York IA. Evolution and Virulence of Influenza A Virus Protein PB1-F2. International Journal of Molecular Sciences. 2018;19(1):96. doi:10.3390/ijms19010096.

F5: Guo J, Chan EWC, Chen S. Mechanism of substrate recognition by the novel Botulinum Neurotoxin subtype F5. Scientific Reports. 2016;6:19875. doi:10.1038/srep19875.

F7: Montagne A, Nikolakopoulou AM, Zhao Z, et al. Pericyte degeneration causes white matter dysfunction in the mouse CNS. Nature medicine. 2018;24(3):326-337. doi:10.1038/nm.4482.

F9: Wurpel DJ, Totsika M, Allsopp LP, et al. F9 Fimbriae of Uropathogenic Escherichia coli Are Expressed at Low Temperature and Recognise Gal?1-3GlcNAc-Containing Glycans. Cascales E, ed. PLoS ONE. 2014;9(3):e93177. doi:10.1371/journal.pone.0093177.

FABP2: Esteves A, Knoll-Gellida A, Canclini L, Silvarrey MC, André M, Babin PJ. Fatty acid binding proteins have the potential to channel dietary fatty acids into enterocyte nuclei. Journal of Lipid Research. 2016;57(2):219-232. doi:10.1194/jlr.M062232.

FADS1: Anthony D. Gromovsky, Rebecca C. Schugar, Amanda L. Brown, Robert N. Helsley, Amy C. Burrows, Daniel Ferguson, Renliang Zhang, Brian E. Sansbury, Richard G. Lee, Richard E. Morton, Daniela S. Allende, John S. Parks, Matthew Spite, J. Mark BrownArterioscler Thromb Vasc Biol. Author manuscript; available in PMC 2019 Jan 1.Published in final edited form as: Arterioscler Thromb Vasc Biol. 2018 Jan; 38(1): 218–231. Published online 2017 Oct 26. doi: 10.1161/ATVBAHA.117.309660PMCID: PMC5746431Lin

FADS2: Xie D, Fu Z, Wang S, et al. Characteristics of the fads2 gene promoter in marine teleost Epinephelus coioides and role of Sp1-binding site in determining promoter activity. Scientific Reports. 2018;8:5305. doi:10.1038/s41598-018-23668-w.

FAM111B: Sandra Mercier, Sébastien Küry, Emmanuelle Salort-Campana, Armelle Magot, Uchenna Agbim, Thomas Besnard, Nathalie Bodak, Chantal Bou-Hanna, Flora Bréhéret, Perrine Brunelle, Florence Caillon, Brigitte Chabrol, Valérie Cormier-Daire, Albert David, Bruno Eymard, Laurence Faivre, Dominique Figarella-Branger, Emmanuelle Fleurence, Mythily Ganapathi, Romain Gherardi, Alice Goldenberg, Antoine Hamel, Jeanine Igual, Alan D. Irvine, Dominique Israël-Biet, Caroline Kannengiesser, Christian Laboisse,

FAM124A: Wang K, Gao M, Yang M, et al. Transcriptome analysis of bronchoalveolar lavage fluid from children with severe Mycoplasma pneumoniae pneumonia reveals novel gene expression and immunodeficiency. Human Genomics. 2017;11:4. doi:10.1186/s40246-017-0101-y.

FAM129A: Alexandra G. Evstafieva, Irina E. Kovaleva, Maria S. Shoshinova, Andrei V. Budanov, Peter M. ChumakovPLoS One. 2018; 13(2): e0191107. Published online 2018 Feb 8. doi: 10.1371/journal.pone.0191107PMCID: PMC5805170Eva Sigstad, Elisabeth Paus, Trine Bjøro, Aasmund Berner, Krystyna Kotanska Grøholt, Lars H Jørgensen, Manuel Sobrinho-Simões, Ruth Holm, David J WarrenMod Pathol. 2012 Apr; 25(4): 537–547. Published online 2011 Dec 9. doi: 10.1038/modpathol.2011.188 PMCID: PMC3318159 Greg L. Shaw

FAM171A2: Schlesinger S, Kaffe B, Melcer S, et al. A hyperdynamic H3.3 nucleosome marks promoter regions in pluripotent embryonic stem cells. Nucleic Acids Research. 2017;45(21):12181-12194. doi:10.1093/nar/gkx817.

FAM19A2: Geoffrey A. Walford, Stefan Gustafsson, Denis Rybin, Alena Stanáková, Han Chen, Ching-Ti Liu, Jaeyoung Hong, Richard A. Jensen, Ken Rice, Andrew P. Morris, Reedik Mägi, Anke Tönjes, Inga Prokopenko, Marcus E. Kleber, Graciela Delgado, Günther Silbernagel, Anne U. Jackson, Emil V. Appel, Niels Grarup, Joshua P. Lewis, May E. Montasser, Claes Landenvall, Harald Staiger, Jian'an Luan, Timothy M. Frayling, Michael N. Weedon, Weijia Xie, Sonsoles Morcillo, María Teresa Martínez-Larrad.

FAM209B: You Li, Xiaosheng Wang, Suleyman Vural, Nitish K. Mishra, Kenneth H. Cowan, Chittibabu Guda PLoS One. 2015; 10(3): e0119383. Published online 2015 Mar 24. doi: 10.1371/journal.pone.0119383 PMCID: PMC4372331 Anastasia G. Efthymiou, Alison M. GoateMol Neurodegener. 2017; 12: 43. Published online 2017 May 26. doi: 10.1186/s13024-017-0184-xPMCID: PMC5446752

FAM58A: Guen VJ, Gamble C, Perez DE, et al. STAR syndrome-associated CDK10/Cyclin M regulates actin network architecture and ciliogenesis . Cell Cycle. 2016;15(5):678-688. doi:10.1080/15384101.2016.1147632.

FAM69A: Ohtsubo Y, Nagata Y, Tsuda M. Efficient N-tailing of blunt DNA ends by Moloney murine leukemia virus reverse transcriptase. Scientific Reports. 2017;7:41769. doi:10.1038/srep41769.

FAM71F1: Ur Rehman Z, Khan FA, Farmanullah, et al. Transcriptome profiling of anti-müllerian hormone treated preantral/small antral mouse ovary follicles. Oncotarget. 2018;9(54):30253-30267. doi:10.18632/oncotarget.25572.

FANCA: Liangyue Qian, Fenghua Yuan, Paola Rodriguez-Tello, Suyog Padgaonkar, Yanbin ZhangPLoS One. 2013; 8(12): e82666. Published online 2013 Dec 4. doi: 10.1371/journal.pone.0082666PMCID: PMC3857783 Anaid Benitez, Fenghua Yuan, Satoshi Nakajima, Leizhen Wei, Liangyue Qian, Richard Myers, Jennifer J. Hu, Li Lan, Yanbin Zhang Nucleic Acids Res. 2014 Feb; 42(3): 1671–1683. Published online 2013 Oct 28. doi: 10.1093/nar/gkt975PMCID: PMC3919598 Patrycja Pawlikowska, Pierre Fouchet, William Vainchenker

FARP1: Lucas Cheadle, Thomas BiedererJ Cell Biol. 2012 Dec 10; 199(6): 985–1001. doi: 10.1083/jcb.201205041PMCID: PMC3518221 Lucas Cheadle, Thomas BiedererJ Neurosci. 2014 Jun 4; 34(23): 7999–8009. doi: 10.1523/JNEUROSCI.3950-13.2014 PMCID: PMC4044256Yi-Chun Kuo, Xiaojing He, Andrew J. Coleman, Yu-Ju Chen, Pranathi Dasari, Jen Liou, Thomas Biederer, Xuewu Zhang Sci Rep. 2018; 8: 10477. Published online 2018 Jul 11. doi: 10.1038/s41598-018-28692-4 PMCID: PMC6041286

FASLG: Li Y, Sun Y, Cai M, et al. Fas Ligand Gene (Faslg) Plays an Important Role in Nerve Degeneration and Regeneration After Rat Sciatic Nerve Injury. Frontiers in Molecular Neuroscience. 2018;11:210. doi:10.3389/fnmol.2018.00210.

FBN2: Gerhard Sengle, Valerie Carlberg, Sara F. Tufa, Noe L. Charbonneau, Silvia Smaldone, Eric J. Carlson, Francesco Ramirez, Douglas R. Keene, Lynn Y. SakaiPLoS Genet. 2015 Jun; 11(6): e1005340. Published online 2015 Jun 26. doi: 10.1371/journal.pgen.1005340PMCID: PMC4482570Yanrong Shi, Yidong Tu, Robert P. Mecham, Steven BassnettInvest Ophthalmol Vis Sci. 2013 Nov; 54(12): 7163–7173. Published online 2013 Nov 1. doi: 10.1167/iovs.13-12687PMCID: PMC3816615Jillian G. Buchan, David M. Alvarado, G

FCER1A: Korosec P, Turner PJ, Silar M, et al. Basophils, high-affinity IgE receptors, and CCL2 in human anaphylaxis. The Journal of Allergy and Clinical Immunology. 2017;140(3):750-758.e15. doi:10.1016/j.jaci.2016.12.989.

FCHSD1: Almeida-Souza L, Frank RAW, García-Nafría J, et al. A Flat BAR Protein Promotes Actin Polymerization at the Base of Clathrin-Coated Pits. Cell. 2018;174(2):325-337.e14. doi:10.1016/j.cell.2018.05.020.

FCN1: A Genome-Wide Association Study of Basal Transepidermal Water Loss Finds that Variants at 9q34.3 Are Associated with Skin Barrier Function Journal of Investigative Dermatology (2017)Carbohydrate recognition properties of human ficolins: glycan array screening reveals the sialic acid binding specificity of M-ficolin. (PMID: 20032467) Gout E … Thielens NM The Journal of biological chemistry 2010 3 4 22 Trivalent recognition unit of innate immunity system: crystal structure of trimeric human M-ficolin fibrinogen-like domain. (PMID: 17148457) Tanio M … Kohno T The Journal of biological chemistry 2007 3 4 22 Polymorphisms in the ficolin 1 gene (FCN1) are associated with susceptibility to the development of rheumatoid arthritis. (PMID: 18032536) Vander Cruyssen B … De Keyser F Rheumatology (Oxford, England) 2007 3 22 40 Human ficolin: cDNA cloning, demonstration of peripheral blood leucocytes as the major site of synthesis and assignment of the gene to chromosome 9. (PMID: 8573080) Lu J … Reid KB The Biochemical journal 1996 2 3 4 Characterization of ficolins as novel elastin-binding proteins and molecular cloning of human ficolin-1. (PMID: 8947836) Harumiya S … Ichijo H Journal of biochemistry 1996

FECH: Basavarajappa HD, Sulaiman RS, Qi X, et al. Ferrochelatase is a therapeutic target for ocular neovascularization. EMBO Molecular Medicine. 2017;9(6):786-801. doi:10.15252/emmm.201606561.

FGD6: Thongnak C, Hnoonual A, Tangviriyapaiboon D, et al. Whole-Exome Sequencing Identifies One De Novo Variant in the FGD6 Gene in a Thai Family with Autism Spectrum Disorder. International Journal of Genomics. 2018;2018:8231547. doi:10.1155/2018/8231547.

FGF20: Biggs LC, Mäkelä OJ, Myllymäki S-M, et al. Hair follicle dermal condensation forms via Fgf20 primed cell cycle exit, cell motility, and aggregation. Horsley V, Schekman R, eds. eLife. 2018;7:e36468. doi:10.7554/eLife.36468.

FGFR1: Luo J, Liu S, Leung S, et al. An mRNA Gene Expression–Based Signature to Identify FGFR1-Amplified Estrogen Receptor–Positive Breast Tumors. The Journal of Molecular Diagnostics?: JMD. 2017;19(1):147-161. doi:10.1016/j.jmoldx.2016.09.007.

FGFR3: Mélanie Mahe, Florent Dufour, Hélène Neyret-Kahn, Aura Moreno-Vega, Claire Beraud, Mingjun Shi, Imene Hamaidi, Virginia Sanchez-Quiles, Clementine Krucker, Marion Dorland-Galliot, Elodie Chapeaublanc, Remy Nicolle, Hervé Lang, Celio Pouponnot, Thierry Massfelder, François Radvanyi, Isabelle Bernard-PierrotEMBO Mol Med. 2018 Apr; 10(4): e8163. Published online 2018 Feb 20. doi: 10.15252/emmm.201708163PMCID: PMC5887543Siru Zhou, Yangli Xie, Junzhou Tang, Junlan Huang, Qizhao Huang, Wei Xu, Zu

FGFR4: Quintanal-Villalonga Á, Ojeda-Márquez L, Marrugal Á, et al. The FGFR4-388arg Variant Promotes Lung Cancer Progression by N-Cadherin Induction. Scientific Reports. 2018;8:2394. doi:10.1038/s41598-018-20570-3.

FKBP5: Criado-Marrero M, Morales Silva RJ, Velazquez B, et al. Dynamic expression of FKBP5 in the medial prefrontal cortex regulates resiliency to conditioned fear. Learning & Memory. 2017;24(4):145-152. doi:10.1101/lm.043000.116.

FLG: Blunder S, Rühl R, Moosbrugger-Martinz V, et al. Alterations in Epidermal Eicosanoid Metabolism Contribute to Inflammation and Impaired Late Differentiation in FLG-Mutated Atopic Dermatitis. The Journal of investigative dermatology. 2017;137(3):706-715. doi:10.1016/j.jid.2016.09.034.

FLJ10986: Singh C, Glaab E, Linster CL. Molecular Identification of d-Ribulokinase in Budding Yeast and Mammals. The Journal of Biological Chemistry. 2017;292(3):1005-1028. doi:10.1074/jbc.M116.760744.

FLJ25967: Tosolini M, Pont F, Poupot M, et al. Assessment of tumor-infiltrating TCRV?9V?2 ?? lymphocyte abundance by deconvolution of human cancers microarrays. Oncoimmunology. 2017;6(3):e1284723. doi:10.1080/2162402X.2017.1284723.

FLJ33534: Wan X, Huang W, Yang S, et al. Identification of androgen-responsive lncRNAs as diagnostic and prognostic markers for prostate cancer. Oncotarget. 2016;7(37):60503-60518. doi:10.18632/oncotarget.11391.

FMN2: Agís-Balboa RC, Pinheiro PS, Rebola N, et al. Formin 2 links neuropsychiatric phenotypes at young age to an increased risk for dementia. The EMBO Journal. 2017;36(19):2815-2828. doi:10.15252/embj.201796821.

FMO3: Shih DM, Wang Z, Lee R, et al. Flavin containing monooxygenase 3 exerts broad effects on glucose and lipid metabolism and atherosclerosis. Journal of Lipid Research. 2015;56(1):22-37. doi:10.1194/jlr.M051680.

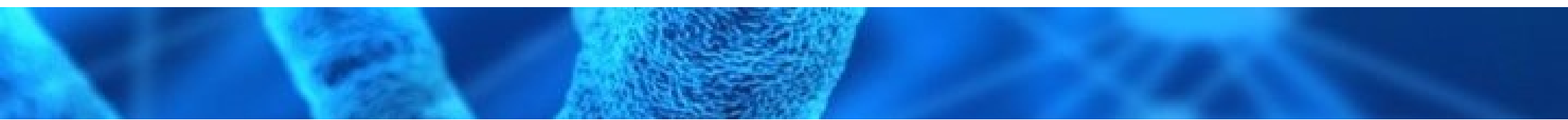
FOXE1: Bullock M, Lim G, Li C, et al. Thyroid transcription factor FOXE1 interacts with ETS factor ELK1 to co-regulate TERT. Oncotarget. 2016;7(52):85948-85962. doi:10.18632/oncotarget.13288.

FOXN4: The finished DNA sequence of human chromosome 12. (PMID: 16541075) Scherer SE ... Baylor College of Medicine Human Genome Sequencing Center Sequence Production Team Nature 2006 3 4 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004 3 4 Expression of the winged helix/forkhead gene, foxn4, during zebrafish development. (PMID: 15464224) Danilova N ... Steiner LA Brain research. Developmental brain research 2004 3 4 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 Identification of FOXN4 as a tumor suppressor of breast carcinogenesis via the activation of TP53 and deactivation of Notch signaling. (PMID: 31430519) Wang X ... Chen Z Gene 2020

FRMD4A: Zheng X, Jia B, Lin X, et al. FRMD4A: A potential therapeutic target for the treatment of tongue squamous cell carcinoma. International Journal of Molecular Medicine. 2016;38(5):1443-1449. doi:10.3892/ijmm.2016.2745.

FSIP1: Meisi Yan, Jinsong Wang, Yanlv Ren, Lin Li, Weidan He, Ying Zhang, Tong Liu, Zhigao LiCell Death Dis. 2019 Mar; 10(3): 204. Published online 2019 Feb 27. doi: 10.1038/s41419-018-1248-8PMCID: PMC6393503 Tong Liu, Hao Zhang, Li Sun, Danyu Zhao, Peng Liu, Meisi Yan, Neeha Zaidi, Sudeh Izadmehr, Animesh Gupta, Wahid Abu-Amer, Minna Luo, Jie Yang, Xunyan Ou, Yining Wang, Xuefeng Bai, Yan Wang, Maria I. New, Mone Zaidi, Tony Yuen, Caigang LiuProc Natl Acad Sci U S A. 2017 Jul 18;

FST: Seachrist DD, Sizemore ST, Johnson E, Abdul-Karim FW, Weber Bonk KL, Keri RA. Follistatin is a metastasis suppressor in a mouse model of HER2-positive breast cancer. Breast Cancer Research?: BCR. 2017;19:66. doi:10.1186/s13058-017-0857-y.



FTO: Y.C. Loraine Tung, Pawan Gulati, Che-Hsiung Liu, Debra Rimmington, Rowena Dennis, Marcella Ma, Vladimir Saudek, Stephen O'Rahilly, Anthony P. Coll, Giles S.H. YeoMol Metab. 2015 Apr; 4(4): 287-298. Published online 2015 Feb 7. doi: 10.1016/j.molmet.2015.01.011 PMCID: PMC4354923Daniel P. S. Osborn, Rosa Maria Roccasecca, Fiona McMurray, Victor Hernandez-Hernandez, Sriparna Mukherjee, Inês Barroso, Derek Stemple, Roger Cox, Philip L. Beales, Sonia Christou-SavinaPLoS One. 2014; 9(2): e87662. Fat mass and obesity-associated (FTO) and leptin receptor (LEPR) gene polymorphisms in Egyptian obese subjects. (PMID: 30767572) Ali EMM ... Settin A Archives of physiology and biochemistry 2021 3 Effect of Posttranslational Modifications on the Structure and Activity of FTO Demethylase. (PMID: 33925955) Marcinkowski M ... Poznański J International journal of molecular sciences 2021 3 Association of FTO gene methylation with incident type 2 diabetes mellitus: A nested case-control study. (PMID: 33753148) Huang S ... Hu D Gene 2021 3 Variants in the Obesity-Linked FTO gene locus modulates psychopathological features of patients with Anorexia Nervosa. (PMID: 33737121) González LM ... Gervasini G Gene 2021

FUT2: Velkova A, Diaz JEL, Pangilinan F, et al. The FUT2 secretor variant p.Trp154Ter influences serum vitamin B12 concentration via holo-haptocorrin, but not holo-transcobalamin, and is associated with haptocorrin glycosylation. Human Molecular Genetics. 2017; The role of the blood group-related glycosyltransferases FUT2 and B4GALNT2 in susceptibility to infectious disease. (PMID: 33662872) Galeev A ... Grassl GA International journal of medical microbiology : IJMM 2021 3 Maternal Fucosyltransferase 2 Status Associates with the Profiles of Human Milk Oligosaccharides and the Fecal Microbiota Composition of Breastfed Infants. (PMID: 33677972) Liu F ... Guo H Journal of agricultural and food chemistry 2021 Interactome Mapping Provides a Network of Neurodegenerative Disease Proteins and Uncovers Widespread Protein Aggregation in Affected Brains. (PMID: 32814053) Haenig C ... Wanker EE Cell reports 2020 3 Association of FUT2 and ABO with Crohn's disease in Koreans. (PMID: 31260595) Ye BD ... Song K Journal of gastroenterology and hepatology 2020 3 "FUT2" a potential genetic modifier in NCF1 deficiency. (PMID: 31494296) Bargir UA ... Madkaikar MR The journal of allergy and clinical immunology. In practice 2020 3 Genetic Manipulation of Human Intestinal Enteroids Demonstrates the Necessity of a Functional Fucosyltransferase 2 Gene for Secretor-Dependent Human Norovirus Infection. (PMID: 32184242) Haga K ... Estes MK mBio 2020 3 Can the FUT 2 Gene Variant Have an Effect on the Body Weight of Patients Undergoing Bariatric Surgery?-Preliminary, Exploratory Study. (PMID: 32872099) Komorniak N ... Stachowska E Nutrients 2020

FUT8: Praveen Agrawal, Barbara Fontanals-Cirera, Elena Sokolova, Samson Jacob, Christopher A. Vaiana, Diana Argibay, Veronica Davalos, Meagan McDermott, Shruti Nayak, Farbod Darvishian, Mireia Castillo, Beatrix Ueberheide, Iman Osman, David Fenyö, Lara K. Mahal, Eva HernandoCancer Cell. Author manuscript; available in PMC 2018 Jun 12. Published in final edited form as: Cancer Cell. 2017 Jun 12; 31(6): 804–819.e7. doi: 10.1016/j.ccell.2017.05.007PMCID: PMC5649440Cheng-Fen Tu, Meng-Ying Wu.

FYCO1: Da Ros M, Lehtiniemi T, Olotu O, et al. FYCO1 and autophagy control the integrity of the haploid male germ cell-specific RNP granules. Autophagy. 2017;13(2):302-321. doi:10.1080/15548627.2016.1261319.

G6PD: Recht J, Ashley EA, White NJ. Use of primaquine and glucose-6-phosphate dehydrogenase deficiency testing: Divergent policies and practices in malaria endemic countries. Sinnis P, ed. PLoS Neglected Tropical Diseases. 2018;12(4):e0006230. doi:10.1371/journ

GAB2: Zhang X, Lavoie G, Méant A, et al. Extracellular Signal-Regulated Kinases 1 and 2 Phosphorylate Gab2 To Promote a Negative-Feedback Loop That Attenuates Phosphoinositide 3-Kinase/Akt Signaling. Molecular and Cellular Biology. 2017;37(7):e00357-16. doi:10.

GAD1: Wang M, Cai E, Fujiwara N, et al. Odorant Sensory Input Modulates DNA Secondary Structure Formation and Heterogeneous Ribonucleoprotein Recruitment on the Tyrosine Hydroxylase and Glutamic Acid Decarboxylase 1 Promoters in the Olfactory Bulb. The Journal

GATA2: Kaimakis P, de Pater E, Eich C, et al. Functional and molecular characterization of mouse Gata2-independent hematopoietic progenitors. Blood. 2016;127(11):1426-1437. doi:10.1182/blood-2015-10-673749.

GATA4: Cayla A. Thompson, Kevin Wojta, Kirthi Pulakanti, Sridhar Rao, Paul Dawson, Michele A. BattleCell Mol Gastroenterol Hepatol. 2017 May; 3(3): 422–446. Published online 2017 Jan 24. doi: 10.1016/j.jcmgh.2016.12.009PMCID: PMC5404030 Su-Ren Chen, Ji-Xin Tang, Jin-Mei Cheng, Jian Li, Cheng Jin, Xiao-Yu Li, Shou-Long Deng, Yan Zhang, Xiu-Xia Wang, Yi-Xun LiuOncotarget. 2015 Nov 10; 6(35): 37012–37027. Published online 2015 Oct 14. doi: 10.18632/oncotarget.6115PMCID: PMC4741912 Boni A. Afouda.

GATM: Turer E, McAlpine W, Wang K, et al. Creatine maintains intestinal homeostasis and protects against colitis. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(7):E1273-E1281. doi:10.1073/pnas.1621400114.

GCH1: Gillian Douglas, Ashley B. Hale, Mark J Crabtree, Brent J. Ryan, Alex Hansler, Katrin Watschinger, Steven S Gross, Craig A. Lygate, Nicholas J. Alp, Keith M. ChannonDev Biol. 2015 Mar 1; 399(1): 129–138. doi: 10.1016/j.ydbio.2014.12.025 PMCID: PMC4347993 Katja Zschiebsch, Caroline Fischer, Annett Wilken-Schmitz, Gerd Geisslinger, Keith Channon, Katrin Watschinger, Irmgard TegederJ Cell Mol Med. 2019 Feb; 23(2): 985–1000. Published online 2018 Nov 18.

GCK: Matthew G. Rees, Mindy I. Davis, Min Shen, Steve Titus, Anne Raimondo, Amy Barrett, Anna L. Gloyn, Francis S. Collins, Anton SimeonovPLoS One. 2014; 9(2): e89335. Published online 2014 Feb 19. doi: 10.1371/journal.pone.0089335PMCID: PMC3929664Brian Lu, Kiran Kurmi, Miguel Munoz-Gomez, Egon J. Jacobus Ambuludi, Jason M. Tonne, Kuntol Rakshit, Taro Hitosugi, Yogish C. Kudva, Aleksey V. Matveyenko, Yasuhiro IkedaDis Model Mech. 2018 Jun 1; 11(6): dmm033316. Published online 2018 Jun 13. doi: 1

GCKR: Zhao Yang Wang, Ling Jin, Huanran Tan, David M. IrwinPLoS One. 2013; 8(4): e60896. Published online 2013 Apr 1. doi: 10.1371/journal.pone.0060896PMCID: PMC3613411Ling Jin, Tingting Guo, Zhixin Li, Zhen Lei, Hui Li, Yiqing Mao, Xi Wang, Na Zhou, Yizhuang Zhang, Ruobi Hu, Xuehui Zhang, Gang Niu, David M. Irwin, Huanran TanInt J Mol Sci. 2015 Apr; 16(4): 7377–7393. Published online 2015 Apr 2. doi: 10.3390/ijms16047377PMCID: PMC4425023Maykel López Rodríguez, Lilian Fernandes Silva, Jagadish Va

GCLC: Cristina Espinosa-Díez, Verónica Miguel, Susana Vallejo, Francisco J. Sánchez, Elena Sandoval, Eva Blanco, Pablo Cannata, Concepción Peiró, Carlos F. Sánchez-Ferrer, Santiago LamasRedox Biol. 2018 Apr; 14: 88–99. Published online 2017 Sep 1. doi: 10.1016/j.redox.2017.08.019PMCID: PMC5596265Alexey Moskalev, Mikhail Shaposhnikov, Ekaterina Proshkina, Alexey Belyi, Alexander Fedintsev, Svetlana Zhikrivetskaya, Zulfiya Guvatova, Asiya Sadritdinova, Anastasia Snezhkina, George Krasnov, Anna Kudr

GCLM: Jinhwan Lim, Brooke N. Nakamura, Isaac Mohar, Terrance J. Kavanagh, Ulrike LudererEndocrinology. 2015 Sep; 156(9): 3329–3343. Published online 2015 Jun 17. doi: 10.1210/en.2015-1206PMCID: PMC4541624Chad S. Weldy, Ian P. Luttrell, Collin C. White, Vicki Morgan-Stevenson, Theo K. Bammler, Richard P. Beyer, Zahra Afsharinejad, Francis Kim, Kanchan Chitaley, Terrance J. KavanaghFree Radic Biol Med. Author manuscript; available in PMC 2013 Sep 15.Published in final edited form as: Free Radic Bio

GHRL: Wu S, Liu J, Wang X, Li M, Chen Z, Tang Y. Aberrant Expression of the Long Non-coding RNA GHRLOS and Its Prognostic Significance in Patients with Colorectal Cancer. Journal of Cancer. 2017;8(19):4040-4047. doi:10.7150/jca.21304.

GJA8: The impact of GJA8 SNPs on susceptibility to age-related cataract. Yu X, et al. Hum Genet, 2018 Dec. PMID 30349978 Detection of c.139G>A (D47N) mutation in GJA8 gene in an extended family with inheritance of autosomal dominant zonular cataract without pulverulent opacities by exome sequencing. Gunda P, et al. J Genet, 2018 Sep. PMID 30262699

GJB2: Mikstiene V, Jakaitiene A, Byckova J, et al. The high frequency of GJB2 gene mutation c.313_326del14 suggests its possible origin in ancestors of Lithuanian population. BMC Genetics. 2016;17:45. doi:10.1186/s12863-016-0354-9.

GLCCI1: Kim S-H, Kim H-J, Kim C-W. GLCCI1 is a novel component associated with the PI3K signaling pathway in podocyte foot processes. Experimental & Molecular Medicine. 2016;48(5):e233-. doi:10.1038/emm.2016.28.

GLIS1: Tosic M, Allen A, Willmann D, et al. Lsd1 regulates skeletal muscle regeneration and directs the fate of satellite cells. Nature Communications. 2018;9:366. doi:10.1038/s41467-017-02740-5.

GLO1: Jandial R, Neman J, Lim PP, et al. Inhibition of GLO1 in Glioblastoma Multiforme Increases DNA-AGEs, Stimulates RAGE Expression, and Inhibits Brain Tumor Growth in Orthotopic Mouse Models. International Journal of Molecular Sciences. 2018;19(2):406. doi:1

GMDS: Xing Wei, Kun Zhang, Haifeng Qin, Jinlong Zhu, Qiaoxi Qin, Yang Yu, Hong WangBMC Cancer. 2018; 18: 600. Published online 2018 May 29. doi: 10.1186/s12885-018-4524-1PMCID: PMC5975429 Magdalena Carlberg, Maigun Edhborg, Lene LindbergAm J Mens Health. 2018 Jul; 12(4): 720–729. Published online 2018 Jan 19. doi: 10.1177/1557988317749071PMCID: PMC6131440 Genevieve D. E. Haliburton, Gabriel L. McKinsey, Katherine S. Pollard Neurogenetics. 2016; 17: 1–9. Published online 2015 Sep 17.

GOLM1: Li RM, Nai MM, Duan SJ, et al. Down-expression of GOLM1 enhances the chemo-sensitivity of cervical cancer to methotrexate through modulation of the MMP13/EMT axis. American Journal of Cancer Research. 2018;8(6):964-980.

GPC5: Siwei Wang, Mantang Qiu, Wenjia Xia, Youtao Xu, Qixing Mao, Jie Wang, Gaochao Dong, Lin Xu, Xin Yang, Rong Yin Oncotarget. 2016 Nov 29; 7(48): 79736–79746. Published online 2016 Oct 27. doi: 10.18632/oncotarget.12945 PMCID: PMC5346747 Lixia Guo, Jingyu Wang, Ting Zhang, Yanan YangBiochem Biophys Rep. 2016 Jul; 6: 108–112. Published online 2016 Mar 21. doi: 10.1016/j.bbrep.2016.03.010 PMCID: PMC4832925 Alexander G. Bassuk, Lakshmi B. Muthuswamy, Riley Boland, Tiffany L. Smith, Alissa M. Huls

GPHN: Dejanovic B, Djémié T, Grünewald N, et al. Simultaneous impairment of neuronal and metabolic function of mutated gephyrin in a patient with epileptic encephalopathy. EMBO Molecular Medicine. 2015;7(12):1580-1594. doi:10.15252/emmm.201505323.

GPX1: Gan X, Chen B, Shen Z, et al. High GPX1 expression promotes esophageal squamous cell carcinoma invasion, migration, proliferation and cisplatin-resistance but can be reduced by vitamin D. International Journal of Clinical and Experimental Medicine. 2014;7

GPX3: Simultaneous genotyping of 11 non-synonymous SNPs in the 4 glutathione peroxidase genes using the multiplex single base extension method. (PMID: 19161995) Iida R ... Yasuda T Clinica chimica acta; international journal of clinical chemistry 2009 3 21 39 Glutathione peroxidase 3 gene polymorphisms and risk of differentiated thyroid cancer. (PMID: 19375609) Lin JC ... Juo SH Surgery 2009 3 21 39 Variation in the selenoenzyme genes and risk of advanced distal colorectal adenoma. (PMID: 18483336) Peters U ... Foster CB Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2008 3 21 39 Promoter polymorphisms in the plasma glutathione peroxidase (GPx-3) gene: a novel risk factor for arterial ischemic stroke among young adults and children. (PMID: 17122425) Voetsch B ... Loscalzo J Stroke 2007 3 21 39 Differential induction of extracellular glutathione peroxidase and nitric oxide synthase 2 in airways of healthy individuals exposed to 100% O(2) or cigarette smoke. (PMID: 10970826) Comhair SA ... Erzurum SC American journal of respiratory cell and molecular biology 2000

GPX4: Rohr-Udilova N, Bauer E, Timelthaler G, et al. Impact of glutathione peroxidase 4 on cell proliferation, angiogenesis and cytokine production in hepatocellular carcinoma. Oncotarget. 2018;9(11):10054-10068. doi:10.18632/oncotarget.24300.

GRIA3: Wei C-H, Wu G, Cai Q, et al. MicroRNA-330-3p promotes cell invasion and metastasis in non-small cell lung cancer through GRIA3 by activating MAPK/ERK signaling pathway. Journal of Hematology & Oncology. 2017;10:125. doi:10.1186/s13045-017-0493-0.

GRID1: Dissection of phenotype reveals possible association between schizophrenia and Glutamate Receptor Delta 1 (GRID1) gene promoter. (PMID: 19346103) Treutlein J ... Rietschel M Schizophrenia research 2009 3 23 41 54 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 41 54 Genetic variants associated with cardiac structure and function: a meta-analysis and replication of genome-wide association data. (PMID: 19584346) Vasan RS ... Blankenberg S JAMA 2009 3 41 54 A case-control association study between the GRID1 gene and schizophrenia in the Chinese Northern Han population. (PMID: 17490860) Guo SZ ... He L Schizophrenia research 2007 3 41 54 Bipolar I disorder and schizophrenia: a 440-single-nucleotide polymorphism screen of 64 candidate genes among Ashkenazi Jewish case-parent trios. (PMID: 16380905) Fallin MD ... Pulver AE American journal of human genetics 2005

GRK3: Chromosome mapping of the human arrestin (SAG), beta-arrestin 2 (ARRB2), and beta-adrenergic receptor kinase 2 (ADRBK2) genes. (PMID: 7695743) Calabrese G ... De Blasi A Genomics 1994 2 3 21 Molecular cloning, functional expression and mRNA analysis of human beta-adrenergic receptor kinase 2. (PMID: 8427589) Parruti G ... De Blasi A Biochemical and biophysical research communications 1993 3 4 21 Allele specific analysis of the ADRBK2 gene in lymphoblastoid cells from bipolar disorder patients. (PMID: 19766236) McCarthy MJ ... Turner EE Journal of psychiatric research 2010 3 21 beta-Adrenoceptor and GRK3 expression in human lymphocytes is related to blood pressure and urinary albumin excretion. (PMID: 20216086) Oliver E ... D'Ocon P Journal of hypertension 2010 3 21 Leukocyte analysis from WHIM syndrome patients reveals a pivotal role for GRK3 in CXCR4 signaling. (PMID: 18274673) Balabanian K ... Bachelerie F The Journal of clinical investigation 2008 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021

GRK5: Hua Xu, Xiaoshan Jiang, Ke Shen, Christopher C. Fischer, Philip B. WedegaertnerMol Biol Cell. 2014 Jul 1; 25(13): 2105–2115. doi: 10.1091/mbc.E13-09-0547PMCID: PMC4072583 Konstantin E. Komolov, Anshul Bhardwaj, Jeffrey L. BenovicJ Biol Chem. 2015 Aug 21; 290(34): 20629–20647. Published online 2015 Jun 1. doi: 10.1074/jbc.M115.647297PMCID: PMC4543624 Konstantin E. Komolov, Yang Du, Nguyen Minh Duc, Robin M. Betz, João P. G. L. M. Rodrigues, Ryan D. Leib, Dhabaleswar Patra, Georgios Skiniotis

GRM5: Genome-wide association study of COVID-19 severity among the Chinese population Yuanfeng Li, Yuehua Ke, ...Gangqiao Zhou Show authors Cell Discovery volume 7, Article number: 76 (2021) Laura T. Haas, Santiago V. Salazar, Mikhail A. Kostylev, Ji Won Um, Adam C. Kaufman, Stephen M. StrittmatterBrain. 2016 Feb; 139(2): 526–546. Published online 2015 Dec 14. doi: 10.1093/brain/awv356PMCID: PMC4840505Despoina Goniotaki, Asvin K. K. Lakkaraju, Amulya N. Shrivastava, Pamela Bakirci, Silvia Sorce, Assunta Senatore, Rajlakshmi Marpakwar, Simone Hornemann, Fabrizio Gasparini, Antoine Triller, Adriano AguzziPLoS Pathog. 2017 Nov; 13(11): e1006733. Published online 2017 Nov 27. doi:

GRN: Arrant AE, Onyilo VC, Unger DE, Roberson ED. Progranulin Gene Therapy Improves Lysosomal Dysfunction and Microglial Pathology Associated with Frontotemporal Dementia and Neuronal Ceroid Lipofuscinosis. The Journal of Neuroscience. 2018;38(9):2341-2358. do

GSDMB: Genetic variation in ORM1-like 3 (ORMDL3) and gasdermin-like (GSDML) and childhood asthma. (PMID: 19133921) Wu H ... London SJ Allergy 2009 3 23 41 54 Differential expression and localisation of gasdermin-like (GSDML), a novel member of the cancer-associated GSDMDC protein family, in neoplastic and non-neoplastic gastric, hepatic, and colon tissues. (PMID: 18038310) Carl-McGrath S ... Röcken C Pathology 2008 3 4 23 54 Members of a novel gene family, Gsdm, are expressed exclusively in the epithelium of the skin and gastrointestinal tract in a highly tissue-specific manner. (PMID: 17350798) Tamura M ... Shiroishi T Genomics 2007 2 3 4 54 Transcriptional control of the HERV-H LTR element of the GSDML gene in human tissues and cancer cells. (PMID: 16625320) Sin HS ... Kim HS Archives of virology 2006 3 4 23 54 Evolutionary recombination hotspot around GSDML-GSDM locus is closely linked to the oncogenomic recombination hotspot around the PPP1R1B-ERBB2-GRB7 amplicon. (PMID: 15010812) Katoh M ... Katoh M International journal of oncology 2004

GSDML: Granell R, Henderson AJ, Timpson N, et al. Examination of the relationship between variation at 17q21 and childhood wheeze phenotypes. The Journal of allergy and clinical immunology. 2013;131(3):685-694. doi:10.1016/j.jaci.2012.09.021.

GSG1L: Mao X, Gu X, Lu W. GSG1L regulates the strength of AMPA receptor-mediated synaptic transmission but not AMPA receptor kinetics in hippocampal dentate granule neurons. Journal of Neurophysiology. 2017;117(1):28-35. doi:10.1152/jn.00307.2016.

GSR: Chul Han, Mi-Jung Kim, Dalian Ding, Hyo-Jin Park, Karessa White, Logan Walker, Tongjun Gu, Masaru Tanokura, Tatsuya Yamasoba, Paul Linser, Richard Salvi, Shinichi SomeyaPLoS One. 2017; 12(7): e0180817. Published online 2017 Jul 7. doi: 10.1371/journal.pone.0180817PMCID: PMC5501606Kai Lüersen, Dirk Stegehake, Jens Daniel, Mike Drescher, Irene Ajonina, Caroline Ajonina, Patrick Hertel, Christian Woltersdorf, Eva LiebauPLoS One. 2013; 8(4): e60731. Published online 2013 Apr 8. doi: 10.1371/jou

GSTM1: Yuan X-P, Liu L-S, Chen C-B, et al. MicroRNA-423-5p facilitates hypoxia/reoxygenation-induced apoptosis in renal proximal tubular epithelial cells by targeting GSTM1 via endoplasmic reticulum stress. Oncotarget. 2017;8(47):82064-82077. doi:10.18632/oncotarget.18289.

GSTP1: Harshbarger W, Gondi S, Ficarro SB, et al. Structural and Biochemical Analyses Reveal the Mechanism of Glutathione S-Transferase Pi 1 Inhibition by the Anti-cancer Compound Piperlongumine. The Journal of Biological Chemistry. 2017;292(1):112-120. doi:10.1

GUCY1A3: Kessler T, Wobst J, Wolf B, et al. Functional characterization of the GUCY1A3coronary artery disease risk locus. Circulation. 2017;136(5):476-489. doi:10.1161/CIRCULATIONAHA.116.024152.

GZMB: Defective gammadelta T-cell function and granzyme B gene polymorphism in a cohort of newly diagnosed breast cancer patients. (PMID: 19446661) Gaafar A ... Al-Hussein K Experimental hematology 2009 3 21 39 Disparate distribution of 16 candidate single nucleotide polymorphisms among racial and ethnic groups of pediatric heart transplant patients. (PMID: 17198275) Girnita DM ... Zeevi A Transplantation 2006 3 21 39 The three-dimensional structure of human granzyme B compared to caspase-3, key mediators of cell death with cleavage specificity for aspartic acid in P1. (PMID: 11325591) Rotonda J ... Becker JW Chemistry & biology 2001 3 4 21 Crystal structure of the caspase activator human granzyme B, a proteinase highly specific for an Asp-P1 residue. (PMID: 11209755) Estébanez-Perpiña E ... Bode W Biological chemistry 2000 3 4 21 Granzyme B mimics apical caspases. Description of a unified pathway for trans-activation of executioner caspase-3 and -7. (PMID: 9852092) Yang X ... Froelich CJ The Journal of biological chemistry 1998

HABP2: Stavenuiter F, Ebberink EHTM, Mertens K, Meijer AB. Role of glycine 221 in catalytic activity of hyaluronan-binding protein 2. The Journal of Biological Chemistry. 2017;292(15):6381-6388. doi:10.1074/jbc.M116.757849.

HBA1: AbdulAzeez S, Borgio JF. In-Silico Computing of the Most Deleterious nsSNPs in HBA1 Gene. Zhang Y, ed. PLoS ONE. 2016;11(1):e0147702. doi:10.1371/journal.pone.0147702.

HBB: Cai L, Bai H, Mahairaki V, et al. A Universal Approach to Correct Various HBBGene Mutations in Human Stem Cells for Gene Therapy of Beta-Thalassemia and Sickle Cell Disease. Stem Cells Translational Medicine. 2018;7(1):87-97. doi:10.1002/sctm.17-0066.

HCG9: Mrinal Pal, Sasha Ebrahimi, Gabriel Oh, Tarang Khare, Aiping Zhang, Zachary A. Kaminsky, Sun-Chong Wang, Arturas PetronisSchizophr Bull. 2016 Jan; 42(1): 170-177. Published online 2015 Jun 15. doi: 10.1093/schbul/sbv079PMCID: PMC4681545 Mu-Yun Wu, Shu-Jing Huang, Fan Yang, Xin-Tian Qin, Dong Liu, Ying Ding, Shu Yang, Xi-Cheng Wang Oncotarget. 2017 Aug 8; 8(32): 52708-52723. Published online 2017 Apr 13. doi: 10.18632/oncotarget.17085 PMCID: PMC5581063 Kevin Y. Urayama, Ruth F. Jarrett. Geno

HDAC4: Three proteins define a class of human histone deacetylases related to yeast Hda1p. (PMID: 10220385) Grozinger CM . Schreiber SL Proceedings of the National Academy of Sciences of the United States of America 1999 2 3 4 23 58 Haploinsufficiency of HDAC4 causes brachydactyly mental retardation syndrome, with brachydactyly type E, developmental delays, and behavioral problems. (PMID: 20691407) Williams SR . Elsea SH American journal of human genetics 2010 2 3 4 58 Involvement of histone deace

HDAC9: Smita Salian-Mehta, Mei Xu, Timothy A. McKinsey, Stuart Tobet, Margaret E. WiermanJ Biol Chem. 2015 May 29; 290(22): 14045–14056. Published online 2015 Apr 14. doi: 10.1074/jbc.M115.640482PMCID: PMC4447976 Christian L. Lino Cardenas, Chase W. Kessinger, Yisha Cheng, Carolyn MacDonald, Thomas MacGillivray, Brian Ghoshhajra, Luai Huleihel, Saifar Nuri, Ashish S. Yeri, Farouc A. Jaffer, Naftali Kaminski, Patrick Ellinor, Neal L. Weintraub, Rajeev Malhotra, Eric M. Isselbacher, Mark E. Lindsay

HERC2: Monica Cubillos-Rojas, Taiane Schneider, Ouadah Hadjebi, Leonardo Pedrazza, Jarbas Rodrigues de Oliveira, Francina Langa, Jean-Louis Guénet, Joan Duran, Josep Maria de Anta, Soledad Alcántara, Rocio Ruiz, Eva María Pérez-Villegas, Francisco J. Aguilar, Ángel M. Carrión, Jose Angel Armengol, Emma Baple, Andrew H. Crosby, Ramon Bartrons, Francesc Ventura, Jose Luis Rosa Oncotarget. 2016 Aug 30; 7(35): 56083–56106. Published online 2016 Aug 12. doi: 10.18632/oncotarget.11270PMCID: PMC5302898

HES1: Liu X-J, Yang B, Huang S-N, et al. Human cytomegalovirus IE1 downregulates Hes1 in neural progenitor cells as a potential E3 ubiquitin ligase. Murphy EA, ed. PLoS Pathogens. 2017;13(7):e1006542. doi:10.1371/journal.ppat.1006542.

HFE: Lee SY, Zhu J, Salzberg AC, et al. Analysis of single nucleotide variants of HFEgene and association to survival in The Cancer Genome Atlas GBM data. Langevin SM, ed. PLoS ONE. 2017;12(3):e0174778. doi:10.1371/journal.pone.0174778.

HHEX: Gauvrit S, Villasenor A, Strilic B, et al. HHEX is a transcriptional regulator of the VEGFC/FLT4/PROX1 signaling axis during vascular development. Nature Communications. 2018;9:2704. doi:10.1038/s41467-018-05039-1.

HIF1A: Wu Y, Yun D, Zhao Y, et al. Down regulation of RNA binding motif, single-stranded interacting protein 3, along with up regulation of nuclear HIF1A correlates with poor prognosis in patients with gastric cancer. Oncotarget. 2017;8(1):1262-1277. doi:10.1863

HIVEP2: Siddharth Srivastava, Hartmut Engels, Ina Schanze, Kirsten Cremer, Thomas Wieland, Moritz Menzel, Max Schubach, Saskia Biskup, Martina Kreiß, Sabine Endele, Tim M Strom, Dagmar Wieczorek, Martin Zenker, Siddharth Gupta, Julie Cohen, Alexander M Zink, SakkuBai NaiduEur J Hum Genet. 2016 Apr; 24(4): 556–561. Published online 2015 Jul 8. doi: 10.1038/ejhg.2015.151PMCID: PMC4929870 Zeina Bash-Imam, Gabriel Thérizols, Anne Vincent, Florian Lafôrets, Micaela Polay Espinoza, Nathalie Pion.

HLA: Goeury T, Creary LE, Brunet L, et al. Deciphering the fine nucleotide diversity of full HLA class I and class II genes in a well-documented population from sub-Saharan Africa. Hla. 2018;91(1):36-51. doi:10.1111/tan.13180.

HLA-B51: Pediatrics in Systemic Autoimmune Diseases I. Koné-Paut, in Handbook of Systemic Autoimmune Diseases, 2016 Pulmonary manifestations of systemic disease Bryan Corrin MD FRCPath, Andrew G. Nicholson DM FRCPath, in Pathology of the Lungs (Third Edition), 2011 Behcet's Syndrome P. Raval, in Reference Module in Biomedical Sciences, 2015

HLA-C: Parolini F, Biswas P, Serena M, et al. Stability and Expression Levels of HLA-C on the Cell Membrane Modulate HIV-1 Infectivity. Silvestri G, ed. Journal of Virology. 2018;92(1):e01711-17. doi:10.1128/JVI.01711-17.

HLA-DQA1: Liu B, Deng T, Zhu L, Zhong J. Association of human leukocyte antigen (HLA)-DQ and HLA-DQA1/DQB1 alleles with Vogt-Koyanagi-Harada disease: A systematic review and meta-analysis. Li. Y, ed. Medicine. 2018;97(7):e9914. doi:10.1097/MD.0000000000009914. A childhood acute lymphoblastic leukemia genome-wide association study identifies novel sex-specific risk variants Sandeep K. Singh, PhD,a,b Philip J. Lupo, PhD,c Michael E. Scheurer, PhD,d Anshul Saxena, MPH,e Amy E. Kennedy, PhD,f Boubakari I

HLA-DQA2: HLA-DQA2 and HLA-DQB2 genes are specifically expressed in human Langerhans cells and encode a new HLA class II molecule. (PMID: 22407913) Lenormand C . Tourne S Journal of immunology (Baltimore, Md. : 1950) 2012 3 4 58 Genome-wide association study of asthma identifies RAD50-IL13 and HLA-DR/DQ regions. (PMID: 20159242) Li X . Bleecker ER The Journal of allergy and clinical immunology 2010 3 45 58 Genome-wide association study identifies new HLA class II haplotypes strongly protective agains

HLA-DQB1: Jin P-P, Sun L-L, Ding B-J, et al. Human Leukocyte Antigen DQB1 (HLA-DQB1) Polymorphisms and the Risk for Guillain-Barré Syndrome: A Systematic Review and Meta-Analysis. Linker RA, ed. PLoS ONE. 2015;10(7):e0131374. doi:10.1371/journal.pone.0131374.

HLA-DRA: Sara Cajander, Elisabet Tina, Anders Bäckman, Anders Magnuson, Kristoffer Strålin, Bo Söderquist, Jan KällmanPLoS One. 2016; 11(5): e0154690. Published online 2016 May 4. doi: 10.1371/journal.pone.0154690PMCID: PMC4856385 Martin Sebastian Winkler, Anne Rissiek, Marion Priefler, Edzard Schwedhelm, Linda Robbe, Antonia Bauer, Corinne Zahrte, Christian Zoellner, Stefan Kluge, Axel NierhausPLoS One. 2017; 12(8): e0182427. Published online 2017 Aug 3. doi: 10.1371/journal.pone.0182427

HLA-DRB1: Yi Tian Ting, Jan Petersen, Sri H. Ramarathinam, Stephen W. Scally, Khai L. Loh, Ranjeny Thomas, Anish Suri, Daniel G. Baker, Anthony W. Purcell, Hugh H. Reid, Jamie RossjohnJ Biol Chem. 2018 Mar 2; 293(9): 3236–3251. Published online 2018 Jan 9. doi: 10.1074/jbc.RA117.001013PMCID: PMC5836122Lara Kular, Yun Liu, Sabrina Ruhrmann, Galina Zheleznyakova, Francesco Marabita, David Gomez-Cabrero, Tojo James, Ewoud Ewing, Magdalena Lindén, Bartosz Górnikiewicz, Shahin Aeinehband, Pernilla Stridh,

HLA-F: Human leukocyte antigen F (HLA-F). An expressed HLA gene composed of a class I coding sequence linked to a novel transcribed repetitive element. (PMID: 1688605) Geraghty DE . Koller BH The Journal of experimental medicine 1990 2 3 4 23 58 Genetic associations of common deletion polymorphisms in families with Avellino corneal dystrophy. (PMID: 19622345) Park M . Kim HL Biochemical and biophysical research communications 2009 3 23 45 58 SNP mapping and candidate gene sequencing in the class I

HMBS: Chen B, Solis-Villa C, Hakenberg J, et al. Acute Intermittent Porphyria: Predicted Pathogenicity of HMBS Variants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. Human mutation. 2016;37(11):1215-1222. doi:10.1002/humu.23067.

HMCN1: Westcot SE, Hatzold J, Urban MD, et al. Protein-Trap Insertional Mutagenesis Uncovers New Genes Involved in Zebrafish Skin Development, Including a Neuregulin 2a-Based ErbB Signaling Pathway Required during Median Fin Fold Morphogenesis. Carney TJ, ed. PL

HMGB1: The genetic variation of the human HMGB1 gene. (PMID: 17610420) Kornblit B ... Garred P Tissue antigens 2007 3 4 23 41 Oxidation of the alarmin high-mobility group box 1 protein (HMGB1) during apoptosis. (PMID: 19811284) Urbonaviciute V ... Voll RE Autoimmunity 2009 3 4 23 Human HMGB1 directly facilitates interactions between nucleotide excision repair proteins on triplex-directed psoralen interstrand crosslinks. (PMID: 19446504) Lange SS ... Vasquez KM DNA repair 2009 3 4 23 High mobility group box 1 protein binding to lipopolysaccharide facilitates transfer of lipopolysaccharide to CD14 and enhances lipopolysaccharide-mediated TNF-alpha production in human monocytes. (PMID: 18354232) Youn JH ... Shin JS Journal of immunology (Baltimore, Md. : 1950) 2008 3 4 23 Proteomic screen defines the hepatocyte nuclear factor 1alpha-binding partners and identifies HMGB1 as a new cofactor of HNF1alpha. (PMID: 18160415) Yu M ... Yang XM Nucleic acids research 2008

HNF1A: Luo Z, Li Y, Wang H, et al. Hepatocyte Nuclear Factor 1A (HNF1A) as a Possible Tumor Suppressor in Pancreatic Cancer. Trevino JG, ed. PLoS ONE. 2015;10(3):e0121082. doi:10.1371/journal.pone.0121082.

HNF1B: Two variants on chromosome 17 confer prostate cancer risk, and the one in TCF2 protects against type 2 diabetes. Gudmundsson J1, Sulem P, Steinthorsdottir V, Bergthorsson JT, Thorleifsson G, Manolescu A, Rafnar T, Gudbjartsson D, Agnarsson BA, Baker A, Sigurdsson A, Benediktsdottir KR, Jakobsdottir M, Blondal T, Stacey SN, Helgason A, Gunnarsdottir S, Olafsdottir A, Kristinsson KT, Birgisdottir B, Ghosh S, Thorlacius S, Magnusdottir D, Stefansdottir G, Kristjansson K, Bagger Y, Wilensky RL,

HR: Kim J-K, Kim I, Choi K, et al. Poly(rC) binding protein 2 acts as a negative regulator of IRES-mediated translation of Hr mRNA. Experimental & Molecular Medicine. 2018;50(2):e441-. doi:10.1038/emm.2017.262.

HRAS: Mutations in LZTR1 drive human disease by dysregulating RAS ubiquitination. (PMID: 30442762) Steklov M . Sablina AA Science (New York, N.Y.) 2018 3 4 58 Postzygotic HRAS and KRAS mutations cause nevus sebaceous and Schimmelpenning syndrome. (PMID: 22683711) Groesser L . Hafner C Nature genetics 2012 3 4 58 Molecular, morphologic, and outcome analysis of thyroid carcinomas according to degree of extrathyroid extension. (PMID: 20860430) Rivera M . Ghossein R Thyroid : official journal of the

HSD11B1: Association study of 11beta-hydroxysteroid dehydrogenase type 1 gene polymorphisms and metabolic syndrome in urban Japanese cohort. (PMID: 19535162) Miyamoto Y . Morisaki T Diabetes research and clinical practice 2009 3 23 43 56 Regulatory effect of common promoter polymorphisms on the expression of the 11beta-hydroxysteroid dehydrogenase type 1 gene. (PMID: 19571556) Ku YH . Park KS Hormone research 2009 3 23 43 56 [Possible pathogenetic role of 11 beta-hydroxysteroid dehydrogenase type 1

HSPD1: Fang Y, Xie T, Xue N, et al. miR-382 Contributes to Renal Tubulointerstitial Fibrosis by Downregulating HSPD1. Oxidative Medicine and Cellular Longevity. 2017;2017:4708516. doi:10.1155/2017/4708516.

HSPE1: Tsai C-H, Chen Y-T, Chang Y-H, et al. Systematic verification of bladder cancer-associated tissue protein biomarker candidates in clinical urine specimens. Oncotarget. 2018;9(56):30731-30747. doi:10.18632/oncotarget.24578.

HTR1B: Guo J, Zhang W, Zhang L, et al. Probable involvement of p11 with interferon alpha induced depression. Scientific Reports. 2016;6:17029. doi:10.1038/srep17029.

HTR2A: Polymorphisms in GRIK4, HTR2A, and FKBP5 show interactive effects in predicting remission to antidepressant treatment. (PMID: 19924111) Horstmann S . Binder EB Neuropsychopharmacology : official publication of the American College of Neuropsychopharmacology 2010 3 23 45 58 A genetic variant of HTR2C may play a role in the manifestation of Tourette syndrome. (PMID: 20010450) Dehning S . Zill P Psychiatric genetics 2010 3 23 45 58 Comorbidity between bipolar disorder and alcohol use disorder:

HTR2C: Yu W, Xu H, Xue Y, et al. 5-HT 2C R antagonist/5-HT 2C R inverse agonist recovered the increased isolation-induced aggressive behavior of BALB/c mice mediated by ADAR1 (p110) expression and Htr2c RNA editing. Brain and Behavior. 2018;8(3):e00929. doi:10.1

HTRA1: Ikawati M, Kawaichi M, Oka C. Loss of HtrA1 serine Proteínase induces synthetic modulation of aortic vascular smooth muscle cells. Bader M, ed. PLoS ONE. 2018;13(5):e0196628. doi:10.1371/journal.pone.0196628.

HYAL1: McAtee CO, Berkebile AR, Elowsky CG, et al. Hyaluronidase Hyal1 Increases Tumor Cell Proliferation and Motility through Accelerated Vesicle Trafficking. The Journal of Biological Chemistry. 2015;290(21):13144-13156. doi:10.1074/jbc.M115.647446.

ICAM1: Salvador AM, Nevers T, Velázquez F, et al. Intercellular Adhesion Molecule 1 Regulates Left Ventricular Leukocyte Infiltration, Cardiac Remodeling, and Function in Pressure Overload–Induced Heart Failure. Journal of the American Heart Association: Cardiov

IDE: Zhang Z, Liang WG, Bailey LJ, et al. Ensemble cryoEM elucidates the mechanism of insulin capture and degradation by human insulin degrading enzyme. Subramaniam S, ed. eLife. 2018;7:e33572. doi:10.7554/eLife.33572.

IFI16: Iqbal J, Ansari MA, Kumar B, et al. Histone H2B-IFI16 Recognition of Nuclear Herpesviral Genome Induces Cytoplasmic Interferon-? Responses. Moses AV, ed. PLoS Pathogens. 2016;12(10):e1005967. doi:10.1371/journal.ppat.1005967.

IFIH1: Asgari S, Schlapbach LJ, Anchisi S, et al. Severe viral respiratory infections in children with IFIH1 loss-of-function mutations. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(31):8342-8347. doi:10.1073/pnas.170

IFNG: Natan Stein, Orit Berhani, Dominik Schmiedel, Alexandra Duev-Cohen, Einat Seidel, Inbal Kol, Pinchas Tsukerman, Merav Hecht, Adi Reches, Moriya Gamliel, Akram Obeidat, Yoav Charpak-Amikam, Rachel Yamin, Ofer MandelboimiScience. 2019 Jan 25; 11: 466–473. Published online 2019 Jan 3. doi: 10.1016/j.isci.2018.12.034PMCID: PMC6354656Anand Balasubramani, Colleen J. Winstead, Henrietta Turner, Karen M. Janowski, Stacey N. Harbour, Yoichiro Shibata, Gregory E. Crawford, Robin D. Hatton, Casey T. W

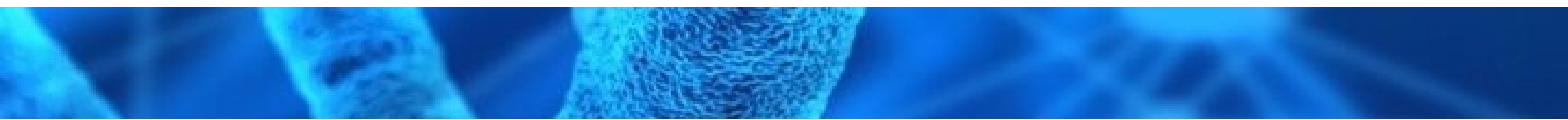
IFNGR1: Londino JD, Gulick DL, Lear TB, et al. Post-translational modification of the interferon-gamma receptor alters its stability and signaling. The Biochemical journal. 2017;474(20):3543-3557. doi:10.1042/BCJ20170548.

IFNGR2: Zhou H, Chen S, Qi Y, et al. Identification of Type II Interferon Receptors in Geese: Gene Structure, Phylogenetic Analysis, and Expression Patterns. BioMed Research International. 2015;2015:537637. doi:10.1155/2015/537637.

IGF1R: Lee H-T, Chang H-T, Lee S, et al. Role of IGF1R+ MSCs in modulating neuroplasticity via CXCR4 cross-interaction. Scientific Reports. 2016;6:32595. doi:10.1038/srep32595.

IGF2BP2: Wu L, Song W, Xie Y, et al. miR-181a-5p suppresses invasion and migration of HTR-8/SVneo cells by directly targeting IGF2BP2. Cell Death & Disease. 2018;9(2):16. doi:10.1038/s41419-017-0045-0.

IKBKE: Rajurkar M, Dang K, Fernandez-Barrena MG, et al. IKBKE is required during KRAS-induced pancreatic tumorigenesis. Cancer research. 2017;77(2):320-329. doi:10.1158/0008-5472.CAN-15-1684.



IKZF3: Human aiolos, an ikaros-related zinc finger DNA binding protein: cDNA cloning, tissue expression pattern, and chromosomal mapping. (PMID: 10552935) Hosokawa Y ... Seto M Genomics 1999 2 3 4 23 54 Combinatorial effects of splice variants modulate function of Aiolos. (PMID: 17646674) Caballero R ... Ballestar E Journal of cell science 2007 3 4 23 54 Both normal and leukemic B lymphocytes express multiple isoforms of the human Aiolos gene. (PMID: 11745366) Liippo J ... Lassila O European journal of immunology 2001 3 4 23 54 Eos and pegasus, two members of the Ikaros family of proteins with distinct DNA binding activities. (PMID: 10978333) Perdomo J ... Crossley M The Journal of biological chemistry 2000 3 4 23 54 Aiolos transcription factor controls cell death in T cells by regulating Bcl-2 expression and its cellular localization. (PMID: 10369681) Romero F ... Rebollo A The EMBO journal 1999

IL-10: Guillot-Sestier M-V, Doty KR, Gate D, et al. Il10 deficiency re-balances innate immunity to mitigate Alzheimer-like pathology. Neuron. 2015;85(3):534-548. doi:10.1016/j.neuron.2014.12.068.

IL-12B: Unger A, Finkernagel F, Hoffmann N, et al. Chromatin Binding of c-REL and p65 Is Not Limiting for Macrophage IL12B Transcription During Immediate Suppression by Ovarian Carcinoma Ascites. Frontiers in Immunology. 2018;9:1425. doi:10.3389/fimmu.2018.01425.

IL-12RB1: Turner AJ, Aggarwal P, Miller HE, et al. The introduction of RNA-DNA differences underlies interindividual variation in the human IL12RB1 mRNA repertoire. Proceedings of the National Academy of Sciences of the United States of America. 2015;112(50):15414-

IL-13: Le Blon D, Guglielmetti C, Hoornaert C, et al. Intracerebral transplantation of interleukin 13-producing mesenchymal stem cells limits microgliosis, oligodendrocyte loss and demyelination in the cuprizone mouse model. Journal of Neuroinflammation. 2016;13

IL-17F: Omrane I, Medimegh I, Baroudi O, et al. Involvement of IL17A, IL17F and IL23R Polymorphisms in Colorectal Cancer Therapy. Lim M, ed. PLoS ONE. 2015;10(6):e0128911. doi:10.1371/journal.pone.0128911.

IL-18RAP: Martínez-Barquero V, de Marco G, Martínez-Hervas S, et al. Are IL18RAPgene polymorphisms associated with body mass regulation? A cross-sectional study. BMJ Open. 2017;7(11):e017875. doi:10.1136/bmjopen-2017-017875.

IL-1A: Su H, Rei N, Zhang L, Cheng J. Meta-analyses of IL1A polymorphisms and the risk of several autoimmune diseases published in databases. Ahuja SK, ed. PLoS ONE. 2018;13(6):e0198693. doi:10.1371/journal.pone.0198693.

IL-1B: Hasegawa T, Hall CJ, Crosier PS, et al. Transient inflammatory response mediated by interleukin-1? is required for proper regeneration in zebrafish fin fold. Stainier DY, ed. eLife. 2017;6:e22716. doi:10.7554/eLife.22716.

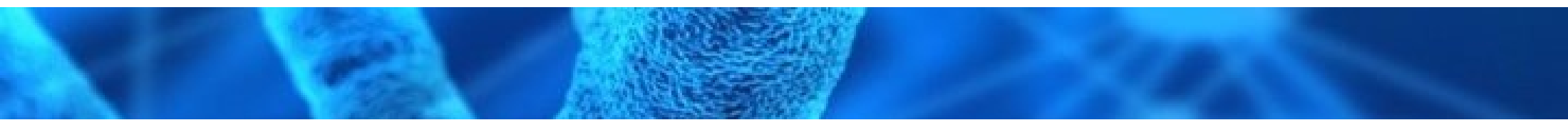
IL-1RL1: Díaz-Jiménez D, Núñez L, De la Fuente M, et al. A functional IL1RL1 variant regulates corticosteroid-induced sST2 expression in ulcerative colitis. Scientific Reports. 2017;7:10180. doi:10.1038/s41598-017-10465-0.

IL-23R: Sivanesan D, Beauchamp C, Quinou C, et al. IL23R (Interleukin 23 Receptor) Variants Protective against Inflammatory Bowel Diseases (IBD) Display Loss of Function due to Impaired Protein Stability and Intracellular Trafficking. The Journal of Biological Ch

IL-2RA: Li L, Yang S-H, Yao Y, et al. Block of both TGF-? and IL-2 signaling impedes Neurophilin-1+ regulatory T cell and follicular regulatory T cell development. Cell Death & Disease. 2016;7(10):e2439-. doi:10.1038/cddis.2016.348.

IL-31: Takamori A, Nambu A, Sato K, et al. IL-31 is crucial for induction of pruritus, but not inflammation, in contact hypersensitivity. Scientific Reports. 2018;8:6639. doi:10.1038/s41598-018-25094-4.

IL-4: Hühner L, Rilka J, Gilsbach R, Zhou X, Machado V, Spittau B. Interleukin-4 Protects Dopaminergic Neurons In vitro but Is Dispensable for MPTP-Induced Neurodegeneration In vivo. Frontiers in Molecular Neuroscience. 2017;10:62. doi:10.3389/fnmol.2017.00062.



IL-6: Huang Q, Zhang Z, Liao Y, et al. 17?-estradiol upregulates IL6 expression through the ER, pathway to promote lung adenocarcinoma progression. Journal of Experimental & Clinical Cancer Research?: CR. 2018;37:133. doi:10.1186/s13046-018-0804-5.

IL-6R: terleukin-6 receptor gene, plasma C-reactive protein, and diabetes risk in women. (PMID: 18852330) Qi L . Hu FB Diabetes 2009 3 23 43 56 IL-6 receptor, IL-8 receptor and TNF-alpha238 (G/A) polymorphisms are not associated with Behçet's disease in patients of German or Turkish origin. (PMID: 19026125) Storz K . Kötter I Clinical and experimental rheumatology 2008 3 23 43 56 Interleukin-6 (IL-6) and receptor (IL6-R) gene haplotypes associate with amniotic fluid protein concentrations in prete

IL-7R: Gaddiel Galarza-Muñoz, Farren B.S. Briggs, Irina Evsyukova, Geraldine Schott-Lerner, Edward M. Kennedy, Tinashe Nyanhete, Liuyang Wang, Laura Bergamaschi, Steven G. Widen, Georgia D. Tomaras, Dennis C. Ko, Shelton S. Bradrick, Lisa F. Barcellos, Simon G. Gregory, Mariano A. Garcia-BlancoCell. Author manuscript; available in PMC 2018 Mar 23.Published in final edited form as: Cell. 2017 Mar 23; 169(1): 72–84.e13. doi: 10.1016/j.cell.2017.03.007PMCID: PMC5456452 Shijiao Cai, Yang Chen, Yue Sha

IL2RA: Large-scale genetic fine mapping and genotype-phenotype associations implicate polymorphism in the IL2RA region in type 1 diabetes. (PMID: 17676041) Lowe CE . Todd JA Nature genetics 2007 2 3 4 43 56 Genetic association between the interleukin-2 receptor-alpha gene and mode of onset of type 1 diabetes in the Japanese population. (PMID: 19106270) Kawasaki E . Japanese Study Group on Type 1 Diabetes Genetics The Journal of clinical endocrinology and metabolism 2009 3 23 43 56 Human IL2RA null

IL4R: The polymorphisms S503P and Q576R in the interleukin-4 receptor alpha gene are associated with atopy and influence the signal transduction. (PMID: 10233717) Kruse S ... Deichmann KA Immunology 1999 3 4 21 39 70 The association of atopy with a gain-of-function mutation in the alpha subunit of the interleukin-4 receptor. (PMID: 9392697) Hershey GK ... Chatila TA The New England journal of medicine 1997 3 4 21 39 70 Effects of common atopy-associated amino acid substitutions in the IL-4 receptor alpha chain on IL-4 induced phenotypes. (PMID: 15712015) Franjkovic I ... Bein G Immunogenetics 2005 3 21 39 70 Interleukin-4/interleukin-4 receptor gene polymorphisms in hand osteoarthritis. (PMID: 20219689) Vargiolu M ... Meliconi R Osteoarthritis and cartilage 2010 3 21 39 Cytokine polymorphisms and Alzheimer disease: possible associations. (PMID: 20213229) Ribizzi G ... Megna M Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology 2010 Interleukin-4/interleukin-4 receptor gene polymorphisms in hand osteoarthritis. (PMID: 20219689) Vargiolu M ... Meliconi R Osteoarthritis and cartilage 2010 3 21 39 Cytokine polymorphisms and Alzheimer disease: possible associations. (PMID: 20213229) Ribizzi G ... Megna M Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology 2010 3 21 39 Involvement of IL-4, IL-13 and Their Receptors in Pancreatic Cancer. (PMID: 33804263) Shi J ... Kornmann M International journal of molecular sciences 2021 3 Detection of association of IL1β, IL4R, and IL6 gene polymorphisms with cervical cancer in the Bangladeshi women by tetra-primer ARMS-PCR method. (PMID: 33187912) Muhammad SB ... Safiqul Islam M International immunopharmacology 2021 3 IL4RA gene expression in relation to I50V, Q551R and C-3223T polymorphisms. (PMID: 33529503) Danielewicz H ... Boznański A Advances in clinical and experimental medicine : official organ Wroclaw Medical University 2021

INSIG2: Prakash J, Mittal B, Srivastava A, Awasthi S, Srivastava P, Srivastava N. Common Genetic Variant of INSIG2 Gene rs7566605 Polymorphism Is Associated with Severe Obesity in North India. Iranian Biomedical Journal. 2017;21(4):261-269. doi:10.18869/acadpub.i

INTERGENIC: Tsai C-H, Liao R, Chou B, Palumbo M, Contreras LM. Genome-Wide Analyses in Bacteria Show Small-RNA Enrichment for Long and Conserved Intergenic Regions. Zhulin IB, ed. Journal of Bacteriology. 2015;197(1):40-50. doi:10.1128/JB.02359-14.

IRF4: Guérin A, Kerner G, Marr N, et al. IRF4 haploinsufficiency in a family with Whipple’s disease. van der Meer JW, ed. eLife. 2018;7:e32340. doi:10.7554/eLife.32340.

IRF5: De S, Zhang B, Shih T, et al. B Cell-Intrinsic Role for IRF5 in TLR9/BCR-Induced Human B Cell Activation, Proliferation, and Plasmablast Differentiation. Frontiers in Immunology. 2017;8:1938. doi:10.3389/fimmu.2017.01938.

IRS1: Insulin receptor substrate 1 gene variation modifies insulin resistance response to weight-loss diets in a 2-year randomized trial: the Preventing Overweight Using Novel Dietary Strategies (POUNDS LOST) trial. Qi Q1, Bray GA, Smith SR, Hu FB, Sacks FM, Qi L.

ITGA11: Ruchi Bansal, Shigeki Nakagawa, Saleh Yazdani, Joop van Baarlen, Anu Venkatesh, Anna P Koh, Won-Min Song, Nicolas Goossens, Hideo Watanabe, Mary B Beasley, Charles A Powell, Gert Storm, Naftali Kaminski, Harry van Goor, Scott L Friedman, Yujin Hoshida, Jai PrakashExp Mol Med. 2017 Nov; 49(11): e396. Published online 2017 Nov 17. doi: 10.1038/emm.2017.213PMCID: PMC5704196 Vincent Grassot, Anne Da Silva, James Saliba, Abderrahman Maftah, Fabrice Dupuy, Jean-Michel Petit BMC Genomics.

ITGA2: Chuang Y-C, Wu H-Y, Lin Y-L, et al. Blockade of ITGA2 Induces Apoptosis and Inhibits Cell Migration in Gastric Cancer. Biological Procedures Online. 2018;20:10. doi:10.1186/s12575-018-0073-x.

ITGA6: Jin Y-P, Hu Y-P, Wu X-S, et al. miR-143-3p targeting of ITGA6 suppresses tumour growth and angiogenesis by downregulating PLGF expression via the PI3K/AKT pathway in gallbladder carcinoma. Cell Death & Disease. 2018;9(2):182. doi:10.1038/s41419-017-0258-2

ITGB3: Nemlich Y, Baruch EN, Besser MJ, et al. ADAR1-mediated regulation of melanoma invasion. Nature Communications. 2018;9:2154. doi:10.1038/s41467-018-04600-2.

JAG1: Stephania Macchiarulo, Bernice E. MorrowBiol Open. 2017 Oct 15; 6(10): 1472–1482. Published online 2017 Aug 24. doi: 10.1242/bio.027359PMCID: PMC5665468Marcia Bellon, Ramona Moles, Hassiba Chaib-Mezrag, Joanna Pancewicz, Christophe NicotJ Hematol Oncol. 2018; 11: 119. Published online 2018 Sep 19. doi: 10.1186/s13045-018-0665-6PMCID: PMC6146899 Wen-Hsin Chang, Bing-Ching Ho, Yi-Jing Hsiao, Jin-Shing Chen, Chien-Hung Yeh, Hsuan-Yu Chen, Gee-Chen Chang, Kang-Yi Su, Sung-Liang YuPLoS One. 2016

JAZF1: Sung Y, Park S, Park SJ, et al. Jazf1 promotes prostate cancer progression by activating JNK/Slug. Oncotarget. 2018;9(1):755-765. doi:10.18632/oncotarget.23146.

JDP2: Marc R. Mansour, Shuning He, Zhaodong Li, Riadh Lobbardi, Brian J. Abraham, Clemens Hug, Sunniyat Rahman, Theresa E. Leon, You-Yi Kuang, Mark W. Zimmerman, Traci Blonquist, Evisa Gjini, Alejandro Gutierrez, Qin Tang, Laura Garcia-Perez, Karin Pike-Overzet, Lars Anders, Alla Berezovskaya, Yi Zhou, Leonard I. Zon, Donna Neuberg, Adele K. Fielding, Frank J.T. Staal, David M. Langenau, Takaomi Sanda, Richard A. Young, A. Thomas LookJ Exp Med. 2018 Jul 2; 215(7): 1929–1945. doi: 10.1084/jem.2017

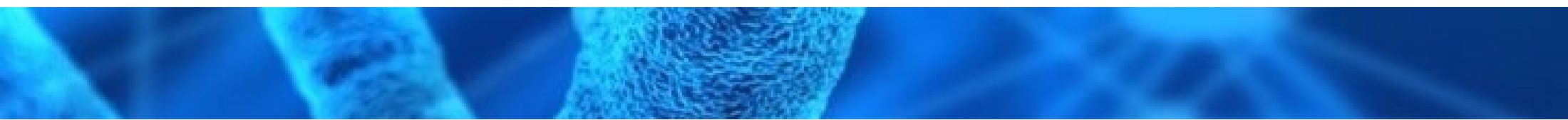
JMJD1C: A genome- and phenome-wide association study to identify genetic variants influencing platelet count and volume and their pleiotropic effects. Shameer K1, Denny JC, Ding K, Jouni H, Crosslin DR, de Andrade M, Chute CG, Peissig P, Pacheco JA, Li R, Bastarache L, Kho AN, Ritchie MD, Masys DR, Chisholm RL, Larson EB, McCarty CA, Roden DM, Jarvik GP, Kullo IJ.

KCNA5: Zhang F, Bian Y, Huang L, Fan W. Association between connexin 40 and potassium voltage-gated channel subfamily A member 5 expression in the atrial myocytes of patients with atrial fibrillation. Experimental and Therapeutic Medicine. 2017;14(5):5170-5176.

KCNB1: Wei Y, Shin MR, Sesti F. Oxidation of KCNB1 channels in the human brain and in mouse model of Alzheimer's disease. Cell Death & Disease. 2018;9(8):820. doi:10.1038/s41419-018-0886-1.

KCND2: Identification of Genes Promoting Skin Youthfulness by Genome-Wide Association Study, Anne L.S. Chang, Gil Atzmon, Aviv Bergman, Samantha Brugmann, Scott X. Atwood, Howard Y. Chang and Nir Barzilai, Characterization of human Kv4.2 mediating a rapidly-inactivating transient voltage-sensitive K+ current. (PMID: 10551270) Zhu XR . Pongs O Receptors & channels 1999 Voltage-dependent K+ channel acts as sex steroid sensor in endocrine cells of the human ovary. (PMID: 15991246) Kunz L . Mayerhofer

KCNE1: Barro-Soria R, Ramentol R, Liin SI, Perez ME, Kass RS, Larsson HP. KCNE1 and KCNE3 modulate KCNQ1 channels by affecting different gating transitions. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(35):E7367-E7376



KCNE2: Hu Z, Crump SM, Zhang P, Abbott GW. Kcne2 deletion attenuates acute post-ischaemia/reperfusion myocardial infarction. Cardiovascular Research. 2016;110(2):227-237. doi:10.1093/cvr/cvw048.

KCNE3: Barro-Soria R, Ramentol R, Liin SI, Perez ME, Kass RS, Larsson HP. KCNE1 and KCNE3 modulate KCNQ1 channels by affecting different gating transitions. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(35):E7367-E7376

KCNJ11: Zhang B, Novitskaya T, Wheeler DG, et al. Kcnj11 Ablation is Associated with Increased Nitro-Oxidative Stress During Ischemia-Reperfusion Injury: Implications for Human Ischemic Cardiomyopathy. Circulation Heart failure. 2017;10(2):e003523. doi:10.1161/CI

KCNMB2: Molecular basis of fast inactivation in voltage and Ca2+-activated K+ channels: a transmembrane beta-subunit homolog. (PMID: 10097176) Wallner M . Toro L Proceedings of the National Academy of Sciences of the United States of America 1999 2 3 4 56 Germline genomic variants associated with childhood acute lymphoblastic leukemia. (PMID: 19684603) Treviño LR . Relling MV Nature genetics 2009 3 43 56 The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Coll

KCNQ1: Barro-Soria R, Ramentol R, Liin SI, Perez ME, Kass RS, Larsson HP. KCNE1 and KCNE3 modulate KCNQ1 channels by affecting different gating transitions. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(35):E7367-E7376

KIAA1211: Zeltner N, Fattahi F, Dubois NC, et al. Capturing the biology of mild versus severe disease in a pluripotent stem cell-based model of famílial Dysautonomia. Nature medicine. 2016;22(12):1421-1427. doi:10.1038/nm.4220.

KIF6: Angelini S, Rosticci M, Massimo G, et al. Relationship between Lipid Phenotypes, Overweight, Lipid Lowering Drug Response and KIF6 and HMG-CoA Genotypes in a Subset of the Brisighella Heart Study Population. International Journal of Molecular Sciences. 20

KIRREL: Zhang M-J, Hong Y-Y, Li N. Overexpression of Kin of IRRE-Like Protein 1 (KIRREL) in Gastric Cancer and Its Clinical Prognostic Significance. Medical Science Monitor?: International Medical Journal of Experimental and Clinical Research. 2018;24:2711-2719.

KL: Rangiani A, Cao Z, Sun Y, et al. Protective Roles of DMP1 in High Phosphate Homeostasis. Mohanraj R, ed. PLoS ONE. 2012;7(8):e42329. doi:10.1371/journal.pone.0042329.

KLC1: Chiba K, Chien K, Sobu Y, et al. Phosphorylation of KLC1 modifies interaction with JIP1 and abolishes the enhanced fast velocity of APP transport by kinesin-1. Kaibuchi K, ed. Molecular Biology of the Cell. 2017;28(26):3857-3869. doi:10.1091/mbc.E17-05-03

KLF6: Zhang Y, Lei C-Q, Hu Y-H, et al. Krüppel-like Factor 6 Is a Co-activator of NF-?B That Mediates p65-dependent Transcription of Selected Downstream Genes. The Journal of Biological Chemistry. 2014;289(18):12876-12885. doi:10.1074/jbc.M113.535831.

KLKB1: Genetic variants associated with deep vein thrombosis: the F11 locus. (PMID: 19583818) Li Y ... Rosendaal FR Journal of thrombosis and haemostasis : JTH 2009 3 23 41 54 Common variation in KLKB1 and essential hypertension risk: tagging-SNP haplotype analysis in a case-control study. (PMID: 17318641) Lu X ... Gu D Human genetics 2007 3 23 41 54 Genomic structure of the human plasma prekallikrein gene, identification of allelic variants, and analysis in end-stage renal disease. (PMID: 11031105) Yu H ... Bowden DW Genomics 2000 3 4 23 54 Identification of human plasma kallikrein gene polymorphisms and evaluation of their role in end-stage renal disease. (PMID: 9535413) Yu H ... Freedman BI Hypertension (Dallas, Tex. : 1979) 1998 2 3 23 54 Location of the disulfide bonds in human plasma prekallikrein: the presence of four novel apple domains in the amino-terminal portion of the molecule. (PMID: 1998666) McMullen BA ... Davie EW Biochemistry 1991

KLRB1: Rother S, Hundrieser J, Pokoyski C, et al. The c.503T>C Polymorphism in the Human KLRB1 Gene Alters Ligand Binding and Inhibitory Potential of CD161 Molecules. Vivier E, ed. PLoS ONE. 2015;10(8):e0135682. doi:10.1371/journal.pone.0135682.

KLRK1: Sheppard S, Guedes J, Mroz A, et al. The immunoreceptor NKG2D promotes tumour growth in a model of hepatocellular carcinoma. Nature Communications. 2017;8:13930. doi:10.1038/ncomms13930.

KRT5: Ray S, Chiba N, Yao C, et al. Rare SOX2+ Airway Progenitor Cells Generate KRT5+ Cells that Repopulate Damaged Alveolar Parenchyma following Influenza Virus Infection. Stem Cell Reports. 2016;7(5):817-825. doi:10.1016/j.stemcr.2016.09.010.

KRT75: A novel human type II cytokeratin, K6hf, specifically expressed in the companion layer of the hair follicle. (PMID: 9856802) Winter H ... Schweizer J The Journal of investigative dermatology 1998 2 3 4 21 An unusual Ala12Thr polymorphism in the 1A alpha-helical segment of the companion layer-specific keratin K6hf: evidence for a risk factor in the etiology of the common hair disorder pseudofolliculitis barbae. (PMID: 15086549) Winter H ... Schweizer J The Journal of investigative dermatology 2004 3 4 70 Is the loose anagen hair syndrome a keratin disorder? A clinical and molecular study. (PMID: 11939812) Chapalain V ... Taïeb A Archives of dermatology 2002 3 4 21 Characterization of a 300 kbp region of human DNA containing the type II hair keratin gene domain. (PMID: 10692104) Rogers MA ... Schweizer J The Journal of investigative dermatology 2000 2 3 4 Cytokeratin 75 expression in central, centrifugal, cicatricial alopecia--new observations in normal and diseased hair follicles. (PMID: 19614992) Sperling LC ... Darling T Journal of cutaneous pathology 2010

LAG3: Andrews LP, Marciscano AE, Drake CG, Vignali DAA. LAG3 (CD223) as a Cancer Immunotherapy Target. Immunological reviews. 2017;276(1):80-96. doi:10.1111/imr.12519.

LAMA3: Li R, Ochs MF, Ahn SM, et al. Expression Microarray Analysis Reveals Alternative Splicing of LAMA3 and DST Genes in Head and Neck Squamous Cell Carcinoma. Dias-Neto E, ed. PLoS ONE. 2014;9(3):e91263. doi:10.1371/journal.pone.0091263.

LAMB3: Seung-Nam Jung, Hyun Sil Lim, Lihua Liu, Jae Won Chang, Young Chang Lim, Ki Sang Rha, Bon Seok KooSci Rep. 2018; 8: 2718. Published online 2018 Feb 9. doi: 10.1038/s41598-018-21216-0PMCID: PMC5807368Yinghao Wang, Yixiang Jin, Adheesh Bhandari, Zhihan Yao, Fan Yang, Yiyuan Pan, Zhouci Zheng, Shixu Lv, Ouchen WangOnco Targets Ther. 2018; 11: 37–46. Published online 2017 Dec 21. doi: 10.2147/OTT.S149613PMCID: PMC5743181Ying Zhou, Jason Dang, Kung-Yen Chang, Edwin Yau, Pedro Aza-Blanc, Jorge Mo

LCT: Fumery M, Speca S, Langlois A, et al. Peroxisome proliferator-activated receptor gamma (PPAR?) regulates lactase expression and activity in the gut. EMBO Molecular Medicine. 2017;9(11):1471-1481. doi:10.15252/emmm.201707795.

LDLR: Poirier S, Hamouda HA, Villeneuve L, Demers A, Mayer G. Trafficking Dynamics of PCSK9-Induced LDLR Degradation: Focus on Human PCSK9 Mutations and C-Terminal Domain. Kanzaki M, ed. PLoS ONE. 2016;11(6):e0157230. doi:10.1371/journal.pone.0157230.

LEP: Silva BJ de A, Barbosa MG de M, Andrade PR, et al. Autophagy Is an Innate Mechanism Associated with Leprosy Polarization. Salgame P, ed. PLoS Pathogens. 2017;13(1):e1006103. doi:10.1371/journal.ppat.1006103.

LEPR: Yang Y, Niu T. A meta-analysis of associations of LEPR Q223R and K109R polymorphisms with Type 2 diabetes risk. Amendola R, ed. PLoS ONE. 2018;13(1):e0189366. doi:10.1371/journal.pone.0189366.

LGALS17A: El-Diwany R, Soliman M, Sugawara S, et al. CMPK2 and BCL-G are associated with type 1 interferon–induced HIV restriction in humans. Science Advances. 2018;4(8):eaat0843. doi:10.1126/sciadv.aat0843.

LHPP: Polimanti R, Wang Q, Meda SA, et al. The Interplay Between Risky Sexual Behaviors and Alcohol Dependence: Genome-Wide Association and Neuroimaging Support for LHPP as a Risk Gene. Neuropsychopharmacology. 2017;42(3):598-605. doi:10.1038/npp.2016.153.

LINC00704: Lu W, Xu Y, Xu J, Wang Z, Ye G. Identification of differential expressed lncRNAs in human thyroid cancer by a genome-wide analyses. Cancer Medicine. 2018;7(8):3935-3944. doi:10.1002/cam4.1627.

LINC01299: Novel genetic loci identified for the pathophysiology of childhood obesity in the Hispanic population. (PMID: 23251661) Comuzzie AG ... Butte NF PloS one 2012 3 Diversification of transcriptional modulation: large-scale identification and characterization of putative alternative promoters of human genes. (PMID: 16344560) Kimura K ... Sugano S Genome research 2006 3 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004

LINC01432: Iván Galván-Femenía, Mireia Obón-Santacana, David Piñeyro, Marta Guindo-Martinez, Xavier Duran, Anna Carreras, Raquel Pluvinet, Juan Velasco, Laia Ramos, Susanna Aussó, J M Mercader, Lluis Puig, Manuel Perucho, David Torrents, Victor Moreno, Lauro Sumoy, Rafael de CidJ Med Genet. 2018 Nov; 55(11): 765–778. Published online 2018 Aug 30. doi: 10.1136/jmedgenet-2018-105437PMCID: PMC6252362Kaustubh Adhikari, Macarena Fuentes-Guajardo, Mirsha Quinto-Sánchez, Javier Mendoza-Revilla, Juan Camilo C

LINC01500: Tatsushi Okayama, Yasuyuki Hashiguchi, Hiroki Kikuyama, Hiroshi Yoneda, Tetsufumi KanazawaTransl Psychiatry. 2018; 8: 221. Published online 2018 Oct 15. doi: 10.1038/s41398-018-0272-xPMCID: PMC6189064

LIPA: Dong L, Feng R, Zhang L, et al. Prospective comparison of hybrid capture 2 and SPF10-LiPA for carcinogenic human papillomavirus detection and risk prediction of cervical cancer: a population-based cohort study in China. Journal of Gynecologic Oncology. 20

LIPC: Wang Y, Wang M, Zhang X, et al. The Association between LIPC rs493258 Polymorphism and the Susceptibility to Age-Related Macular Degeneration. Tchounwou PB, ed. International Journal of Environmental Research and Public Health. 2016;13(10):1022. doi:10.33

LMNA: Janin A, Bauer D, Ratti F, et al. SMAD6 overexpression leads to accelerated myogenic differentiation of LMNA mutated cells. Scientific Reports. 2018;8:5618. doi:10.1038/s41598-018-23918-x.

LOX: Molecular cloning of human lysyl oxidase and assignment of the gene to chromosome 5q23.3-31.2. (PMID: 1685472) Hämäläinen ER . Kivirikko KI Genomics 1991 2 3 4 23 56 Association between lysyl oxidase polymorphisms and oral submucous fibrosis in older male areca chewers. (PMID: 18764858) Shieh TM . Liu CJ Journal of oral pathology & medicine : official publication of the International Association of Oral Pathologists and the American Academy of Oral Pathology 2009 3 23 43 56 Epigenetic inhib

LOXHD1: TANG H, ZHANG W, YAN X-M, et al. Analysis of SLC4A11, ZEB1, LOXHD1, COL8A2 and TCF4 gene sequences in a multi-generational family with late-onset Fuchs corneal dystrophy. International Journal of Molecular Medicine. 2016;37(6):1487-1500. doi:10.3892/ijmm.

LPL: Additive effects of LPL, APOA5 and APOE variant combinations on triglyceride levels and hypertriglyceridemia: results of the ICARIA genetic sub-study. Ariza MJ1, Sánchez-Chaparro MA, Barón FJ, Hornos AM, Calvo-Bonacho E, Rioja J, Valdivielso P, Gelpi JA, González-Santos P. Genetic-epidemiological evidence on genes associated with HDL cholesterol levels: a systematic in-depth review. Boes E1, Coassin S, Kollerits B, Heid IM, Kronenberg F.

LPP: Elaine Ngan, Konstantin Stoletov, Harvey W. Smith, Jessica Common, William J. Muller, John D. Lewis, Peter M. SiegelNat Commun. 2017; 8: 15059. Published online 2017 Apr 24. doi: 10.1038/ncomms15059PMCID: PMC5413977 Cecilia S. Leung, Tsz-Lun Yeung, Kay-Pong Yip, Kwong-Kwok Wong, Samuel Y. Ho, Lingegowda S. Mangala, Anil K. Sood, Gabriel Lopez-Berestein, Jianting Sheng, Stephen T.C. Wong, Michael J. Birrer, Samuel C. MokJ Clin Invest. 2018 Feb 1; 128(2): 589–606. Published online 2017 Dec 18

LRP6: Wang L, Chai Y, Li C, et al. Oxidized phospholipids are ligands for LRP6. Bone Research. 2018;6:22. doi:10.1038/s41413-018-0023-x.

LRP8: Waltmann MD, Basford JE, Konaniah ES, Weintraub NL, Hui DY. Apolipoprotein E receptor-2 deficiency enhances macrophage susceptibility to lipid accumulation and cell death to augment atherosclerotic plaque progression and necrosis. Biochimica et biophysica

LRRC3C: DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. (PMID: 16625196) Zody MC ... Nusbaum C Nature 2006 3 4 54 Gene Expression-Genotype Analysis Implicates GSDMA, GSDMB, and LRRC3C as Contributors to Inflammatory Bowel Disease Susceptibility. (PMID: 26484354) Söderman J ... Almer S BioMed research international 2015 3 54 Phylogenetic-based propagation of functional annotations within the Gene Ontology consortium. (PMID: 21873635) Gaudet P ... Thomas PD Briefings in bioinformatics 2011 3 54 A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. (PMID: 19820697) Soranzo N ... Gieger C Nature genetics 2009 3 54 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004

LTA: Kang S-S, Kim SK, Baik JE, et al. Staphylococcal LTA antagonizes the B cell-mitogenic potential of LPS. Scientific Reports. 2018;8:1496. doi:10.1038/s41598-018-19653-y.

LYPLAL1: Discovery of a Selective Covalent Inhibitor of Lysophospholipase-like 1 (LYPLAL1) as a Tool to Evaluate the Role of this Serine Hydrolase in Metabolism. Ahn K, Boehm M, Brown MF, Calloway J, Che Y, Chen J, Fennell KF, Geoghegan KF, Gilbert AM, Gutierrez JA, Kalgutkar AS, Lanba A, Limberakis C, Magee TV, O'Doherty I, Oliver R, Pabst B, Pandit J, Parris K, Pfefferkorn JA, Rolph TP, Patel R, Schuff B, Shanmugasundaram V, Starr JT, Varghese AH, Vera NB, Vernochet C, Yan J. Genetic predictors of

M6PR: Caster AH, Sztul E, Kahn RA. A Role for Cargo in Arf-dependent Adaptor Recruitment. The Journal of Biological Chemistry. 2013;288(21):14788-14804. doi:10.1074/jbc.M113.453621.

MACROD2: Van den Broek E, den Uil SH, Coupé VMH, et al. MACROD2 expression predicts response to 5-FU-based chemotherapy in stage III colon cancer. Oncotarget. 2018;9(50):29445-29452. doi:10.18632/oncotarget.25655.

MACROH2A2: Histone H2A variants and the inactive X chromosome: identification of a second macroH2A variant. (PMID: 11331621) Chadwick BP ... Willard HF Human molecular genetics 2001 2 3 4 21 MACROH2A2, a new member of the MARCOH2A core histone family. (PMID: 11262398) Costanzi C ... Pehrson JR The Journal of biological chemistry 2001 2 3 4 Formation of MacroH2A-containing senescence-associated heterochromatin foci and senescence driven by ASF1a and HIRA. (PMID: 15621527) Zhang R ... Adams PD Developmental cell 2005 3 4 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004 3 4 The DNA sequence and comparative analysis of human chromosome 10. (PMID: 15164054) Deloukas P ... Rogers J Nature 2004 3 https://www.ncbi.nlm.nih.gov/pubmed/29654602

MAF: Kim M, Wende H, Walcher J, et al. Maf links Neuregulin1 signaling to cholesterol synthesis in myelinating Schwann cells. Genes & Development. 2018;32(9-10):645-657. doi:10.1101/gad.310490.117.

MAGEC3: Eng KH, Szender JB, Etter JL, et al. Paternal lineage early onset hereditary ovarian cancers: A famílial Ovarian Cancer Registry study. Eng C, ed. PLoS Genetics. 2018;14(2):e1007194. doi:10.1371/journal.pgen.1007194.

MAGI2: Bierzynska A, Soderquest K, Dean P, et al. MAGI2 Mutations Cause Congenital Nephrotic Syndrome. Journal of the American Society of Nephrology?: JASN. 2017;28(5):1614-1621. doi:10.1681/ASN.2016040387.

MALT1: Kip E, Staal J, Verstrepen L, et al. MALT1 Controls Attenuated Rabies Virus by Inducing Early Inflammation and T Cell Activation in the Brain. Dutch RE, ed. Journal of Virology. 2018;92(8):e02029-17. doi:10.1128/JVI.02029-17.

Name Sample

Age | Gender F | Report date 12/09/2025

Prescriber | Health insurance

Sample

MAOA: Monoamine oxidase A gene (MAOA) predicts behavioral aggression following provocation. (PMID: 19168625) McDermott R ... Johnson DD Proceedings of the National Academy of Sciences of the United States of America 2009 3 21 39 69 A non-additive interaction of a functional MAO-A VNTR and testosterone predicts antisocial behavior. (PMID: 17429405) Sjöberg RL ... Goldman D Neuropsychopharmacology : official publication of the American College of Neuropsychopharmacology 2008 3 21 24 39 Monoamine oxidase-a genetic variations influence brain activity associated with inhibitory control: new insight into the neural correlates of impulsivity. (PMID: 16202396) Passamonti L ... Quattrone A Biological psychiatry 2006 3 21 39 69 20 ans après: a second mutation in MAOA identified by targeted high-throughput sequencing in a family with altered behavior and cognition. (PMID: 24169519) Piton A ... Mandel JL European journal of human genetics : EJHG 2014 3 4 69 Association of VNTR polymorphisms in the MAOA promoter and DRD4 exon 3 with heroin dependence in male Chinese addicts. (PMID: 20218801) Chien CC ... Lung FW The world journal of biological psychiatry : the official journal of the World Federation of Societies of Biological Psychiatry 2010 A proximity-dependent biotinylation map of a human cell. (PMID: 34079125) Go CD ... Gingras AC Nature 2021 3 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 MAOA alters the effects of heavy drinking and childhood physical abuse on risk for severe impulsive acts of violence among alcoholic violent offenders. (PMID: 20201935) Tikkanen R ... Virkkunen M Alcoholism, clinical and experimental research 2010 3 39 Does prior traumatization affect the treatment outcome of CBT for panic disorder? The potential role of the MAOA gene and depression symptoms. (PMID: 28712090) Trautmann S ... Reif A European archives of psychiatry and clinical neuroscience 2019 3 PLEKHA4/kramer Attenuates Dishevelled Ubiquitination to Modulate Wnt and Planar Cell Polarity Signaling. (PMID: 31091453) Shami Shah A ... Baskin JM Cell reports 2019 3 Moderator Effects of Life Stress on the Association between MAOA-uVNTR, Depression, and Burnout. (PMID: 30943524) Plieger T ... Reuter M Neuropsychobiology 2019 3 Role of monoamine-oxidase-A-gene variation in the development of glioblastoma in males: a case control study. (PMID: 31556016) Sjöberg RL ... Melin B Journal of neuro-oncology 2019 Serotonin, genetic variability, behaviour, and psychiatric disorders--a review. (PMID: 20187845) Nordquist N ... Oreland L Upsala journal of medical sciences 2010 3 21 An interaction between the norepinephrine transporter and monoamine oxidase A polymorphisms, and novelty-seeking personality traits in Korean females. (PMID: 17920180) Lee BC ... Ham BJ Progress in neuro-psychopharmacology & biological psychiatry 2008 3 21 The Altered Brain Activation of Phonological Working Memory, Dual Tasking, and Distraction Among Participants With Adult ADHD and the Effect of the MAOA Polymorphism. (PMID: 25777072) Ko CH ... Yen JY Journal of attention disorders 2018 3 Deficiency of Sustained Attention in ADHD and Its Potential Genetic Contributor MAOA. (PMID: 25784069) Liu L ... Wang Y Journal of attention disorders 2018 3 Histone Interaction Landscapes Visualized by Crosslinking Mass Spectrometry in Intact Cell Nuclei. (PMID: 30021884) Fasci D ... Heck AJR Molecular & cellular proteomics : MCP 2018

MAP2K1: Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. (PMID: 19156172) Dentici ML ... Dallapiccola B European journal of human genetics : EJHG 2009 3 22 40 71 Mutation and phenotypic spectrum in patients with cardio-facio-cutaneous and Costello syndrome. (PMID: 18042262) Schulz AL ... Zenker M Clinical genetics 2008 3 4 22 71 Germline mutations in genes within the MAPK pathway cause cardio-facio-cutaneous syndrome. (PMID: 16439621) Rodriguez-Viciana P ... Rauen KA Science (New York, N.Y.) 2006 3 4 22 71 Cloning and characterization of two distinct human extracellular signal-regulated kinase activator kinases, MEK1 and MEK2. (PMID: 8388392) Zheng CF ... Guan KL The Journal of biological chemistry 1993 2 3 4 22 Somatic activating mutations in MAP2K1 cause melorheostosis. (PMID: 29643386) Kang H ... Bhattacharyya T Nature communications 2018

MAT1A: Yuntao Bing, Siying Zhu, Guozheng Yu, Ting Li, Weijun Liu, Changsheng Li, Yitao Wang, Haolong Qi, Tao Guo, Yufeng Yuan, Yueming He, Zhisu Liu, Quanyan LiuJ Biol Chem. 2014 Nov 21; 289(47): 32639–32655. Published online 2014 Sep 30. doi: 10.1074/jbc.M114.589689PMCID: PMC4239617Heping Yang, Michele E. Cho, Tony W.H. Li, Hui Peng, Kwang Suk Ko, Jose M. Mato, Shelly C. LuJ Clin Invest. 2013 Jan 2; 123(1): 285–298. Published online 2012 Dec 17. doi: 10.1172/JCI63861PMCID: PMC3533284Komal Ramani,

MC1R: Chen S, Zhu B, Yin C, et al. Palmitoylation-dependent activation of MC1R prevents melanomagenesis. Nature. 2017;549(7672):399-403. doi:10.1038/nature23887.

MC4R: Giuseppe Bruschetta, Jung Dae Kim, Sabrina Diano, Li F. ChanMol Metab. 2018 Dec; 18: 79-87. Published online 2018 Oct 4. doi: 10.1016/j.molmet.2018.09.010PMCID: PMC6308034 Faith K. McDaniel, Brent M. Molden, Sameer Mohammad, Giovanna Baldini, Lakisha McPike, Paola Narducci, Susana Granell, Giulia BaldiniJ Biol Chem. 2012 Jun 22; 287(26): 21873-21890. Published online 2012 Apr 27. doi: 10.1074/jbc.M112.346890PMCID: PMC3381150 Brent M. Molden, Kimberly A. Cooney, Kirk West. Association of the

MCM6: Liu M, Hu Q, Tu M, et al. MCM6 promotes metastasis of hepatocellular carcinoma via MEK/ERK pathway and serves as a novel serum biomarker for early recurrence. Journal of Experimental & Clinical Cancer Research?: CR. 2018;37:10. doi:10.1186/s13046-017-0669

MCR1R: Radchenko V, Engle JW, Roy C, et al. 18th European Symposium on Radiopharmacy and Radiopharmaceuticals: Salzburg, Austria. 7-10 April 2016. Ejnmmi Radiopharmacy and Chemistry. 2016;1(Suppl 1):10. doi:10.1186/s41181-016-0012-6.

MCT1: Noor SI, Jamali S, Ames S, Langer S, Deitmer JW, Becker HM. A surface proton antenna in carbonic anhydrase II supports lactate transport in cancer cells. Vander Heiden MG, ed. eLife. 2018;7:e35176. doi:10.7554/eLife.35176.

MDFIC: Robert H. Oakley, John M. Busillo, John A. CidlowskiJ Biol Chem. 2017 Apr 7; 292(14): 5825–5844. Published online 2017 Feb 21. doi: 10.1074/jbc.M116.758888PMCID: PMC5392576 Faizan H. Khan, Vijayabaskar Pandian, Satishkumar Ramraj, Mohan Natarajan, Sheeja Aravindan, Terence S. Herman, Natarajan AravindanBMC Cancer. 2015; 15: 514. Published online 2015 Jul 10. doi: 10.1186/s12885-015-1463-yPMCID: PMC4496850Haiqing Ma, Desheng Weng, Yibing Chen, Wei Huang, Ke Pan, Hui Wang, Jiancong Sun.

MDM2: Raja R, Ronsard L, Lata S, Trivedi S, Banerjea AC. HIV-1 Tat potently stabilises Mdm2 and enhances viral replication. Biochemical Journal. 2017;474(14):2449-2464. doi:10.1042/BCJ20160825.

MEN1: Yuan Z, Claros CS, Suzuki M, et al. Loss of MEN1 activates DNMT1 implicating DNA hypermethylation as a driver of MEN1 tumorigenesis. Oncotarget. 2016;7(11):12633-12650. doi:10.18632/oncotarget.7279.

MERTK: Yukako Kayashima, Natalia Makhanova, Nobuyo MaedaArterioscler Thromb Vasc Biol. Author manuscript; available in PMC 2018 Jul 1.Published in final edited form as: Arterioscler Thromb Vasc Biol. 2017 Jul; 37(7): e82–e91. Published online 2017 May 4. doi: 10.1161/ATVBAHA.117.309522PMCID: PMC5497749 Khanh-Quynh N. Nguyen, Wen-I Tsou, Daniel A. Calarese, Stanley G. Kimani, Sukhwinder Singh, Shelly Hsieh, Yongzhang Liu, Bin Lu, Yi Wu, Scott J. Garforth, Steve C. Almo, Sergei V. Kotenko.

MLXIPL: Singh P, Irwin DM. Contrasting Patterns in the Evolution of Vertebrate MLX Interacting Protein (MLXIP) and MLX Interacting Protein-Like (MLXIPL) Genes. Bertrand S, ed. PLoS ONE. 2016;11(2):e0149682. doi:10.1371/journal.pone.0149682.

MME: Auer-Grumbach M, Toegel S, Schabhüttl M, et al. Rare Variants in MME, Encoding MetalloProteínase Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. American Journal of Human Genetics. 2016;99(3):607-623. doi:10.1016/j.ajhg.2016

MMP1: Depetris-Chauvin A, Fernández-Gamba Á, Gorostiza EA, Herrero A, Castaño EM, Ceriani MF. Mmp1 Processing of the PDF Neuropeptide Regulates Circadian Structural Plasticity of Pacemaker Neurons. Taghert PH, ed. PLoS Genetics. 2014;10(10):e1004700. doi:10.1371/journal.pgen.1004700.

MMP3: Wu Y, Lee M-J, Ido Y, Fried SK. High-fat diet-induced obesity regulates MMP3 to modulate depot- and sex-dependent adipose expansion in C57BL/6J mice. American Journal of Physiology - Endocrinology and Metabolism. 2017;312(1):E58-E71. doi:10.1152/ajpendo.0

MMP9: Appleby TC, Greenstein AE, Hung M, et al. Biochemical characterization and structure determination of a potent, selective antibody inhibitor of human MMP9. The Journal of Biological Chemistry. 2017;292(16):6810-6820. doi:10.1074/jbc.M116.760579.

MOV10: Skariah G, Seimetz J, Norsworthy M, et al. Mov10 suppresses retroelements and regulates neuronal development and function in the developing brain. BMC Biology. 2017;15:54. doi:10.1186/s12915-017-0387-1.

MPO: Pulli B, Wojtkiewicz G, Iwamoto Y, et al. Molecular MR Imaging of Myeloperoxidase Distinguishes Steatosis from Steatohepatitis in Nonalcoholic Fatty Liver Disease. Radiology. 2017;284(2):390-400. doi:10.1148/radiol.2017160588.

MPV17L2: Mai N, Chrzanowska-Lightowlers ZMA, Lightowlers RN. The process of mammalian mitochondrial protein synthesis. Cell and Tissue Research. 2017;367(1):5-20. doi:10.1007/s00441-016-2456-0.

MRAS: Higgins EM, Bos JM, Mason-Suares H, et al. Elucidation of MRAS-mediated Noonan syndrome with cardiac hypertrophy. JCI Insight. 2017;2(5):e91225. doi:10.1172/jci.insight.91225.

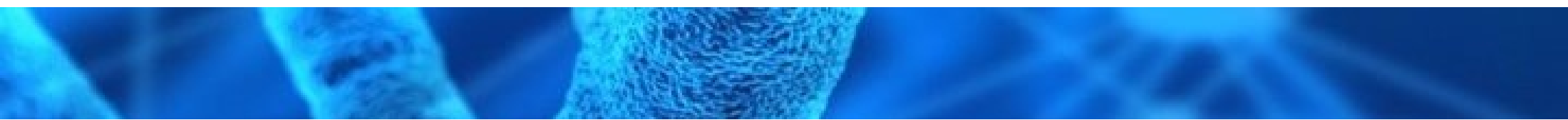
MS4A6A: Lacher SE, Alazizi A, Wang X, et al. A hypermorphic antioxidant response element is associated with increased MS4A6A expression and Alzheimer's disease. Redox Biology. 2018;14:686-693. doi:10.1016/j.redox.2017.10.018. Alzheimer's disease susceptibility variants in the MS4A6A gene are associated with altered levels of MS4A6A expression in blood. Proitsi P1, Lee SH, Lunnon K, Keohane A, Powell J, Troakes C, Al-Sarraj S, Furney S, Soininen H, Kloszewska I, Mecocci P, Tsolaki M, Vellas B, Loves

MSMB: Lou H, Li H, Yeager M, et al. Promoter variants in the MSMB gene associated with prostate cancer regulate MSMB/NCOA4 fusion transcripts. Human genetics. 2012;131(9):1453-1466. doi:10.1007/s00439-012-1182-2.

MSRA: Molecular cloning and functional expression of a human peptide methionine sulfoxide reductase (hMsrA). (PMID: 10452521) Kuschel L ... Heinemann SH FEBS letters 1999 2 3 4 23 Two new Loci for body-weight regulation identified in a joint analysis of genome-wide association studies for early-onset extreme obesity in French and german study groups. (PMID: 20421936) Scherag A ... Meyre D PLoS genetics 2010 3 23 41 Methionine sulfoxide reductase: a novel schizophrenia candidate gene. (PMID: 18506707) Walss-Bass C ... Raventos H American journal of medical genetics. Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics 2009 3 23 41 Gene structure, localization and role in oxidative stress of methionine sulfoxide reductase A (MSRA) in the monkey retina. (PMID: 16364291) Lee JW ... Rodriguez IR Experimental eye research 2006 3 4 23 Functioning methionine sulfoxide reductases A and B are present in human epidermal melanocytes in the cytosol and in the nucleus. (PMID: 16480945) Schallreuter KU ... Wood JM Biochemical and biophysical research communications 2006

MTHFD1: Kamynina E, Lachenauer ER, DiRisio AC, Liebenthal RP, Field MS, Stover PJ. Arsenic trioxide targets MTHFD1 and SUMO-dependent nuclear de novo thymidylate biosynthesis. Proceedings of the National Academy of Sciences of the United States of America. 2017;1

MTHFD1L: Lee D, Xu IM-J, Chiu DK-C, et al. Folate cycle enzyme MTHFD1L confers metabolic advantages in hepatocellular carcinoma. The Journal of Clinical Investigation. 2017;127(5):1856-1872. doi:10.1172/JCI90253.



MTHFR: Human methylenetetrahydrofolate reductase: isolation of cDNA, mapping and mutation identification. (PMID: 7920641) Goyette P ... Rozen R Nature genetics 1994 2 3 4 21 70 Systematic meta-analyses and field synopsis of genetic association studies in schizophrenia: the SzGene database. (PMID: 18583979) Allen NC ... Bertram L Nature genetics 2008 3 4 39 70 A common haplotype on methylenetetrahydrofolate reductase gene modifies the effect of angiotensin-converting enzyme inhibitor on blood pressure in essential hypertension patients--a family-based association study. (PMID: 16081343) Jiang S ... Xu X Clinical and experimental hypertension (New York, N.Y. : 1993) 2005 3 21 24 39 Methylenetetrahydrofolate reductase genotype, vitamin B12, and folate influence plasma homocysteine in hemodialysis patients. (PMID: 11979347) Nakamura T ... Hata A American journal of kidney diseases : the official journal of the National Kidney Foundation 2002 3 21 24 39 Polymorphisms in the methylenetetrahydrofolate reductase gene are associated with susceptibility to acute leukemia in adults. (PMID: 10536004) Skibola CF ... Morgan G Proceedings of the National Academy of Sciences of the United States of America 1999 Association of methylenetetrahydrofolate reductase (MTHFR 677C>T and 1298A>C) polymorphisms and haplotypes with silent brain infarction and homocysteine levels in a Korean population. (PMID: 20191019) Han IB ... Kim NK Yonsei medical journal 2010 3 21 39 MTHFR C677T and A1298C variant genotypes and the risk of microsatellite instability among Iranian colorectal cancer patients. (PMID: 20193847) Naghibalhossaini F ... Abdollahi K Cancer genetics and cytogenetics 2010 3 39 Polymorphism C776G in the transcobalamin II gene and homocysteine, folate and vitamin B12 concentrations. Association with MTHFR C677T and A1298C and MTRR A66G polymorphisms in healthy children. (PMID: 16820193) Aléssio AC ... Annichino-Bizzacchi JM Thrombosis research 2007 3 39 Hyperhomocysteinaemia and MTHFR C677T gene polymorphism in renal transplant recipients. (PMID: 11420199) Szabó AJ ... Reusz GS Archives of disease in childhood 2001 3 39 Food Intervention with Folate Reduces TNF-α and Interleukin Levels in Overweight and Obese Women with the MTHFR C677T Polymorphism: A Randomized Trial. (PMID: 32019154) Lisboa JVC ... Costa MJC Nutrients 2020

MTNR1B: G-allele of intronic rs10830963 in MTNR1B confers increased risk of impaired fasting glycemia and type 2 diabetes through an impaired glucose-stimulated insulin release: studies involving 19,605 Europeans. Sparsø T1, Bonnefond A, Andersson E, Bouatia-Naji N, Holmkvist J, Wegner L, Grarup N, Gjesing AP, Banasik K, Cavalcanti-Proença C, Marchand M, Vaxillaire M, Charpentier G, Jarvelin MR, Tichet J, Balkau B, Marre M, Lévy-Marchal C, Faerch K, Borch-Johnsen K, Jørgensen T, Madsbad S, Poulsen

MTR: Barker JW, Han PK, Choi SH, Bae KT, Park S-H. Investigation of Inter-Slice Magnetization Transfer Effects as a New Method for MTR Imaging of the Human Brain. Lui S, ed. PLoS ONE. 2015;10(2):e0117101. doi:10.1371/journal.pone.0117101.

MTRR: Orozco LD, Morselli M, Rubbi L, et al. Epigenome-wide association of liver methylation patterns and complex metabolic traits in mice. Cell metabolism. 2015;21(6):905-917. doi:10.1016/j.cmet.2015.04.025. Chen J, Wang Q, Yin F-Q, Zhang W, Yan L-H, Li L. MTRR silencing inhibits growth and cisplatin resistance of ovarian carcinoma via inducing apoptosis and reducing autophagy. American Journal of Translational Research. 2015;7(9):1510-1527.

MTTP: Newberry EP, Xie Y, Kennedy SM, et al. Prevention of hepatic fibrosis with liver microsomal triglyceride transfer protein deletion in Liver fatty acid binding protein null mice. Hepatology (Baltimore, Md). 2017;65(3):836-852. doi:10.1002/hep.28941.

MYBPC1: Ha K, Buchan JG, Alvarado DM, et al. MYBPC1 mutations impair skeletal muscle function in zebrafish models of arthrogryposis. Human Molecular Genetics. 2013;22(24):4967-4977. doi:10.1093/hmg/ddt344.

MYBPC3: Maksymilian Prondzynski, Elisabeth Krämer, Sandra D. Laufer, Aya Shibamiya, Ole Pless, Frederik Flenner, Oliver J. Müller, Julia Münch, Charles Redwood, Arne Hansen, Monica Patten, Thomas Eschenhagen, Giulia Mearini, Lucie CarrierMol Ther Nucleic Acids. 2017 Jun 16; 7: 475–486. Published online 2017 May 17. doi: 10.1016/j.omtn.2017.05.008PMCID: PMC5458066 Amelia A. Glazier, Neha Hafeez, Dattatreya Mellacheruvu, Venkatesha Basrur, Alexey I. Nesvizhskii, Lap Man Lee, Hao Shao, Vi Tang.

MYNN: Genetic polymorphisms associated with telomere length and risk of developing myeloproliferative neoplasms Matteo Giaccherini, Angelica Macauda, Nicola Sgherza, Juan Sainz, Federica Gemignani, Josè Manuel Sanchez Maldonado, Manuel Jurado, Francesca Tavano, Grzegorz Mazur, Andrés Jerez, Joanna Góra-Tybor, Aleksandra Gołos, Francisca Hernández Mohedo, Joaquin Martinez Lopez, Judit Várkonyi, Raffaele Spadano, Aleksandra Butrym, Federico Canzian & Daniele Campa Myoneurin, a novel member of the BTB/POZ-zinc finger family highly expressed in human muscle. (PMID: 10873615) Alliel PM ... Périn JP Biochemical and biophysical research communications 2000 2 3 4 22 Neuromuscular expression of the BTB/POZ and zinc finger protein myoneurin. (PMID: 14694499) Cifuentes-Diaz C ... Alliel PM Muscle & nerve 2004 3 4 22 Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. (PMID: 21833088) International Multiple Sclerosis Genetics Consortium ... Compston A Nature 2011 3 40 Genetic susceptibility to distinct bladder cancer subphenotypes. (PMID: 19692168) Guey LT ... EPICURO/Spanish Bladder Cancer Study investigators European urology 2010 3 40 Multiple common variants for celiac disease influencing immune gene expression. (PMID: 20190752) Dubois PC ... van Heel DA Nature genetics 2010

MYO16: Ying F. Liu, Sarah M. Sowell, Yue Luo, Alka Chaubey, Richard S. Cameron, Hyung-Goo Kim, Anand K. SrivastavaPLoS One. 2015; 10(4): e0123106. Published online 2015 Apr 22. doi: 10.1371/journal.pone.0123106PMCID: PMC4406691 Article Pub Reader Laura Rodriguez-Murillo, Bin Xu, J Louw Roos, Gonçalo R Abecasis, Joseph A Gogos, Maria Karayiorgou Neuropsychopharmacology. 2014 Mar; 39(4): 934–943. Prepublished online 2013 Oct 21. Published online 2013 Nov 20.

MYRF: cDNA cloning and genomic structure of a novel gene (C11orf9) localized to chromosome 11q12-->q13.1 which encodes a highly conserved, potential membrane-associated protein. (PMID: 10828591) Stöhr H . Weber BH Cytogenetics and cell genetics 2000 2 3 4 56 De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. (PMID: 29446546) Pinz H . Bhoj EJ American journal of medical genetics. Part A 2018 3 4 56 De novo variants in congenital

NAA25: Polimorfismos Genéticos Associados à Razão Neutrófilo-Linfócito e suas Implicações Clínicas para os Fatores de Risco Metabólicos Os genes Park B, Choe EK, Kang HY, Shin E, Lee S e Won S. Jornal de Medicina Clínica. 2018; 7 (8): 204. doi: 10.3390 / jcm7080204

NAF1: hNaf1 is required for accumulation of human box H/ACA snoRNPs, scaRNPs, and telomerase. (PMID: 16601202) Hoareau-Aveilla C ... Henry Y RNA (New York, N.Y.) 2006 2 3 4 Stepwise RNP assembly at the site of H/ACA RNA transcription in human cells. (PMID: 16618814) Darzacq X ... Meier UT The Journal of cell biology 2006 2 3 4 Analysis of copy number variation in 8,842 Korean individuals reveals 39 genes associated with hepatic biomarkers AST and ALT. (PMID: 20797317) Kim HY ... Kim H BMB reports 2010 3 39 Common variants in the trichohyalin gene are associated with straight hair in Europeans. (PMID: 19896111) Medland SE ... Martin NG American journal of human genetics 2009 3 39 Dynamic association and localization of human H/ACA RNP proteins. (PMID: 17135485) Kittur N ... Meier UT RNA (New York, N.Y.) 2006 3 Telomere Length and Male Fertility. (PMID: 33921254) Gentiluomo M ... Campa D International journal of molecular sciences 2021

NAMPT: Cloning and characterization of the cDNA encoding a novel human pre-B-cell colony-enhancing factor. (PMID: 8289818) Samal B ... McNiece I Molecular and cellular biology 1994 2 3 4 21 Genetic variant in visfatin gene promoter is associated with decreased risk of coronary artery disease in a Chinese population. (PMID: 19804767) Yan JJ ... Huang J Clinica chimica acta; international journal of clinical chemistry 2010 3 21 39 The visfatin (PBEF1) G-948T gene polymorphism is associated with increased high-density lipoprotein cholesterol in obese subjects. (PMID: 18940394) Johansson LM ... Ridderstråle M Metabolism: clinical and experimental 2008 3 21 39 Molecular basis for the inhibition of human NMPRTase, a novel target for anticancer agents. (PMID: 16783377) Khan JA ... Tong L Nature structural & molecular biology 2006 3 4 21 Visfatin as a novel mediator released by inflamed human endothelial cells. (PMID: 24130902) Romacho T ... Peiró C PloS one 2013

NAT2: Podgorná E, Diallo I, Vangenot C, et al. Variation in NAT2 acetylation phenotypes is associated with differences in food-producing subsistence modes and ecoregions in Africa. BMC Evolutionary Biology. 2015;15:263. doi:10.1186/s12862-015-0543-6.

NCKAP5: A novel ligand for an SH3 domain of the adaptor protein Nck bears an SH2 domain and nuclear signaling motifs. (PMID: 9344857) Matuoka K ... Takenawa T Biochemical and biophysical research communications 1997 2 3 4 A genome-wide association study in the Japanese population confirms 9p21 and 14q23 as susceptibility loci for primary open angle glaucoma. (PMID: 22419738) Osman W ... Nakamura Y Human molecular genetics 2012 3 39 A genome-wide meta-analysis identifies novel loci associated with schizophrenia and bipolar disorder. (PMID: 20889312) Wang KS ... Aragam N Schizophrenia research 2010 3 39 Genome-wide association analysis of susceptibility and clinical phenotype in multiple sclerosis. (PMID: 19010793) Baranzini SE ... Oksenberg JR Human molecular genetics 2009 3 39 Genome-wide association scan of quantitative traits for attention deficit hyperactivity disorder identifies novel associations and confirms candidate gene associations. (PMID: 18821565) Lasky-Su J ... Faraone SV American journal of medical genetics. Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics 2008

NCOA6: Yun Qing, Feng Yin, Wei Wang, Yonggang Zheng, Pengfei Guo, Frederick Schozer, Hua Deng, Duojia PaneLife. 2014; 3: e02564. Published online 2014 Jul 15. doi: 10.7554/eLife.02564PMCID: PMC4118621Jae-il Roh, Cheolho Cheong, Young Hoon Sung, Jeehyun Lee, Jaewon Oh, Beom Seob Lee, Jong-Eun Lee, Yong Song Gho, Duk-Kyung Kim, Chan Bae Park, Ji Hyun Lee, Jae Woon Lee, Seok-Min Kang, Han-Woong LeeCell Rep. Author manuscript; available in PMC 2015 Aug 21.Published in final edited form as: Cell Rep. 2

NDUFA8: The nuclear-encoded human NADH:ubiquinone oxidoreductase NDUFA8 subunit: cDNA cloning, chromosomal localization, tissue distribution, and mutation detection in complex-I-deficient patients. (PMID: 9860297) Triepels R ... Smeitink J Human genetics 1998 3 4 21 Accessory subunits are integral for assembly and function of human mitochondrial complex I. (PMID: 27626371) Stroud DA ... Ryan MT Nature 2016 3 4 Protein import and oxidative folding in the mitochondrial intermembrane space of intact mammalian cells. (PMID: 23676665) Fischer M ... Riemer J Molecular biology of the cell 2013 3 4 NDUFB7 and NDUFA8 are located at the intermembrane surface of complex I. (PMID: 21310150) Szklarczyk R ... Huynen MA FEBS letters 2011 3 4 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 Interactome Mapping Provides a Network of Neurodegenerative Disease Proteins and Uncovers Widespread Protein Aggregation in Affected Brains. (PMID: 32814053) Haenig C ... Wanker EE Cell reports 2020 3 Unbiased Profiling of the Human Proinsulin Biosynthetic Interaction Network Reveals a Role for Peroxiredoxin 4 in Proinsulin Folding. (PMID: 32457219) Tran DT ... Itkin-Ansari P Diabetes 2020 3 Systems analysis of RhoGEF and RhoGAP regulatory proteins reveals spatially organized RAC1 signalling from integrin adhesions. (PMID: 32203420) Müller PM ... Rocks O Nature cell biology 2020 3 A High-Density Human Mitochondrial Proximity Interaction Network. (PMID: 32877691) Antonicka H ... Shoubridge EA Cell metabolism 2020 3 Biallelic mutations in NDUFA8 cause complex I deficiency in two siblings with favorable clinical evolution. (PMID: 33153867) Tort F ... Ribes A Molecular genetics and metabolism 2020

NEDD4: Zhu J, Lee KY, Jewett KA, Man H-Y, Chung HJ, Tsai N-P. Epilepsy-associated gene Nedd4-2 mediates neuronal activity and seizure susceptibility through AMPA receptors. Frankel WN, ed. PLoS Genetics. 2017;13(2):e1006634. doi:10.1371/journal.pgen.1006634.

NEDD4L: Ding Y, Zhang Y, Xu C, Tao Q-H, Chen Y-G. HECT Domain-containing E3 Ubiquitin Ligase NEDD4L Negatively Regulates Wnt Signaling by Targeting Dishevelled for proteínasomal Degradation. The Journal of Biological Chemistry. 2013;288(12):8289-8298. doi:10.1074/jbc.M112.433185.

NFE2L2: Pajares M, Jiménez-Moreno N, García-Yagüe ÁJ, et al. Transcription factor NFE2L2/NRF2 is a regulator of macroautophagy genes. Autophagy. 2016;12(10):1902-1916. doi:10.1080/15548627.2016.1208889.

NFKBIA: Kinker GS, Thomas AM, Carvalho VJ, Lima FP, Fujita A. Deletion and low expression of NFKBIA are associated with poor prognosis in lower-grade glioma patients. Scientific Reports. 2016;6:24160. doi:10.1038/srep24160.

NGF: Ahluwalia A, Jones MK, Hoa N, Zhu E, Brzozowski T, Tarnawski AS. Reduced NGF in Gastric Endothelial Cells Is One of the Main Causes of Impaired Angiogenesis in Aging Gastric Mucosa. Cellular and Molecular Gastroenterology and Hepatology. 2018;6(2):199-213

NINJ2: Guozhuan Zhang, Yingjiao Sun, Lei Wang, Hui Tian, Lishuang LiangOncotarget. 2017 Sep 22; 8(42): 71520–71527. Published online 2017 May 4. doi: 10.18632/oncotarget.17640PMCID: PMC5641067Joshua C. Bis, Anita DeStefano, Xiaoming Liu, Jennifer A. Brody, Seung Hoan Choi, Benjamin F. J. Verhaaren, Stéphanie Debette, M. Arfan Ikram, Eyal Shahar, Kenneth R. Butler, Jr, Rebecca F. Gottesman, Donna Muzny, Christie L. Kovar, Bruce M. Psaty, Albert Hofman, Thomas Lumley, Mayetri Gupta, Philip A. Wolf,

NIPSNAP3B: Olga Y. Gorlova, Yafang Li, Ivan Gorlov, Jun Ying, Wei V. Chen, Shervin Assassi, John D. Reveille, Frank C. Arnett, Xiaodong Zhou, Lara Bossini-Castillo, Elena Lopez-Isac, Marialbert Acosta-Herrera, Peter K. Gregersen, Annette T. Lee, Virginia D. Steen, Barri J. Fessler, Dinesh Khanna, Elena Schiopu, Richard M. Silver, Jerry A. Molitor, Daniel E. Furst, Suzanne Kafaja, Robert W. Simms, Robert A. Lafyatis, Patricia Carreira, Carmen Pilar Simeon, Ivan Castellvi, Emma Beltran, Norberto Ortego.

NKAIN3: Xu X, Zheng Z, Jia L, et al. Overexpression of SMARCA2 or CAMK2D is associated with cisplatin resistance in human epithelial ovarian cancer. Oncology Letters. 2018;16(3):3796-3804. doi:10.3892/ol.2018.9109.

NLRP1: Jin Y, Birlea SA, PR Fain, Spritz RA. Variações genéticas na NALP1 estão associadas ao vitiligo generalizado em uma população romena. J Invest Dermatol. Novembro de 2007; 127 (11): 2558-62. Epub 2007 19 de julho.Citação no PubMed Jin Y, Mailloux CM, Gowan K, Riccardi SL, LaBerge G, Bennett DC, PR Fain, Spritz RA. NALP1 em doença auto-imune múltipla associada ao vitiligo. N Engl J Med. 22 de março de 2007; 356 (12): 1216-25.Citação no PubMed

NLRP3: Wang W, Xiao F, Wan P, et al. EV71 3D Protein Binds with NLRP3 and Enhances the Assembly of Inflammasome Complex. Luo GG, ed. PLoS Pathogens. 2017;13(1):e1006123. doi:10.1371/journal.ppat.1006123.

NLRP8: Jia-Qi Chu, Ge Shi, Yi-Ming Fan, In-Wook Choi, Guang-Ho Cha, Yu Zhou, Young-Ha Lee, Juan-Hua QuanKorean J Parasitol. 2016 Dec; 54(6): 711–717. Published online 2016 Dec 31. doi: 10.3347/kjp.2016.54.6.711PMCID: PMC5266351 Gabrielle Bradshaw, Robbie R. Lualhati, Cassie L. Albury, Neven Maksemous, Deidre Roos-Araujo, Robert A. Smith, Miles C. Benton, David A. Eccles, Rod A. Lea, Heidi G. Sutherland, Larisa M. Haupt, Lyn R. GriffithsFront Immunol. 2018; 9: 420. Published online 2018 Mar 5.

NMNAT2: Yousuf O. Ali, Hunter M. Allen, Lei Yu, David Li-Kroeger, Dena Bakhshizadehmahmoudi, Asante Hatcher, Cristin McCabe, Jishu Xu, Nicole Bjorklund, Giulio Taglialatela, David A. Bennett, Philip L. De Jager, Joshua M. Shulman, Hugo J. Bellen, Hui-Chen LuPLoS Biol. 2016 Jun; 14(6): e1002472. Published online 2016 Jun 2. doi: 10.1371/journal.pbio.1002472PMCID: PMC4890852 Yousuf O. Ali, Gillian Bradley, Hui-Chen LuSci Rep. 2017; 7: 43846. Published online 2017 Mar 7.

NOD2: Gresnigt MS, Cunha C, Jaeger M, et al. Genetic deficiency of NOD2 confers resistance to invasive aspergillosis. Nature Communications. 2018;9:2636. doi:10.1038/s41467-018-04912-3.

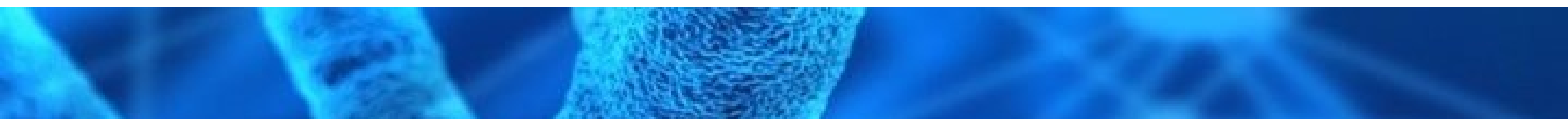
NOS3: Postberg J, Kanders M, Forcob S, et al. CpG signalling, H2A.Z/H3 acetylation and microRNA-mediated deferred self-attenuation orchestrate foetal NOS3 expression. Clinical Epigenetics. 2015;7(1):9. doi:10.1186/s13148-014-0042-4.

NOTCH2: Wang W, Yu S, Myers J, et al. Notch2 blockade enhances hematopoietic stem cell mobilization and homing. Haematologica. 2017;102(10):1785-1795. doi:10.3324/haematol.2017.168674.

NOTCH4: Lin X, Sun B, Zhu D, et al. Notch4+ cancer stem-like cells promote the metastatic and invasive ability of melanoma. Cancer Science. 2016;107(8):1079-1091. doi:10.1111/cas.12978.

NOV: Imen Nouioui, Lorena Carro, Marina García-López, Jan P. Meier-Kolthoff, Tanja Woyke, Nikos C. Kyrpides, Rüdiger Pukall, Hans-Peter Klenk, Michael Goodfellow, Markus GökerFront Microbiol. 2018; 9: 2007. Published online 2018 Aug 22. doi: 10.3389/fmicb.2018.02007PMCID: PMC6113628 Moran Brouk, Yuval Nov, Ayelet FishmanAppl Environ Microbiol. 2010 Oct; 76(19): 6397–6403. Published online 2010 Aug 13. doi: 10.1128/AEM.00878-10PMCID: PMC2950464 Chun-Xia Wang, Xin Xu, Shu-Qiang Li.

NPAS2: Ozburn AR, Kern J, Parekh PK, et al. NPAS2 Regulation of Anxiety-Like Behavior and GABAA Receptors. Frontiers in Molecular Neuroscience. 2017;10:360. doi:10.3389/fnmol.2017.00360.



NPM2: Ellard K, Serpa JJ, Petrotchenko EV, Borchers CH, Ausió J. Expression and purification of the full murine NPM2 and study of its interaction with protamines and histones. Biochemistry and Biophysics Reports. 2016;6:165-171. doi:10.1016/j.bbrep.2016.04.002.

NPPA: Man J, Barnett P, Christoffels VM. Structure and function of the Nppa–Nppbcluster locus during heart development and disease. Cellular and Molecular Life Sciences. 2018;75(8):1435-1444. doi:10.1007/s00018-017-2737-0. Pereira NL, Tosakulwong N, Scott CG, et al. Circulating ANP Genetic Association Study Identifies a Novel Gene Cluster Associated with Stroke in Caucasians. Circulation Cardiovascular genetics. 2015;8(1):141-149. doi:10.1161/CIRCGENETICS.114.000624.

NQO1: Siegel D, Dehn DD, Bokatzian SS, et al. Redox modulation of NQO1. Bratton SB, ed. PLoS ONE. 2018;13(1):e0190717. doi:10.1371/journal.pone.0190717.

NQO2: Kinetic mechanism of quinone oxidoreductase 2 and its inhibition by the antimalarial quinolines. (PMID: 15078100) Kwiek JJ ... Rudolph J Biochemistry 2004 3 4 23 26 Crystal structure of human quinone reductase type 2, a metalloflavoprotein. (PMID: 10433694) Foster CE ... Amzel LM Biochemistry 1999 3 4 23 26 Nucleotide and deduced amino acid sequence of a human cDNA (NQO2) corresponding to a second member of the NAD(P)H:quinone oxidoreductase gene family. Extensive polymorphism at the NQO2 gene locus on chromosome 6. (PMID: 1691923) Jaiswal AK ... McBride OW Biochemistry 1990 2 3 4 23 Investigation of a functional quinine oxidoreductase (NQO2) polymorphism and cognitive decline. (PMID: 18538895) Payton A ... Horan M Neurobiology of aging 2010 3 23 41 The structure of the leukemia drug imatinib bound to human quinone reductase 2 (NQO2). (PMID: 19236722) Winger JA ... Kuriyan J BMC structural biology 2009

NR2F2-AS1: Li X, liang X, Zhou L, et al. Analysis of Differential Expressions of Long Non-coding RNAs in Nasopharyngeal Carcinoma Using Next-generation Deep Sequencing. Journal of Cancer. 2018;9(11):1943-1950. doi:10.7150/jca.23481.

NR3C1: Ash GI, Kostek MA, Lee H, et al. Glucocorticoid Receptor (NR3C1) Variants Associate with the Muscle Strength and Size Response to Resistance Training. Fine ML, ed. PLoS ONE. 2016;11(1):e0148112. doi:10.1371/journal.pone.0148112.

NT5C3A: Livingstone M, Sikström K, Robert PA, Uzé G, Larsson O, Pellegrini S. Assessment of mTOR-Dependent Translational Regulation of Interferon Stimulated Genes. Langsley G, ed. PLoS ONE. 2015;10(7):e0133482. doi:10.1371/journal.pone.0133482.

NTF3: Peng W, Zhu S, Wang J, Chen L, Weng J, Chen S. Lnc-NTF3-5 promotes osteogenic differentiation of maxillary sinus membrane stem cells via sponging miR-93-3p. Clinical Implant Dentistry and Related Research. 2018;20(2):110-121. doi:10.1111/cid.12553.

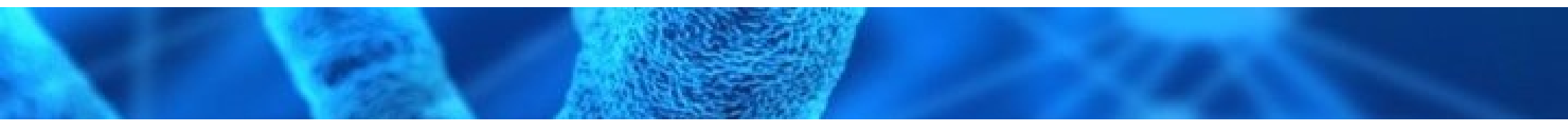
NXPH1: Vallée Marcotte B, Guénard F, Cormier H, et al. Plasma Triglyceride Levels May Be Modulated by Gene Expression of IQCJ, NXPH1, PHF17 and MYB in Humans. Ferguson L, Parslow VR, eds. International Journal of Molecular Sciences.

OCA2: Xiong Wang, Yaowu Zhu, Na Shen, Jing Peng, Chunyu Wang, Haiyi Liu, Yanjun LuOncotarget. 2017 Sep 19; 8(41): 70345–70355. Published online 2017 Jul 31. doi: 10.18632/oncotarget.19697PMCID: PMC5642559Anand Sitaram, Megan K. Dennis, Rittik Chaudhuri, Wilfredo De Jesus-Rojas, Danièle Tenza, Subba Rao Gangi Setty, Christopher S. Wood, Elena V. Sviderskaya, Dorothy C. Bennett, Graça Raposo, Juan S. Bonifacino, Michael S. MarksMol Biol Cell. 2012 Aug 15; 23(16): 3178–3192. doi: 10.1091/mbc.E11-06-

OGG1: Wang R, Li C, Qiao P, et al. OGG1-initiated base excision repair exacerbates oxidative stress-induced parthanatos. Cell Death & Disease. 2018;9(6):628. doi:10.1038/s41419-018-0680-0.

OLR1: Arslan C, Bayoglu B, Tel C, Cengiz M, Dirican A, Besirli K. Upregulation of OLR1 and IL17A genes and their association with blood glucose and lipid levels in femoropopliteal artery disease. Experimental and Therapeutic Medicine. 2017;13(3):1160-1168. doi:

OPRM1: Yamano S, Viet CT, Dang D, et al. Ex vivo non-viral gene delivery of ?-opioid receptor (OPRM1) to attenuate cancer-induced pain. Pain. 2017;158(2):240-251.



OR4A46P: Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. (PMID: 19060911) Aulchenko YS . ENGAGE Consortium Nature genetics 2009

OVOL1: Tsuji G, Hashimoto-Hachiya A, Kiyomatsu-Oda M, et al. Aryl hydrocarbon receptor activation restores filaggrin expression via OVOL1 in atopic dermatitis. Cell Death & Disease. 2017;8(7):e2931-. doi:10.1038/cddis.2017.322.

OXTR: He J, Buil JM, Koot HM, van Lier PAC. Associations between Oxytocin Receptor (OXTR) Genotype and Elementary School Children's Likability, Dis-likability and Friendship among Classroom Peers: A Longitudinal Study. Journal of Youth and Adolescence. 2018 Polymorphisms in the oxytocin receptor gene are associated with the development of psychopathy. Dadds MR1, Moul C1, Cauchi A1, Dobson-Stone C1, Hawes DJ2, Brennan J3, Urwin R4, Ebstein RE5. The association between oxytocin receptor gene polymo

PACERR: Teng H, Mao F, Liang J, et al. Transcriptomic signature associated with carcinogenesis and aggressiveness of papillary thyroid carcinoma. Theranostics. 2018;8(16):4345-4358. doi:10.7150/thno.26862.

PARK7: Yasuomi Urano, Chinatsu Mori, Ayano Fuji, Keito Konno, Takayuki Yamamoto, Shohei Yashirogi, Mayu Ando, Yoshiro Saito, Noriko NoguchiAutophagy. 2018; 14(11): 1943–1958. Published online 2018 Aug 16. doi: 10.1080/15548627.2018.1493043PMCID: PMC6152502Bao Wang, Zhibiao Cai, Kai Tao, Weijun Zeng, Fangfang Lu, Ruixin Yang, Dayun Feng, Guodong Gao, Qian YangAutophagy. 2016 Aug; 12(8): 1215–1228. Published online 2016 May 12. doi: 10.1080/15548627.2016.1179401PMCID: PMC4968227Hui Yu, Jolena N. Wad

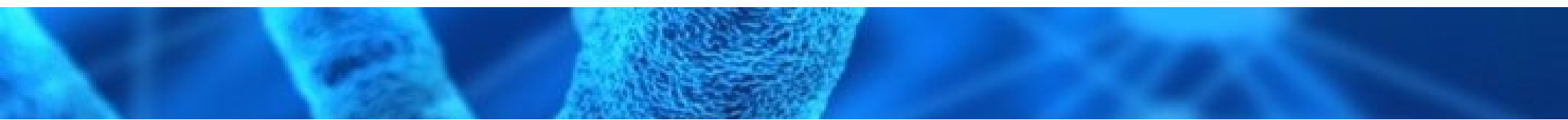
PAX4: Xu Y, Wang Y, Song Y, et al. Generation and Phenotype Identification of PAX4Gene Knockout Rabbit by CRISPR/Cas9 System. G3: Genes|Genomes|Genetics. 2018;8(8):2833-2840. doi:10.1534/g3.118.300448.

PCAT19: Ronchetti D, Agnelli L, Pietrelli A, et al. A compendium of long non-coding RNAs transcriptional fingerprint in multiple myeloma. Scientific Reports. 2018;8:6557. doi:10.1038/s41598-018-24701-8.

PCDH11X: Johansson MM, Lundin E, Qian X, et al. Spatial sexual dimorphism of X and Y homolog gene expression in the human central nervous system during early male development. Biology of Sex Differences. 2016;7:5. doi:10.1186/s13293-015-0056-4.

PCDH9: Xie Z, Zhou F, Yang Y, et al. Lnc-PCDH9-13:1 Is a Hypersensitive and Specific Biomarker for Early Hepatocellular Carcinoma. EBioMedicine. 2018;33:57-67. doi:10.1016/j.ebiom.2018.06.026.

PCIF1: Kathryn C. Claiborn, Mira M. Sachdeva, Corey E. Cannon, David N. Groff, Jeffrey D. Singer, Doris A. StoffersJ Clin Invest. 2010 Oct 1; 120(10): 3713–3721. Published online 2010 Sep 1. doi: 10.1172/JCI40440PMCID: PMC2947215Aihua Liu, Biva M. Desai, Doris A. StoffersMol Cell Biol. 2004 May; 24(10): 4372–4383. doi: 10.1128/MCB.24.10.4372-4383.2004PMCID: PMC400448Sabrina Klein, Rui Meng, Mathias Montenarh, Claudia GötzPharmaceuticals (Basel) 2017 Mar; 10(1): 2. Published online 2016 Dec 28. doi



PCK1: A Leu184Val polymorphism in PCK1 gene is associated with type 2 diabetes in Eastern Chinese population with BMI<23 kg/m2. (PMID: 19070910) Dong Y ... Su Q Diabetes research and clinical practice 2009 3 21 39 The promoter polymorphism -232C/G of the PCK1 gene is associated with type 2 diabetes in a UK-resident South Asian population. (PMID: 19725958) Rees SD ... Kelly MA BMC medical genetics 2009 3 21 39 The estrogen hypothesis of schizophrenia implicates glucose metabolism: association study in three independent samples. (PMID: 18460190) Olsen L ... Werge T BMC medical genetics 2008 3 21 39 Association of the promoter polymorphism -232C/G of the phosphoenolpyruvate carboxykinase gene (PCK1) with Type 2 diabetes mellitus. (PMID: 16620271) Gouni-Berthold I ... Krone W Diabetic medicine : a journal of the British Diabetic Association 2006 3 21 39 Large-scale study of the -232C > G polymorphism of PCK1 in Type 2 diabetes. (PMID: 16978381) Wegner L ... Pedersen O Diabetic medicine : a journal of the British Diabetic Association 2006 PCK1 and DHODH drive colorectal cancer liver metastatic colonization and hypoxic growth by promoting nucleotide synthesis. (PMID: 31841108) Yamaguchi N ... Tavazoie SF eLife 2019 3 RNA-seq reveals outcome-specific gene expression of MMP7 and PCK1 in biliary atresia. (PMID: 31342296) Ramachandran P ... Mahalingam S Molecular biology reports 2019 3 Relationships of SLC2A4, RBP4, PCK1, and PI3K Gene Polymorphisms with Gestational Diabetes Mellitus in a Chinese Population. (PMID: 30805369) Hu S ... Tan H BioMed research international 2019 3 Increased expression of phosphoenolpyruvate carboxykinase cytoplasmic isoform by hepatitis B virus X protein affects hepatitis B virus replication. (PMID: 30168585) Tang X ... Tang H Journal of medical virology 2019 3 PCK1 negatively regulates cell cycle progression and hepatoma cell proliferation via the AMPK/p27Kip1 axis. (PMID: 30717766) Tuo L ... Tang N Journal of experimental & clinical cancer research : CR 2019 The gluconeogenic enzyme PCK1 phosphorylates INSIG1/2 for lipogenesis. (PMID: 32322062) Xu D ... Lu Z Nature 2020 3 4 Recognition of the gluconeogenic enzyme, Pck1, via the Gid4 E3 ligase: An in silico perspective. (PMID: 31883179) Ismail AM ... Elshemey WM Journal of molecular recognition : JMR 2020 3 Hypoxia increases the rate of renal gluconeogenesis via hypoxia-inducible factor-1-dependent activation of phosphoenolpyruvate carboxykinase expression. (PMID: 32045650) Owczarek A ... Winiarska K Biochimie 2020 3 Hepatocellular carcinoma cell-derived extracellular vesicles encapsulated microRNA-584-5p facilitates angiogenesis through PCK1-mediated nuclear factor E2-related factor 2 signaling pathway. (PMID: 32522621) Shao Z ... Zhang Y The international journal of biochemistry & cell biology 2020 3 Cytosolic phosphoenolpyruvate carboxykinase is expressed in α-cells from human and murine pancreas. (PMID: 31180589) Westermeier F ... Bertinat R Journal of cellular physiology 2020

PCSK1: Ramos-Molina B, Martin MG, Lindberg I. PCSK1 Variants and Human Obesity. Progress in molecular biology and translational science. 2016;140:47-74. doi:10.1016/bs.pmbts.2015.12.001.

PCSK9: Glerup S, Schulz R, Laufs U, Schlüter K-D. Physiological and therapeutic regulation of PCSK9 activity in cardiovascular disease. Basic Research in Cardiology. 2017;112(3):32. doi:10.1007/s00395-017-0619-0.

PDE11A: Kelly MP. A Role for Phosphodiesterase 11A (PDE11A) in the Formation of Social Memories and the Stabilization of Mood. Advances in neurobiology. 2017;17:201-230. doi:10.1007/978-3-319-58811-7_8.

PDE4B: Suhasini AN, Wang L, Holder KN, et al. A phosphodiesterase 4B-dependent interplay between tumor cells and the microenvironment regulates angiogenesis in B-cell lymphoma. Leukemia. 2016;30(3):617-626. doi:10.1038/leu.2015.302.

PDE4D: Böttcher R, Dulla K, van Strijp D, et al. Human PDE4D isoform composition is deregulated in primary prostate cancer and indicative for disease progression and development of distant metastases. Oncotarget. 2016;7(43):70669-70684. doi:10.18632/oncotarget.1

PDE9A: Lee DI, Zhu G, Sasaki T, et al. Phosphodiesterase 9A Controls Nitric-oxide Independent cGMP and Hypertrophic Heart Disease. Nature. 2015;519(7544):472-476. doi:10.1038/nature14332.

PDSS1: Julie Mollet, Irina Giurgea, Dimitri Schlemmer, Gustav Dallner, Dominique Chretien, Agnès Delahodde, Delphine Bacq, Pascale de Lonlay, Arnold Munnich, Agnès RötigJ Clin Invest. 2007 Mar 1; 117(3): 765–772. Published online 2007 Mar 1. doi: 10.1172/JCI29089PMCID: PMC1804361Ryoichi Saiki, Adam L. Lunceford, Yuchen Shi, Beth Marbois, Rhonda King, Justin Pachuski, Makoto Kawamukai, David L. Gasser, Catherine F. ClarkeAm J Physiol Renal Physiol. 2008 Nov; 295(5): F1535–F1544. Published online 20

PDSS2: Chungang Feng, Yu Gao, Ben Dorshorst, Chi Song, Xiaorong Gu, Qingyuan Li, Jinxiu Li, Tongxin Liu, Carl-Johan Rubin, Yiqiang Zhao, Yanqiang Wang, Jing Fei, Huifang Li, Kuanwei Chen, Hao Qu, Dingming Shu, Chris Ashwell, Yang Da, Leif Andersson, Xiaoxiang Hu, Ning LiPLoS Genet. 2014 Aug; 10(8): e1004576. Published online 2014 Aug 28. doi: 10.1371/journal.pgen.1004576PMCID: PMC4148213Mitsuro Kanda, Shuji Nomoto, Hisaharu Oya, Ryoji Hashimoto, Hideki Takami, Dai Shimizu, Fuminori Sonohara, Daisu

PECAM1: Sun X, Huang S, Wang X, Zhang X, Wang X. CD300A promotes tumor progression by PECAM1, ADCY7 and AKT pathway in acute myeloid leukemia. Oncotarget. 2018;9(44):27574-27584. doi:10.18632/oncotarget.24164.

PEX5L: Markus Kunze, Naila Malkani, Sebastian Maurer-Stroh, Christoph Wiesinger, Johannes A. Schmid, Johannes BergerJ Biol Chem. 2015 Feb 20; 290(8): 4928–4940. Published online 2014 Dec 23. doi: 10.1074/jbc.M114.601575PMCID: PMC4335231 Tony A. Rodrigues, Inês S. Alencastre, Tânia Francisco, Pedro Brites, Marc Fransen, Cláudia P. Grou, Jorge E. AzevedoMol Cell Biol. 2014 Aug; 34(15): 2917–2928. doi: 10.1128/MCB.01727-13PMCID: PMC4135580 Erika Celis-Aguilar, Luis Lassaletta, Miguel Torres-Martín.

PEX6: Gardner BM, Castanzo DT, Chowdhury S, et al. The peroxisomal AAA-ATPase Pex1/Pex6 unfolds substrates by processive threading. Nature Communications. 2018;9:135. doi:10.1038/s41467-017-02474-4.

PFKP: Lee J-H, Liu R, Li J, et al. Stabilization of phosphofructokinase 1 platelet isoform by AKT promotes tumorigenesis. Nature Communications. 2017;8:949. doi:10.1038/s41467-017-00906-9.

PHACTR1: Annina Kelloniemi, Zoltan Szabo, Raisa Serpi, Juha Näpänkangas, Pauli Ohukainen, Olli Tenhunen, Leena Kaikkonen, Elina Koivisto, Zsolt Bagyura, Risto Kerkelä, Margret Leosdottir, Thomas Hedner, Olle Melander, Heikki Ruskoaho, Jaana RysäPLoS One. 2015; 10(6): e0130502. Published online 2015 Jun 22. doi: 10.1371/journal.pone.0130502PMCID: PMC4476650 Valérie-Anne Codina-Fauteux, Mélissa Beaudoin, Simon Lalonde, Ken Sin Lo, Guillaume Lettre BMC Med Genet. 2018; 19: 97. Published online 2018 Jun

PHYHIP: Skvortsova YV, Kondratieva SA, Zinovyeva MV, Nikolaev LG, Azhikina TL, Gainetdinov IV. Intragenic Locus in Human PIWIL2 Gene Shares Promoter and Enhancer Functions. Grosso M, ed. PLoS ONE. 2016;11(6):e0156454. doi:10.1371/journal.pone.0156454.

PICALM: Stern, J. A., White, S. N., Lehmkuhl, L. B., Reina-Doreste, Y., Ferguson, J. L., Nascone-Yoder, N. M., Meurs, K. M. A single codon insertion in PICALM is associated with development of familial subvalvular aortic stenosis in Newfoundland dogs. Hum. Genet. 133: 1139-1148, 2014. [PubMed: 24898977, images, related citations] Treusch, S., Hamamichi, S., Goodman, J. L., Matlack, K. E. S., Chung, C. Y., Baru, V., Shulman, J. M., Parrado, A., Bevis, B. J., Valastyan, J. S., Han, H., Lindhagen-Per

PIGU: Pflueger D, Terry S, Sboner A, et al. Discovery of non-ETS gene fusions in human prostate cancer using next-generation RNA sequencing. Genome Research. 2011;21(1):56-67. doi:10.1101/gr.110684.110.

PIK3CA: Nicole Bäumer, Jan Rehkämper, Neele Appel, Lisa Terheyden, Wolfgang Hartmann, Eva Wardelmann, Frank Buchholz, Carsten Müller-Tidow, Wolfgang E. Berdel, Sebastian BäumerPLoS One. 2018; 13(7): e0200163. Published online 2018 Jul 12. doi: 10.1371/journal.pone.0200163PMCID: PMC6042707Vanessa F. Merino, Soonweng Cho, Xiaohui Liang, Sunju Park, Kideok Jin, Qian Chen, Duojia Pan, Cynthia A. Zahnow, Alan R. Rein, Saraswati SukumarMol Oncol. 2017 May; 11(5): 552–566. Published online 2017 Apr 6. doi

PIP4K2A: Liao F, Yin D, Zhang Y, et al. Association Between PIP4K2A Polymorphisms and Acute Lymphoblastic Leukemia Susceptibility. Wu. Y, ed. Medicine. 2016;95(18):e3542. doi:10.1097/MD.0000000000003542.

PITX2: Waite M, Skidmore J, Billi A, Martin J, Martin D. GABAergic and glutamatergic identities of developing midbrain Pitx2 neurons. Developmental dynamics?: an official publication of the American Association of Anatomists. 2011;240(2):333-346. doi:10.1002/dvd

PKNOX2: Toivonen J, Kivioja T, Jolma A, Yin Y, Taipale J, Ukkonen E. Modular discovery of monomeric and dimeric transcription factor binding motifs for large data sets. Nucleic Acids Research. 2018;46(8):e44. doi:10.1093/nar/gky027.

PLAU: Hayward CPM, Liang M, Tasneem S, et al. The duplication mutation of Quebec platelet disorder dysregulates PLAU, but not C10orf55, selectively increasing production of normal PLAU transcripts by megakaryocytes but not granulocytes. Schulz C, ed. PLoS ONE.

PLCD1: Hereditary leukonychia, or porcelain nails, resulting from mutations in PLCD1. (PMID: 21665001) Kiuru M ... Christiano AM American journal of human genetics 2011 3 4 69 Genomic structure of the human PLCD1 (phospholipase C delta 1) locus on 3p22-->p21.3. (PMID: 9345909) Ishikawa S ... Nakamura Y Cytogenetics and cell genetics 1997 2 3 21 Cloning and identification of amino acid residues of human phospholipase C delta 1 essential for catalysis. (PMID: 7890667) Cheng HF ... King K The Journal of biological chemistry 1995 3 4 21 Variation at the NFATC2 locus increases the risk of thiazolidinedione-induced edema in the Diabetes REduction Assessment with ramipril and rosiglitazone Medication (DREAM) study. (PMID: 20628086) Bailey SD ... DREAM investigators Diabetes care 2010 3 39 The small GTPase Ral couples the angiotensin II type 1 receptor to the activation of phospholipase C-delta 1. (PMID: 20018811) Godin CM ... Ferguson SS Molecular pharmacology 2010

PLCL1: Zhou R, Lin X, Li D-Y, et al. Identification of novel genetic loci for osteoporosis and/or rheumatoid arthritis using cFDR approach. Wei Z, ed. PLoS ONE. 2017;12(8):e0183842. doi:10.1371/journal.pone.0183842.

PLCL2: Arismendi M, Giraud M, Ruzehaji N, et al. Identification of NF-?B and PLCL2 as new susceptibility genes and highlights on a potential role of IRF8 through interferon signature modulation in systemic sclerosis. Arthritis Research & Therapy. 2015;17(1):71.

PLD3: Satoh J, Kino Y, Yamamoto Y, et al. PLD3 is accumulated on neuritic plaques in Alzheimer's disease brains. Alzheimer's Research & Therapy. 2014;6(9):70. doi:10.1186/s13195-014-0070-5.

PLEKHG1: Wu Y, Luna MJ, Bonilla LS, Ryba NJP, Pickel JM. Characterization of knockin mice at the Rosa26, Tac1 and Plekhg1 loci generated by homologous recombination in oocytes. Hu W, ed. PLoS ONE. 2018;13(2):e0193129. doi:10.1371/journal.pone.0193129.

PLPP3: Silvia Aldi, Ljubica Perisic Matic, Gregory Hamm, Daniëlle van Keulen, Dennie Tempel, Kim Holmstrøm, Agnieszka Szwajda, Boye Schnack Nielsen, Valur Emilsson, Rima Ait-Belkacem, Mariette Lengquist, Gabrielle Paulsson-Berne, Per Eriksson, Jan H.N. Lindeman, Alain J. Gool, Jonathan Stauber, Ulf Hedin, Eva Hurt-Camejo Mol Ther Methods Clin Dev. 2018 Sep 21; 10: 17–28. Published online 2018 Jun 27. doi: 10.1016/j.omtm.2018.05.003PMCID: PMC6039967 Marco Busnelli, Stefano Manzini, Mika Hilvo.

PLTP: Audo R, Deckert V, Daien CI, et al. PhosphoLipid transfer protein (PLTP) exerts a direct pro-inflammatory effect on rheumatoid arthritis (RA) fibroblasts-like-synoviocytes (FLS) independently of its lipid transfer activity. Zhou H, ed. PLoS ONE. 2018;13(3

PNMT: Lee SE, Oh E, Lee B, et al. Phenylethanolamine N-methyltransferase downregulation is associated with malignant pheochromocytoma/paraganglioma. Oncotarget. 2016;7(17):24141-24153. doi:10.18632/oncotarget.8234.

POC5: Shunmoogum A. Patten, Patricia Margaritte-Jeannin, Jean-Claude Bernard, Eudeline Alix, Audrey Labalme, Alicia Besson, Simon L. Girard, Khaled Fendri, Nicolas Fraisse, Bernard Biot, Coline Poizat, Amandine Campan-Fournier, Kariman Abelin-Genevois, Vincent Cunin, Charlotte Zaouter, Meijiang Liao, Raphaelle Lamy, Gaetan Lesca, Rita Menassa, Charles Marcaillou, Melanie Letexier, Damien Sanlaville, Jerome Berard, Guy A. Rouleau, Françoise Clerget-Darpoux, Pierre Drapeau, Florina Moldovan.

POLN: Takata K, Reh S, Yousefzadeh MJ, et al. Analysis of DNA polymerase ? function in meiotic recombination, immunoglobulin class-switching, and DNA damage tolerance. Cohen PE, ed. PLoS Genetics. 2017;13(6):e1006818. doi:10.1371/journal.pgen.1006818.

PON2: Krüger M, Amort J, Wilgenbus P, et al. The anti-apoptotic PON2 protein is Wnt/?-catenin-regulated and correlates with radiotherapy resistance in OSCC patients. Oncotarget. 2016;7(32):51082-51095. doi:10.18632/oncotarget.9013.

POU5F1: Daigneault BW, Rajput S, Smith GW, Ross PJ. Embryonic POU5F1 is Required for Expanded Bovine Blastocyst Formation. Scientific Reports. 2018;8:7753. doi:10.1038/s41598-018-25964-x.

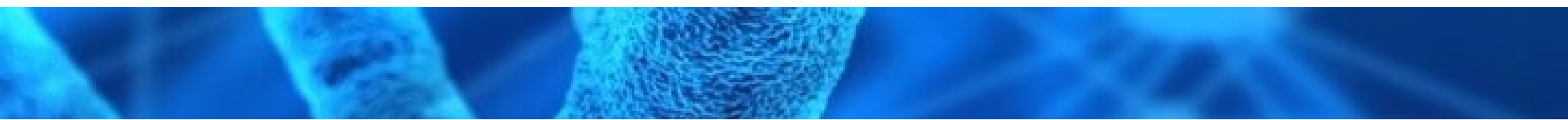
PPARA: Brocker CN, Yue J, Kim D, Qu A, Bonzo JA, Gonzalez FJ. Hepatocyte-specific PPARA expression exclusively promotes agonist-induced cell proliferation without influence from nonparenchymal cells. American Journal of Physiology - Gastrointestinal and Liver Ph

PPARG: Dominant negative mutations in human PPARgamma associated with severe insulin resistance, diabetes mellitus and hypertension. (PMID: 10622252) Barroso I ... O'Rahilly S Nature 1999 3 4 21 70 Association of the PPARGC1A gene polymorphism with diabetic nephropathy in an Asian Indian population (CURES-41). (PMID: 19900151) Gayathri SB ... Mohan V Metabolic syndrome and related disorders 2010 3 21 39 Relationship of five type 2 diabetes candidate gene polymorphisms to the age at diagnosis of diabetes in the Slovakian population. (PMID: 20437825) Kozarova M ... Tkac I Bratislavske lekarske listy 2010 3 21 39 PPARgamma gene C161T substitution alters lipid profile in Chinese patients with coronary artery disease and type 2 diabetes mellitus. (PMID: 20334678) Wan J ... Roy S Cardiovascular diabetology 2010 3 21 39 PPARG and ADIPOQ gene polymorphisms increase type 2 diabetes mellitus risk in Asian Indian Sikhs: Pro12Ala still remains as the strongest predictor. (PMID: 19846176) Sanghera DK ... Kamboh IM Metabolism: clinical and experimental 2010 [The Expression of Genes Encoding ABCA1 and ABCG1 Transporters and PPARγ, LXRβ, and RORα Transcriptional Factors in Subcutaneous and Visceral Adipose Tissue in Women with Metabolic Syndrome]. (PMID: 33566026) Panteleeva AA ... Miroshnikova VV Molekuliarnaia biologiia 2021 3 The Emerging Role of COX-2, 15-LOX and PPARγ in Metabolic Diseases and Cancer: An Introduction to Novel Multi-target Directed Ligands (MTDLs). (PMID: 32867639) Alaaeddine RA ... El-Yazbi AF Current medicinal chemistry 2021 3 A Transcriptional Regulatory Loop of Master Regulator Transcription Factors, PPARG, and Fatty Acid Synthesis Promotes Esophageal Adenocarcinoma. (PMID: 33402390) Ma S ... Lin DC Cancer research 2021 3 Estrogen receptor-associated receptor α and peroxisome proliferator-activated receptor γ in metabolism and disease (Review). (PMID: 33355368) Huang WY ... Sun PM Molecular medicine reports 2021 3 Effects of FTO and PPARγ variants on intrauterine growth restriction in a Brazilian birth cohort. (PMID: 33729310) Barbieri MR ... Bettiol H Brazilian journal of medical and biological research = Revista brasileira de pesquisas medicas e biologicas 2021 Frequency of PPAR-γ, FTO and ABCC8 genetic variation in Pakistani cardiovascular smokers. (PMID: 32712935) Rehman K ... Akash MSH Environmental science and pollution research international 2020 3 ACSL1 affects Triglyceride Levels through the PPARγ Pathway. (PMID: 32218693) Li T ... Meng F International journal of medical sciences 2020 Nuclear hormone receptors: Ancient 9aaTAD and evolutionally gained NCoA activation pathways. (PMID: 30468856) Piskacek M ... Knight A The Journal of steroid biochemistry and molecular biology 2019 4 LncRNA AC096664.3/PPAR-γ/ABCG1-dependent signal transduction pathway contributes to the regulation of cholesterol homeostasis. (PMID: 30938872) Xu BM ... Wang Q Journal of cellular biochemistry 2019 3 Interaction Between AGTR1 and PPARγ Gene Polymorphisms on the Risk of Nonalcoholic Fatty Liver Disease. (PMID: 30793973) Zhu P ... Dong C Genetic testing and molecular biomarkers 2019 DHA-enriched fish oil upregulates cyclin-dependent kinase inhibitor 2A (P16INK) expression and downregulates telomerase activity without modulating effects of PPARγ Pro12Ala polymorphism in type 2 diabetic patients: A randomized, double-blind, placebo-controlled clinical trial. (PMID: 28024882) Toupchian O ... Koohdani F Clinical nutrition (Edinburgh, Scotland) 2018 3 DJ-1 Alleviates Angiotensin II-Induced Endothelial Progenitor Cell Damage by Activating the PPARγ/HO-1 Pathway. (PMID: 28600848) Han T ... Yang S Journal of cellular biochemistry 2018 3 Influence of expression of UCP3, PLIN1 and PPARG2 on the oxidation of substrates after hypocaloric dietary intervention. (PMID: 28651828) Cortes de Oliveira C ... Nonino CB Clinical nutrition (Edinburgh, Scotland) 2018 3 Diabetes mellitus, superoxide dismutase and peroxisome proliferator activated receptor gamma polymorphisms modify the outcome of end-stage renal disease patients of Han Chinese origin. (PMID: 27925431) Chao CT ... Yen CJ Nephrology (Carlton, Vic.) 2018

PPARGC1A: Zhu L, Huang Q, Xie Z, et al. PPARGC1A rs3736265 G>A polymorphism is associated with decreased risk of type 2 diabetes mellitus and fasting plasma glucose level. Oncotarget. 2017;8(23):37308-37320. doi:10.18632/oncotarget.16307.

PPARGC1B: Zhang S, Jiang J, Chen Z, et al. Relationship of PPARG, PPARGC1A, and PPARGC1B polymorphisms with susceptibility to hepatocellular carcinoma in an eastern Chinese Han population. OncoTargets and therapy. 2018;11:4651-4660. doi:10.2147/OTT.S168274.

PPM1H: Cai M, Li L. Subtype identification from heterogeneous TCGA datasets on a genomic scale by multi-view clustering with enhanced consensus. BMC Medical Genomics. 2017;10(Suppl 4):75. doi:10.1186/s12920-017-0306-x.



PPOX: Barbaro M, Kotajärvi M, Harper P, Floderus Y. Partial protoporphyrinogen oxidase (PPOX) gene deletions, due to different Alu-mediated mechanisms, identified by MLPA analysis in patients with variegate porphyria. Orphanet Journal of Rare Diseases. 2013;8:1

PPP1R3B: Li W-J, Yin R-X, Huang J-H, Bin Y, Chen W-X, Cao X-L. Association between the PPP1R3B polymorphisms and serum lipid traits, the risk of coronary artery disease and ischemic stroke in a southern Chinese Han population. Nutrition & Metabolism. 2018;15:27. d

PRDM6: Mutations in the Histone Modifier PRDM6 Are Associated with Isolated Nonsyndromic Patent Ductus Arteriosus. (PMID: 27716515) Li N . Mani A American journal of human genetics 2016 2 3 56 Mutations in the Histone Modifier PRDM6 Are Associated with Isolated Nonsyndromic Patent Ductus Arteriosus. (PMID: 27181681) Li N . Mani A American journal of human genetics 2016 3 4 56 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 2037

PRKCH: Pedros C, Canonigo-Balancio AJ, Kong K-F, Altman A. Requirement of Treg-intrinsic CTLA4/PKC? signaling pathway for suppressing tumor immunity. JCI Insight. 2017;2(23):e95692. doi:10.1172/jci.insight.95692.

PROC: Musia? R, Ocho?ska K, Proc A, et al. Veno-arterial extracorporeal membrane oxygenation as cardiogenic shock therapy support in adult patients after heart surgery. Kardiochirurgia i Torakochirurgia Polska = Polish Journal of Cardio-Thoracic Surgery. 2017;1

PRR5L: Genetic and epigenetic studies of atopic dermatitis Lianghua Bin and Donald Y. M. Leungcorresponding author Multi-ethnic genome-wide association study of 21,000 cases and 95,000 controls identifies new risk loci for atopic dermatitis Lavinia Paternoster,#1,2 Marie Standl,#3 Johannes Waage,4 Hansjörg Baurecht,5 Melanie Hotze,5 David P Strachan,6 John A Curtin,7 Klaus Bønnelykke,4 Chao Tian,8 Atsushi Takahashi,9 Jorge Esparza-Gordillo,10,11 Alexessander Couto Alves,12 Jacob P Thyssen,13 Herma

PRRC2C: Biswas K, Wagner Mackenzie B, Waldvogel-Thurlow S, et al. Differentially Regulated Host Proteins Associated with Chronic Rhinosinusitis Are Correlated with the Sinonasal Microbiome. Frontiers in Cellular and Infection Microbiology. 2017;7:504. doi:10.3389

PSEN1: Pathway-based approaches to imaging genetics association studies: Wnt signaling, GSK3beta substrates and major depression. (PMID: 20219685) Inkster B ... Matthews PM NeuroImage 2010 3 39 Archaeal roots of intramembrane aspartyl protease siblings signal peptide peptidase and presenilin. (PMID: 32935885) Raut P ... Lieberman RL Proteins 2021 3 Neuroprotective effect of emodin against Alzheimer's disease via Nrf2 signaling in U251 cells and APP/PS1 mice. (PMID: 33300068) Li Z ... Fei X Molecular medicine reports 2021 3 Alzheimer's Disease Associated Presenilin 1 and 2 Genes Dysregulation in Neonatal Lymphocytes Following Perinatal Asphyxia. (PMID: 34067945) Tarkowska A ... Pluta R International journal of molecular sciences 2021 3 PSEN1 Compound Heterozygous Mutations Associated with Cerebral Amyloid Angiopathy and Cognitive Decline Phenotype. (PMID: 33918046) Palmieri I ... Cereda C International journal of molecular sciences 2021 A novel mutation in PSEN1 (p.T119I) in an Argentine family with early- and late-onset Alzheimer's disease. (PMID: 31153663) Itzcovich T ... Surace EI Neurobiology of aging 2020 3 70 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 Systematic validation of variants of unknown significance in APP, PSEN1 and PSEN2. (PMID: 32087291) Hsu S ... Karch CM Neurobiology of disease 2020 Recognition of the amyloid precursor protein by human γ-secretase. (PMID: 30630874) Zhou R ... Shi Y Science (New York, N.Y.) 2019 Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. (PMID: 18667258) Guerreiro RJ ... Clarimón J Neurobiology of aging 2010 3 21 39 70 A novel presenilin-1 mutation (Leu85Pro) in early-onset Alzheimer disease with spastic paraparesis. (PMID: 15534188) Ataka S ... Miki T Archives of neurology 2004 3 4 21 70 A novel presenilin 1 mutation associated with Pick's disease but not beta-amyloid plaques. (PMID: 15122701) Dermaut B ... De Deyn PP Annals of neurology 2004 3 4 21 70 A novel presenilin 1 mutation (L174 M) in a large Cuban family with early onset Alzheimer disease. (PMID: 12484344) Bertoli Avella AM ... Heutink P Neurogenetics 2002 3 4 21 70 Molecular evidence of presenilin 1 mutation in familial early onset dementia. (PMID: 11920851) Matsubara-Tsutsui M ... Miki T American journal of medical genetics 2002

PSEN2: Giau VV, Pyun J-M, Bagyinszky E, An SSA, Kim S. A pathogenic PSEN2p.His169Asn mutation associated with early-onset Alzheimer's disease. Clinical Interventions in Aging. 2018;13:1321-1329. doi:10.2147/CIA.S170374.

PSMA6: Kakumu T, Sato M, Goto D, et al. Identification of Proteínasomal catalytic subunit PSMA6 as a therapeutic target for lung cancer. Cancer Science. 2017;108(4):732-743. doi:10.1111/cas.13185.

PSORS1C3: Wi?niewski A, Matusiak ?, Szczerkowska-Dobosz A, Nowak I, Ku?nierczyk P. HLA-C*06:02-independent, gender-related association of PSORS1C3 and PSORS1C1/CDSN single-nucleotide polymorphisms with risk and severity of psoriasis. Molecular Genetics and Genomics

PSRC1: Xi D, Zhao J, Guo K, et al. Serum amyloid P component therapeutically attenuates atherosclerosis in mice via its effects on macrophages. Theranostics. 2018;8(12):3214-3223. doi:10.7150/thno.22704.

PTEN: Amit Gupta, Nicholas R. LeslieJ Biol Chem. 2016 Aug 26; 291(35): 18465–18473. Published online 2016 Jul 12. doi: 10.1074/jbc.M116.727750PMCID: PMC5000091Arman Javadi, Ravi K Deevi, Emma Evergren, Elodie Blondel-Tepaz, George S Baillie, Mark GH Scott, Frederick C CampbelleLife. 2017; 6: e24578. Published online 2017 Jul 27. doi: 10.7554/eLife.24578PMCID: PMC5576923Xiaoxiao Wang, Xin Cao, Ruifang Sun, Charlene Tang, Alexandar Tzankov, Jun Zhang, Ganiraju C. Manyam, Min Xiao, Yi Miao, Kausar J

PTGS2: Vogel LK, Sæbø M, Høyer H, et al. Intestinal PTGS2 mRNA Levels, PTGS2Gene Polymorphisms, and Colorectal Carcinogenesis. Scheurer M, ed. PLoS ONE. 2014;9(8):e105254. doi:10.1371/journal.pone.0105254.

PTPN22: Spalinger MR, Kasper S, Gottier C, et al. NLRP3 tyrosine phosphorylation is controlled by protein tyrosine phosphatase PTPN22. The Journal of Clinical Investigation. 2016;126(5):1783-1800. doi:10.1172/JCI83669.

PTPRD: Berenice Ortiz, Armida W. M. Fabius, Wei H. Wu, Alicia Pedraza, Cameron W. Brennan, Nikolaus Schultz, Kenneth L. Pitter, Jacqueline F. Bromberg, Jason T. Huse, Eric C. Holland, Timothy A. Chan Proc Natl Acad Sci U S A. 2014 Jun 3; 111(22): 8149–8154. Published online 2014 May 19. doi: 10.1073/pnas.1401952111 PMCID: PMC4050622 Noah D. Peyser, Yu Du, Hua Li, Vivian Lui, Xiao Xiao, Timothy A. Chan, Jennifer R. Grandis PLoS One. 2015; 10(8): e0135750. Published online 2015 Aug 12.

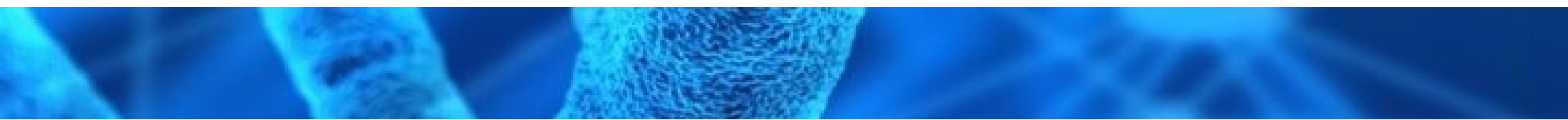
PTPRN2: Sengelaub CA, Navrazhina K, Ross JB, Halberg N, Tavazoie SF. PTPRN2 and PLC?1 promote metastatic breast cancer cell migration through PI(4,5)P2-dependent actin remodeling. The EMBO Journal. 2016;35(1):62-76. doi:10.15252/embj.201591973.

PTPRS: Davis TB, Yang M, Schell MJ, et al. PTPRS Regulates Colorectal Cancer RAS Pathway Activity by Inactivating Erk and Preventing Its Nuclear Translocation. Scientific Reports. 2018;8:9296. doi:10.1038/s41598-018-27584-x.

PUS10: Multi-ethnic genome-wide association study of 21,000 cases and 95,000 controls identifies new risk loci for atopic dermatitis Lavinia Paternoster,#1,2 Marie Standl,#3 Johannes Waage,4 Hansjörg Baurecht,5 Melanie Hotze,5 David P Strachan,6 John A Curtin,7 Klaus Bønnelykke,4 Chao Tian,8 Atsushi Takahashi,9 Jorge Esparza-Gordillo,10,11 Alexessander Couto Alves,12 Jacob P Thyssen,13 Herman T den Dekker,14,15,16 Manuel A Ferreira,17 Elisabeth Altmaier,18,19,20 Patrick MA Sleiman,21,22 Feng Li Xi

PVALB: Dipoppa M, Ranson A, Krumin M, Pachitariu M, Carandini M, Harris KD.

PXK: Characterization of PXK as a protein involved in epidermal growth factor receptor trafficking. (PMID: 20086096) Takeuchi H … Hirata M Molecular and cellular biology 2010 2 3 22 MONaKA, a novel modulator of the plasma membrane Na,K-ATPase. (PMID: 16135750) Mao H … Levitan IB The Journal of neuroscience : the official journal of the Society for Neuroscience 2005 3 4 22 Associations between PXK and TYK2 polymorphisms and systemic lupus erythematosus: a meta-analysis. (PMID: 22592861) Lee YH … Song GG Inflammation research : official journal of the European Histamine Research Society … [et al.] 2012 3 71 A targeted association study in systemic lupus erythematosus identifies multiple susceptibility alleles. (PMID: 20962850) Budarf ML … Rioux JD Genes and immunity 2011 3 40 Early disease onset is predicted by a higher genetic risk for lupus and is associated with a more severe phenotype in lupus patients. (PMID: 20881011) Webb R … Sawalha AH Annals of the rheumatic diseases 2011



QTRT1: Zallot R, Brochier-Armanet C, Gaston KW, et al. Plant, Animal, and Fungal Micronutrient Queuosine Is Salvaged by Members of the DUF2419 Protein Family. ACS Chemical Biology. 2014;9(8):1812-1825. doi:10.1021/cb500278k.

RAB11B: Zhixu Chen, Zezheng Liu, Yang Yang, Zhaoyin Zhu, Ridong Liang, Bin Huang, Di Wu, Lei Yang, Hai Lu, Dadi Jin, Qingchu LiOncotarget. 2018 Jun 1; 9(42): 26770–26786. Published online 2018 Jan 13. doi: 10.18632/oncotarget.24247PMCID: PMC6003561Neil J. Grimsey, Luisa J. Coronel, Isabel Canto Cordova, JoAnn TrejoJ Biol Chem. 2016 Jan 29; 291(5): 2223–2236. Published online 2015 Dec 3. doi: 10.1074/jbc.M115.702993PMCID: PMC4732206Albert S. Reger, Matthew P. Yang, Shizuyo Koide-Yoshida, Elaine Guo,

RAB17: Striz AC, Tuma PL. The GTP-bound and Sumoylated Form of the rab17 Small Molecular Weight GTPase Selectively Binds Syntaxin 2 in Polarized Hepatic WIF-B Cells. The Journal of Biological Chemistry. 2016;291(18):9721-9732. doi:10.1074/jbc.M116.723353.

RASEF: Yang Z-K, Yang J-Y, Xu Z-Z, Yu W-H. DNA Methylation and Uveal Melanoma. Chinese Medical Journal. 2018;131(7):845-851. doi:10.4103/0366-6999.228229.

RASGRP1: Winter S, Martin E, Boutboul D, et al. Loss of RASGRP1 in humans impairs T-cell expansion leading to Epstein-Barr virus susceptibility. EMBO Molecular Medicine. 2018;10(2):188-199. doi:10.15252/emmm.201708292.

RBBP6: Xiao C, Wang Y, Zheng M, et al. RBBP6 increases radioresistance and serves as a therapeutic target for preoperative radiotherapy in colorectal cancer. Cancer Science. 2018;109(4):1075-1087. doi:10.1111/cas.13516.

RBFOX1: Kucherenko MM, Shcherbata HR. Stress-dependent miR-980 regulation of Rbfox1/A2bp1 promotes ribonucleoprotein granule formation and cell survival. Nature Communications. 2018;9:312. doi:10.1038/s41467-017-02757-w.

RBMS1: Leppek K, Stoecklin G. An optimized streptavidin-binding RNA aptamer for purification of ribonucleoprotein complexes identifies novel ARE-binding proteins. Nucleic Acids Research. 2014;42(2):e13. doi:10.1093/nar/gkt956.

RBMS3: Cloning of a human gene closely related to the genes coding for the c-myc single-strand binding proteins. (PMID: 10675610) Penkov D ... Tanaka S Gene 2000 2 3 4 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 39 Deciphering the impact of common genetic variation on lung cancer risk: a genome-wide association study. (PMID: 19654303) Broderick P ... Houlston RS Cancer research 2009 3 39 Genome-wide association with bone mass and geometry in the Framingham Heart Study. (PMID: 17903296) Kiel DP ... Karasik D BMC medical genetics 2007 3 39 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004 3 4 The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). (PMID: 15489334) Gerhard DS ... MGC Project Team Genome research 2004 3 4 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 RBMS3 delays disc degeneration by inhibiting Wnt/β-catenin signaling pathway. (PMID: 32016951) Wang JJ ... Zheng YP European review for medical and pharmacological sciences 2020 3 Increased expression of RBMS3 predicts a favorable prognosis in human gallbladder carcinoma. (PMID: 32627033) Wu Y ... Sun D Oncology reports 2020 3 BCAR1 promotes proliferation and cell growth in lung adenocarcinoma via upregulation of POLR2A. (PMID: 33001583) Mao CG ... Deng B Thoracic cancer 2020 3 Loss of RBMS3 Confers Platinum Resistance in Epithelial Ovarian Cancer via Activation of miR-126-5p/β-catenin/CBP signaling. (PMID: 30279231) Wu G ... Li J Clinical cancer research : an official journal of the American Association for Cancer Research 2019 3 The RNA binding protein RBMS3 inhibits the metastasis of breast cancer by regulating Twist1 expression. (PMID: 30819235) Zhu L ... Wei JF Journal of experimental & clinical cancer research : CR 2019 3 RBMS3 Inhibits the Proliferation and Metastasis of Breast Cancer Cells. (PMID: 28409548) Yang Y ... Ling Y Oncology research 2018 3

RERE: Protein binding of a DRPLA family through arginine-glutamic acid dipeptide repeats is enhanced by extended polyglutamine. (PMID: 10814707) Yanagisawa H ... Yamada M Human molecular genetics 2000 2 3 4 21 De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. (PMID: 27087320) Fregeau B ... Sherr EH American journal of human genetics 2016 3 4 70 Atrophin proteins interact with the Fat1 cadherin and regulate migration and orientation in vascular smooth muscle cells. (PMID: 19131340) Hou R ... Sibinga NE The Journal of biological chemistry 2009 3 4 21 Identification and characterization of novel genes located at the t(1;15)(p36.2;q24) translocation breakpoint in the neuroblastoma cell line NGP. (PMID: 10729226) Amler LC ... Hampton GM Genomics 2000 2 3 4 Genome-wide association study identifies five new schizophrenia loci. (PMID: 21926974) Schizophrenia Psychiatric Genome-Wide Association Study (GWAS) Consortium Nature genetics 2011 3 39 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 39 Variant of TYR and autoimmunity susceptibility loci in generalized vitiligo. (PMID: 20410501) Jin Y ... Spritz RA The New England journal of medicine 2010 3 39

REST: McLeod TCV, Lewis JH, Whelihan K, Bacon CEW. Rest and Return to Activity After Sport-Related Concussion: A Systematic Review of the Literature. Journal of Athletic Training. 2017;52(3):262-287. doi:10.4085/1052-6050-51.6.06.

RETN: Association of variants in RETN with plasma resistin levels and diabetes-related traits in the Framingham Offspring Study. (PMID: 19074981) Hivert MF . Florez JC Diabetes 2009 3 23 43 56 Adiponectin and resistin gene polymorphisms in patients with anorexia nervosa and obesity and its influence on metabolic phenotype. (PMID: 17705672) Krízová J . Haluzík M Physiological research 2008 3 23 43 56 Heritability of serum resistin and its genetic correlation with insulin resistance-related feature

REV3L: Lange SS, Tomida J, Boulware KS, Bhetawal S, Wood RD. The Polymerase Activity of Mammalian DNA Pol ? Is Specifically Required for Cell and Embryonic Viability. McKinnon P, ed. PLoS Genetics. 2016;12(1):e1005759. doi:10.1371/journal.pgen.1005759.

RHOU: Sara Canovas Nunes, Martina Manzoni, Marco Pizzi, Elisa Mandato, Marilena Carrino, Laura Quotti Tubi, Renato Zambello, Fausto Adami, Andrea Visentin, Gregorio Barilà, Livio Trentin, Sabrina Manni, Antonino Neri, Gianpietro Semenzato, Francesco PiazzaBlood Cancer J. 2018 Feb; 8(2): 20. Published online 2018 Feb 13. doi: 10.1038/s41408-018-0053-zPMCID: PMC5811530 Anna E. Dart, Gary M. Box, William Court, Madeline E. Gale, John P. Brown, Sarah E. Pinder, Suzanne A. Eccles, Claire M. Wells.

RIC3: Puschmann A. New Genes Causing Hereditary Parkinson's Disease or Parkinsonism. Current Neurology and Neuroscience Reports. 2017;17(9):66. doi:10.1007/s11910-017-0780-8.

RIMS1: Codner GF, Mianné J, Caulder A, et al. Application of long single-stranded DNA donors in genome editing: generation and validation of mouse mutants. BMC Biology. 2018;16:70. doi:10.1186/s12915-018-0530-7.

RLN3: Calvez J, de Ávila C, Timofeeva E. Sex-specific effects of relaxin-3 on food intake and body weight gain. British Journal of Pharmacology. 2017;174(10):1049-1060. doi:10.1111/bph.13530.

RNASEL: Nguyen-Dumont T, Teo ZL, Hammet F, et al. Is RNASEL:p.Glu265* a modifier of early-onset breast cancer risk for carriers of high-risk mutations BMC Cancer. 2018;18:165. doi:10.1186/s12885-018-4028-z.

RNASET2: RNASET2-deficient cystic leukoencephalopathy resembles congenital cytomegalovirus brain infection. (PMID: 19525954) Henneke M ... Gärtner J Nature genetics 2009 3 4 70 Tumor and metastasis suppression by the human RNASET2 gene. (PMID: 15809705) Acquati F ... Taramelli R International journal of oncology 2005 3 4 21 Mammalian Rh/T2/S-glycoprotein ribonuclease family genes: cloning of a human member located in a region of chromosome 6 (6q27) frequently deleted in human malignancies. (PMID: 9192857) Trubia M ... Taramelli R Genomics 1997 2 3 4 TLR8 Is a Sensor of RNase T2 Degradation Products. (PMID: 31778653) Greulich W ... Hornung V Cell 2019 3 4 Regulation of mitochondrion-associated cytosolic ribosomes by mammalian mitochondrial ribonuclease T2 (RNASET2). (PMID: 30385512) Huang J ... Wang G The Journal of biological chemistry 2018

RPL5: Fancello L, Kampen KR, Hofman IJF, Verbeeck J, Keersmaecker KD. The ribosomal protein gene RPL5 is a haploinsufficient tumor suppressor in multiple cancer types. Oncotarget. 2017;8(9):14462-14478. doi:10.18632/oncotarget.14895.

RPS6KB1: Chen B, Yang L, Zhang R, et al. Hyperphosphorylation of RPS6KB1, rather than overexpression, predicts worse prognosis in non-small cell lung cancer patients. Coleman WB, ed. PLoS ONE. 2017;12(8):e0182891. doi:10.1371/journal.pone.0182891.

RPSAP52: Panagopoulos I, Gorunova L, Brunetti M, et al. Genetic heterogeneity in leiomyomas of deep soft tissue. Oncotarget. 2017;8(30):48769-48781. doi:10.18632/oncotarget.17953.

RPTOR: Yang C, Tsaih S-W, Lemke A, Flister MJ, Thakar MS, Malarkannan S. mTORC1 and mTORC2 differentially promote natural killer cell development. Yokoyama WM, ed. eLife. 2018;7:e35619. doi:10.7554/eLife.35619.

RSU1: Human RSU1 is highly homologous to mouse Rsu-1 and localizes to human chromosome 10. (PMID: 8288261) Tsuda T ... Cutler ML Genomics 1993 2 3 4 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 39 A scan of chromosome 10 identifies a novel locus showing strong association with late-onset Alzheimer disease. (PMID: 16385451) Grupe A ... Goate A American journal of human genetics 2006 3 39 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T ... Sugano S Nature genetics 2004 3 4 The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). (PMID: 15489334) Gerhard DS ... MGC Project Team Genome research 2004

RTEL1: Porreca RM, Glousker G, Awad A, et al. Human RTEL1 stabilizes long G-overhangs allowing telomerase-dependent over-extension. Nucleic Acids Research. 2018;46(9):4533-4545. doi:10.1093/nar/gky173.

RYR2: Gerhard MeissnerJ Gen Physiol. 2017 Dec 4; 149(12): 1065–1089. doi: 10.1085/jgp.201711878PMCID: PMC5715910Zhichao Xiao, Wenting Guo, Siobhan M. Wong King Yuen, Ruiwu Wang, Lin Zhang, Filip Van Petegem, S. R. Wayne ChenPLoS One. 2015; 10(9): e0139058. Published online 2015 Sep 25. doi: 10.1371/journal.pone.0139058PMCID: PMC4583508 Levent Kaya, Barbara Meissner, Maria Christine Riedl, Martin Muik, Christoph Schwarzer, Francesco Ferraguti, Bettina Sarg, Herbert Lindner, Rüdiger Schweigreiter.

S100P: Srinivasa R. Penumutchu, Ruey-Hwang Chou, Chin YuPLoS One. 2014; 9(8): e103947. Published online 2014 Aug 1. doi: 10.1371/journal.pone.0103947PMCID: PMC4118983 Maral E. A. Tabrizi, Tara L. Lancaster, Thamir M. Ismail, Athina Georgiadou, Ankana Ganguly, Jayna J. Mistry, Keqing Wang, Philip S. Rudland, Shakil Ahmad, Stephane R. GrossSci Rep. 2018; 8: 11488. Published online 2018 Jul 31. doi: 10.1038/s41598-018-29852-2PMCID: PMC6068119 Filip Prica, Tomasz Radon, Yuzhu Cheng, Tatjana Crnogorac-

SAMD12: Webb BT, Edwards AC, Wolen AR, et al. Molecular Genetic Influences on Normative and Problematic Alcohol Use in a Population-Based Sample of College Students. Frontiers in Genetics. 2017;8:30. doi:10.3389/fgene.2017.00030.

SCG3: Sun M, Sun T, He Z, Xiong B. Identification of two novel biomarkers of rectal carcinoma progression and prognosis via co-expression network analysis. Oncotarget. 2017;8(41):69594-69609. doi:10.18632/oncotarget.18646.

SCN5A: Hu R-M, Tester DJ, Li R, et al. Mexiletine rescues a mixed biophysical phenotype of the cardiac sodium channel arising from the SCN5A mutation, N406K, found in LQT3 patients. Channels. 2018;12(1):176-186. doi:10.1080/19336950.2018.1475794.

SDC3: Chang BC-C, Hwang L-C, Huang W-H. Positive Association of Metabolic Syndrome with a Single Nucleotide Polymorphism of Syndecan-3 (rs2282440) in the Taiwanese Population. International Journal of Endocrinology. 2018;2018:9282598. doi:10.1155/2018/9282598.

SDHAF4: Belt K, Van Aken O, Murcha M, Millar AH, Huang S. An Assembly Factor Promotes Assembly of Flavinated SDH1 into the Succinate Dehydrogenase Complex[OPEN]. Plant Physiology. 2018;177(4):1439-1452. doi:10.1104/pp.18.00320.

SEC23B: Congenital dyserythropoietic anemia type II (CDAII) is caused by mutations in the SEC23B gene. (PMID: 19621418) Bianchi P . Zanella A Human mutation 2009 2 3 4 23 58 Cloning and functional characterization of mammalian homologues of the COPII component Sec23. (PMID: 8898360) Paccaud JP . Orci L Molecular biology of the cell 1996 2 3 4 58 Germline Heterozygous Variants in SEC23B Are Associated with Cowden Syndrome and Enriched in Apparently Sporadic Thyroid Cancer. (PMID: 26522472) Yehia L .

SELE: Beaufils F, Cmiljanovic N, Cmiljanovic V, et al. 5-(4,6-Dimorpholino-1,3,5-triazin-2-yl)-4-(trifluoromethyl)pyridin-2-amine (PQR309), a Potent, Brain-Penetrant, Orally Bioavailable, Pan-Class I PI3K/mTOR Inhibitor as Clinical Candidate in Oncology. Journa

SELENOF: Serum selenium and single-nucleotide polymorphisms in genes for selenoproteins: relationship to markers of oxidative stress in men from Auckland, New Zealand. Karunasinghe N1, Han DY, Zhu S, Yu J, Lange K, Duan H, Medhora R, Singh N, Kan J, Alzaher W, Chen B, Ko S, Triggs CM, Ferguson LR.

SELENOP: Serum selenium and single-nucleotide polymorphisms in genes for selenoproteins: relationship to markers of oxidative stress in men from Auckland, New Zealand Nishi Karunasinghe,1 Dug Yeo Han,2 Shuotun Zhu,1 Jie Yu,2 Katja Lange,2 He Duan,1 Roxanne Medhora,2 Nabitha Singh,2 James Kan,2 Waseem Alzaher,2 Benson Chen,2 Sarah Ko,2 Christopher M. Triggs,3 and Lynnette R. Fergusoncorresponding author1,2

SERPINA12: Astafev AA, Patel SA, Kondratov RV. Calorie restriction effects on circadian rhythms in gene expression are sex dependent. Scientific Reports. 2017;7:9716. doi:10.1038/s41598-017-09289-9.

SERPINF1: Ziff JL, Crompton M, Powell HRF, et al. Mutations and altered expression of SERPINF1 in patients with famílial otosclerosis. Human Molecular Genetics. 2016;25(12):2393-2403. doi:10.1093/hmg/ddw106.

SERPING1: Gorelik A, Sapir T, Woodruff TM, Reiner O. Serping1/C1 Inhibitor Affects Cortical Development in a Cell Autonomous and Non-cell Autonomous Manner. Frontiers in Cellular Neuroscience. 2017;11:169. doi:10.3389/fncel.2017.00169.

SERPINI1: Tapocik JD, Ceniccola K, Mayo CL, et al. MicroRNAs Are Involved in the Development of Morphine-Induced Analgesic Tolerance and Regulate Functionally Relevant Changes in Serpini1. Frontiers in Molecular Neuroscience. 2016;9:20. doi:10.3389/fnmol.2016.00020

SEZ6L: Causevic M, Dominko K, Malnar M, et al. BACE1-cleavage of Sez6 and Sez6L is elevated in Niemann-Pick type C disease mouse brains. Ohno M, ed. PLoS ONE. 2018;13(7):e0200344. doi:10.1371/journal.pone.0200344.

SH2B3: Ge Z, Gu Y, Xiao L, et al. Co-existence of IL7R high and SH2B3 low expression distinguishes a novel high-risk acute lymphoblastic leukemia with Ikaros dysfunction. Oncotarget. 2016;7(29):46014-46027. doi:10.18632/oncotarget.10014.

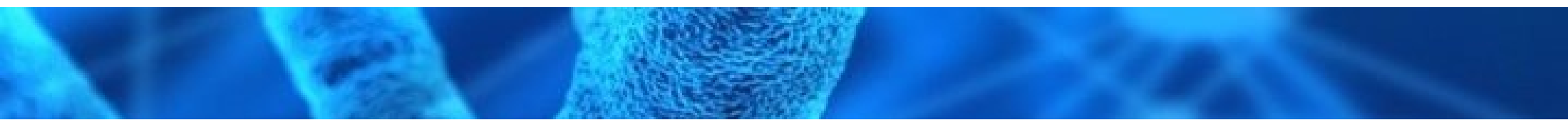
SHBG: Ayd?n B, Winters SJ. Sex Hormone-Binding Globulin in Children and Adolescents. Journal of Clinical Research in Pediatric Endocrinology. 2016;8(1):1-12. doi:10.4274/jcrpe.2764.

SHC4: Genome-wide association analysis of gender differences in major depressive disorder in the Netherlands NESDA and NTR population-based samples. (PMID: 21621269) Aragam N . Pan Y Journal of affective disorders 2011 3 43 56 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE . Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 43 56 Analysis of a Shc family adaptor protein, ShcD/Shc4, that associates with mus

SHISA6: Remco V. Klaassen, Jasper Stroeder, Françoise Coussen, Anne-Sophie Hafner, Jennifer D. Petersen, Cedric Renancio, Leanne J. M. Schmitz, Elisabeth Normand, Johannes C. Lodder, Diana C. Rotaru, Priyanka Rao-Ruiz, Sabine Spijker, Huibert D. Mansvelder, Daniel Choquet, August B. SmitNat Commun. 2016; 7: 10682. Published online 2016 Mar 2. doi: 10.1038/ncomms10682PMCID: PMC4778035Moe Tokue, Kanako Ikami, Seiya Mizuno, Chiyo Takagi, Asuka Miyagi, Ritsuko Takada, Chiyo Noda, Yu Kitadate, Kenshiro

SHMT1: Changwei Dou, Qiuran Xu, Jie Liu, Yufeng Wang, Zhenyu Zhou, Weifeng Yao, Kai Jiang, Jian Cheng, Chengwu Zhang, Kangsheng TuJ Exp Clin Cancer Res. 2019; 38: 70. Published online 2019 Feb 12. doi: 10.1186/s13046-019-1067-5PMCID: PMC6373090Elena V. Abarinov, Anna E. Beaudin, Martha S. Field, Cheryll A. Perry, Robert H. Allen, Sally P. Stabler, Patrick J. StoverJ Nutr. 2013 Jul; 143(7): 1028–1035. Published online 2013 May 22. doi: 10.3945/jn.113.174417PMCID: PMC3681542Donald D. Anderson, Colly

SIRT1: Choi SE, Kwon S, Seok S, et al. Obesity-Linked Phosphorylation of SIRT1 by Casein Kinase 2 Inhibits Its Nuclear Localization and Promotes Fatty Liver. Molecular and Cellular Biology. 2017;37(15):e00006-17. doi:10.1128/MCB.00006-17.



SIRT3: The human SIRT3 protein deacetylase is exclusively mitochondrial. (PMID: 18215119) Cooper HM ... Spelbrink JN The Biochemical journal 2008 2 3 4 Substrates and regulation mechanisms for the human mitochondrial sirtuins Sirt3 and Sirt5. (PMID: 18680753) Schlicker C ... Steegborn C Journal of molecular biology 2008 3 4 21 Reversible lysine acetylation controls the activity of the mitochondrial enzyme acetyl-CoA synthetase 2. (PMID: 16788062) Schwer B ... Verdin E Proceedings of the National Academy of Sciences of the United States of America 2006 3 4 21 A novel VNTR enhancer within the SIRT3 gene, a human homologue of SIR2, is associated with survival at oldest ages. (PMID: 15676284) Bellizzi D ... De Benedictis G Genomics 2005 3 21 39 The human silent information regulator (Sir)2 homologue hSIRT3 is a mitochondrial nicotinamide adenine dinucleotide-dependent deacetylase. (PMID: 12186850) Schwer B ... Verdin E The Journal of cell biology 2002

SIRT6: Ghosh S, Wong SK, Jiang Z, et al. Haploinsufficiency of Trp53 dramatically extends the lifespan of Sirt6-deficient mice. Fässler R, ed. eLife. 2018;7:e32127. doi:10.7554/eLife.32127.

SKIV2L: Aly HH, Suzuki J, Watashi K, et al. RNA Exosome Complex Regulates Stability of the Hepatitis B Virus X-mRNA Transcript in a Non-stop-mediated (NSD) RNA Quality Control Mechanism. The Journal of Biological Chemistry. 2016;291(31):15958-15974. doi:10.1074/j

SLC14A2: Genetic variation in the human urea transporter-2 is associated with variation in blood pressure. (PMID: 11590132) Ranade K ... Botstein D Human molecular genetics 2001 3 23 41 Molecular characterization of a new urea transporter in the human kidney. (PMID: 8647271) Olivès B ... Bailly P FEBS letters 1996 2 3 4 Large-scale genome-wide association studies in East Asians identify new genetic loci influencing metabolic traits. (PMID: 21909109) Kim YJ ... Cho YS Nature genetics 2011 3 41 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 41 Genome-wide association study of hematological and biochemical traits in a Japanese population. (PMID: 20139978) Kamatani Y ... Kamatani N Nature genetics 2010

SLC1A3: Nielson CM, Liu C-T, Smith AV, et al. Novel Genetic Variants Associated With Increased Vertebral Volumetric BMD, Reduced Vertebral Fracture Risk, and Increased Expression of SLC1A3 and EPHB2. Journal of bone and mineral research?: the official journal of

SLC22A2: Human organic cation transporter (OCT1 and OCT2) gene polymorphisms and therapeutic effects of metformin. (PMID: 17111267) Shikata E ... Ieiri I Journal of human genetics 2007 3 4 21 39 Association of the SLC22A1, SLC22A2, and SLC22A3 genes encoding organic cation transporters with diabetic nephropathy and hypertension. (PMID: 20429798) Sallinen R ... Finnish Diabetic Nephropathy Study Group Annals of medicine 2010 3 21 39 Differential transport of platinum compounds by the human organic cation transporter hOCT2 (hSLC22A2). (PMID: 20067471) Burger H ... Wiemer EA British journal of pharmacology 2010 3 21 24 The effects of genetic polymorphisms in the organic cation transporters OCT1, OCT2, and OCT3 on the renal clearance of metformin. (PMID: 19536068) Tzvetkov MV ... Brockmöller J Clinical pharmacology and therapeutics 2009 3 21 39 Effect of genetic variation in the organic cation transporter 2 on the renal elimination of metformin. (PMID: 19483665) Chen Y ... Giacomini KM Pharmacogenetics and genomics 2009 Low HDL concentration in rs2048327-G carriers can predispose men to develop coronary heart disease: Tehran Cardiometabolic genetic study (TCGS). (PMID: 33581269) Najd Hassan Bonab L ... Daneshpour MS Gene 2021 3 Rosai-Dorfman Disease Displays a Unique Monocyte-Macrophage Phenotype Characterized by Expression of OCT2. (PMID: 33177341) Ravindran A ... Mayo Clinic Histiocytosis Working Group The American journal of surgical pathology 2021 The failure of DAC to induce OCT2 expression and its remission by hemoglobin-based nanocarriers under hypoxia in renal cell carcinoma. (PMID: 32206108) Chen L ... Yu L Theranostics 2020 3 Impact of the organic cation transporter 2 inhibitor cimetidine on the single-dose pharmacokinetics of the glucosylceramide synthase inhibitor lucerastat in healthy subjects. (PMID: 31836927) Boof ML ... Dingemanse J European journal of clinical pharmacology 2020 3 Genetic variations of the SLC22A5 gene in the Chinese and Indian populations of Singapore. (PMID: 20208395) Toh DS ... Lee EJ Drug metabolism and pharmacokinetics 2010

SLC22A23: Ekizoglu S, Seven D, Ulutin T, Guliyev J, Buyru N. Investigation of the SLC22A23 gene in laryngeal squamous cell carcinoma. BMC Cancer. 2018;18:477. doi:10.1186/s12885-018-4381-y.

SLC23A1: Genetic variation at the SLC23A1 locus is associated with circulating concentrations of L-ascorbic acid (vitamin C): evidence from 5 independent studies with >15,000 participants. Timpson NJ1, Forouhi NG, Brion MJ, Harbord RM, Cook DG, Johnson P, McConnachie A, Morris RW, Rodriguez S, Luan J, Ebrahim S, Padmanabhan S, Watt G, Bruckdorfer KR, Wareham NJ, Whincup PH, Chanock S, Sattar N, Lawlor DA, Davey Smith G.Eck P, Kwon O, Chen S, Mian O, Levine M. The human sodium-dependent ascorbic acid

SLC23A2: Polymorphisms in the estrogen receptor 1 and vitamin C and matrix metalloproteinase gene families are associated with susceptibility to lymphoma. (PMID: 18636124) Skibola CF ... Smith MT PloS one 2008 3 23 41 54 Genetic variation in the sodium-dependent vitamin C transporters, SLC23A1, and SLC23A2 and risk for preterm delivery. (PMID: 16357110) Erichsen HC ... Chanock SJ American journal of epidemiology 2006 3 23 41 54 Cloning and functional characterization of the human sodium-dependent vitamin C transporters hSVCT1 and hSVCT2. (PMID: 10556521) Daruwala R ... Levine M FEBS letters 1999 3 4 23 54 A human nucleobase transporter-like cDNA (SLC23A1): member of a transporter family conserved from bacteria to mammals. (PMID: 10395795) Hogue DL ... Ling V Genomics 1999 3 4 23 54 Human placental sodium-dependent vitamin C transporter (SVCT2): molecular cloning and transport function. (PMID: 10471399) Rajan DP ... Prasad PD Biochemical and biophysical research communications 1999

SLC24A5: Williams RM, Winkfein RJ, Ginger RS, Green MR, Schnetkamp PP, Wheeler GN. A functional approach to understanding the role of NCKX5 in Xenopus pigmentation. Verheyen EM, ed. PLoS ONE. 2017;12(7):e0180465. doi:10.1371/journal.pone.0180465.

SLC29A3: Alexandre Bolze, Avinash Abhyankar, Audrey V. Grant, Bhavi Patel, Ruchi Yadav, Minji Byun, Daniel Caillez, Jean-Francois Emile, Marçal Pastor-Anglada, Laurent Abel, Anne Puel, Rajgopal Govindarajan, Loic de Pontual, Jean-Laurent CasanovaPLoS One. 2012; 7(1): e29708. Published online 2012 Jan 4. doi: 10.1371/journal.pone.0029708PMCID: PMC3251605 Neil V. Morgan, Mark R. Morris, Hakan Cangul, Diane Gleeson, Anna Straatman-Iwanowska, Nicholas Davies, Stephen Keenan, Shanaz Pasha, Fatimah Rahman

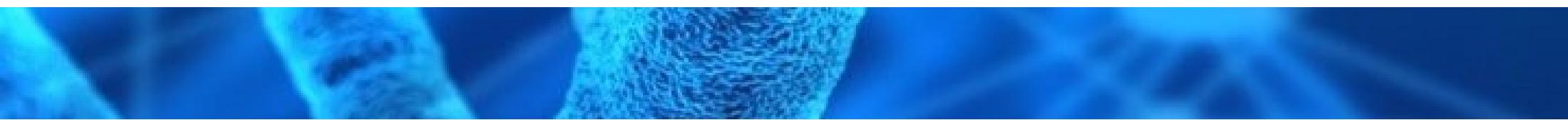
SLC2A14: Mandana Amir Shaghaghi, Haonan Zhouyao, Hongbin Tu, Hani El-Gabalawy, Gary H Crow, Mark Levine, Charles N Bernstein, Peter EckAm J Clin Nutr. 2017 Dec; 106(6): 1508–1513. Published online 2017 Sep 27. doi: 10.3945/ajcn.116.147603PMCID: PMC5698836Colin D Veal, Katherine E Reekie, Johnny C Lorentzen, Peter K Gregersen, Leonid Padyukov, Anthony J BrookesHum Mutat. 2014 Feb; 35(2): 248–256. Published online 2013 Dec 2. doi: 10.1002/humu.22471PMCID: PMC3995011Abhishek Nag, Cristina Venturini, Pi

SLC30A7: Syring KE, Boortz KA, Oeser JK, et al. Combined Deletion of Slc30a7 and Slc30a8 Unmasks a Critical Role for ZnT8 in Glucose-Stimulated Insulin Secretion. Endocrinology. 2016;157(12):4534-4541. doi:10.1210/en.2016-1573.

SLC30A8: A genome-wide association study identifies novel risk loci for type 2 diabetes. (PMID: 17293876) Sladek R ... Froguel P Nature 2007 3 4 39 70 Meta-analysis and functional effects of the SLC30A8 rs13266634 polymorphism on isolated human pancreatic islets. (PMID: 20138556) Cauchi S ... Marchetti P Molecular genetics and metabolism 2010 3 21 39 Autoantibodies to zinc transporter 8 and SLC30A8 genotype stratify type 1 diabetes risk. (PMID: 19590848) Achenbach P ... Ziegler AG Diabetologia 2009 3 21 39 SLC30A8 (ZnT8) Polymorphism is Associated with Young Age at Type 1 Diabetes Onset. (PMID: 18548167) Gohlke H ... Ziegler AG The review of diabetic studies : RDS 2008 3 21 39 Association between anti-ZnT8 autoantibody specificities and SLC30A8 Arg325Trp variant in Japanese patients with type 1 diabetes. (PMID: 18850084) Kawasaki E ... Eguchi K Diabetologia 2008

SLC39A6: Wan X, Kong Z, Chu K, et al. Co-expression analysis revealed PTCH1-3'UTR promoted cell migration and invasion by activating miR-101-3p/SLC39A6 axis in non-small cell lung cancer: implicating the novel function of PTCH1. Oncotarget. 2018;9(4):4798-4813. do

SLC45A2: Park J, Talukder A, Lim S, et al. SLC45A2: A Melanoma Antigen with High Tumor Selectivity and Reduced Potential for Autoimmune Toxicity. Cancer immunology research. 2017;5(8):618-629. doi:10.1158/2326-6066.CIR-17-0051.



SLC52A2: Identification of receptors for pig endogenous retrovirus. (PMID: 12740431) Ericsson TA . Patience C Proceedings of the National Academy of Sciences of the United States of America 2003 2 3 4 56 SLC52A2 [p.P141T] and SLC52A3 [p.N21S] causing Brown-Vialetto-Van Laere Syndrome in an Indian patient: First genetically proven case with mutations in two riboflavin transporters. (PMID: 27702554) Udhayabanu T . Ashokkumar B Clinica chimica acta; international journal of clinical chemistry 2016 3 4

SLC52A3: Identification and functional characterization of rat riboflavin transporter 2. (PMID: 19122205) Yamamoto S . Yuasa H Journal of biochemistry 2009 2 3 4 56 The DNA sequence and comparative analysis of human chromosome 20. (PMID: 11780052) Deloukas P . Rogers J Nature 2001 2 3 4 56 SLC52A3 expression is activated by NF-?B p65/Rel-B and serves as a prognostic biomarker in esophageal cancer. (PMID: 29428966) Long L . Xu LY Cellular and molecular life sciences : CMLS 2018 3 4 56 Functional invo

SLC64A: Lebois LAM, Wolff JD, Ressler KJ. Neuroimaging genetic approaches to Posttraumatic Stress Disorder. Experimental neurology. 2016;284(Pt B):141-152. doi:10.1016/j.expneurol.2016.04.019.

SLC6A2: Verschure DO, Baas F, van Eck-Smit BLF, Somsen GA, Verberne HJ. Polymorphism of SLC6A2 gene does not influence outcome of myocardial 123I-mIBG scintigraphy in patients with chronic heart failure. Journal of Nuclear Cardiology. 2018;25(3):900-906. doi:10.1

SLC6A3: Huang C-C, Kuo S-C, Yeh Y-W, et al. The SLC6A3 gene possibly affects susceptibility to late-onset alcohol dependence but not specific personality traits in a Han Chinese population. Lin ZC, ed. PLoS ONE. 2017;12(2):e0171170. doi:10.1371/journal.pone.01711

SLC7A2: Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 A proximity-dependent biotinylation map of a human cell. (PMID: 34079125) Go CD ... Gingras AC Nature 2021 3 SLC7A2 deficiency promotes hepatocellular carcinoma progression by enhancing recruitment of myeloid-derived suppressors cells. (PMID: 34108444) Xia S ... Liao J Cell death & disease 2021 The prognostic biomarker L-homoarginine is a substrate of the cationic amino acid transporters CAT1, CAT2A and CAT2B. (PMID: 28684763) Chafai A ... Maas R Scientific reports 2017 3 A chimera carrying the functional domain of the orphan protein SLC7A14 in the backbone of SLC7A2 mediates trans-stimulated arginine transport. (PMID: 22787143) Jaenecke I ... Closs EI The Journal of biological chemistry 2012 3 Regulation of arginine transport and metabolism by protein kinase Calpha in endothelial cells: stimulation of CAT2 transporters and arginase activity. (PMID: 20430034) Visigalli R ... Dall'Asta V Journal of molecular and cellular cardiology 2010 3 Polyamines Impair Immunity to Helicobacter pylori by Inhibiting L-Arginine Uptake Required for Nitric Oxide Production. (PMID: 20600019) Chaturvedi R ... Wilson KT Gastroenterology 2010 3 Rapamycin stimulates arginine influx through CAT2 transporters in human endothelial cells. (PMID: 17397797) Visigalli R ... Dall'Asta V Biochimica et biophysica acta 2007 21 System y+ arginine transport and NO production in peripheral blood mononuclear cells in pregnancy and preeclampsia. (PMID: 16344361) McCord N ... Redman C Hypertension (Dallas, Tex. : 1979) 2006 21 The arginine-arginase balance in asthma and lung inflammation. (PMID: 16458291) Zimmermann N ... Rothenberg ME European journal of pharmacology 2006 21 Macrophages require distinct arginine catabolism and transport systems for proliferation and for activation. (PMID: 16703566) Yeramian A ... Celada A European journal of immunology 2006 24 Arginine in asthma and lung inflammation. (PMID: 15465795) King NE ... Zimmermann N The Journal of nutrition 2004 21 CAT2 arginine transporter deficiency significantly reduces iNOS-mediated NO production in astrocytes. (PMID: 12675924) Manner CK ... MacLeod CL Journal of neurochemistry 2003 21 Coinduction of endothelial nitric oxide synthase and arginine recycling enzymes in aorta of diabetic rats. (PMID: 11384198) Oyadomari S ... Mori M Nitric oxide : biology and chemistry 2001 21 Physiological cyclic stretch directs L-arginine transport and metabolism to collagen synthesis in vascular smooth muscle. (PMID: 10973927) Durante W ... Schafer AI FASEB journal : official publication of the Federation of American Societies for Experimental Biology 2000 21 Overexpression of dominant negative mutant hepatocyte nuclear factor (HNF)-1alpha inhibits arginine-induced insulin secretion in MIN6 cells. (PMID: 10440133) Tanizawa Y ... Oka Y Diabetologia 1999

SLC9A3R1: NHE-RF, a regulatory cofactor for Na(+)-H+ exchange, is a common interactor for merlin and ERM (MERM) proteins. (PMID: 9430655) Murthy A . Ramesh V The Journal of biological chemistry 1998 2 3 4 23 56 Identification of EBP50: A PDZ-containing phosphoprotein that associates with members of the ezrin-radixin-moesin family. (PMID: 9314537) Reczek D . Bretscher A The Journal of cell biology 1997 2 3 4 23 56 NHERF1 mutations and responsiveness of renal parathyroid hormone. (PMID: 18784102) Karim

SLC9A9: Zhang-James Y, Middleton FA, Sagvolden T, Faraone SV. Differential expression of SLC9A9 and interacting molecules in the hippocampus of rat models for attention-deficit/hyperactivity disorder. Developmental neuroscience. 2012;34(2-3):218-227. doi:10.1159/

SMAD3: Tang PM-K, Zhou S, Meng X-M, et al. Smad3 promotes cancer progression by inhibiting E4BP4-mediated NK cell development. Nature Communications. 2017;8:14677. doi:10.1038/ncomms14677.

SMOC2: Homozygosity mapping and candidate prioritization identify mutations, missed by whole-exome sequencing, in SMOC2, causing major dental developmental defects. (PMID: 22152679) Bloch-Zupan A ... Dollfus H American journal of human genetics 2011 3 4 70 Identification of a novel smooth muscle associated protein, smap2, upregulated during neointima formation in a rat carotid endarterectomy model. (PMID: 12031507) Nishimoto S ... Komurasaki T Biochimica et biophysica acta 2002 2 3 4 Recessive oligodontia linked to a homozygous loss-of-function mutation in the SMOC2 gene. (PMID: 23317772) Alfawaz S ... Kelsell DP Archives of oral biology 2013 3 70 Genome-wide association study of generalized vitiligo in an isolated European founder population identifies SMOC2, in close proximity to IDDM8. (PMID: 19890347) Birlea SA ... Spritz RA The Journal of investigative dermatology 2010 3 39 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010

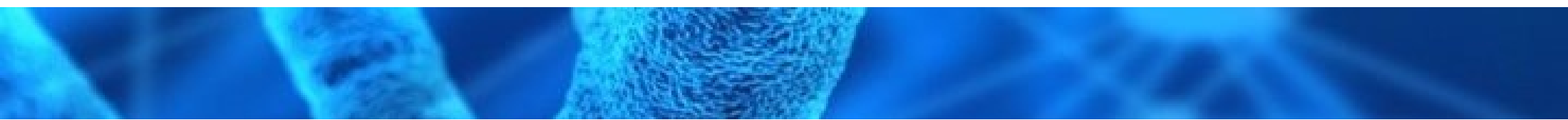
SMYD3: Kim J-M, Kim K, Schmidt T, et al. Cooperation between SMYD3 and PC4 drives a distinct transcriptional program in cancer cells. Nucleic Acids Research. 2015;43(18):8868-8883. doi:10.1093/nar/gkv874.

SNAP25: Weber P, Batoulis H, Rink KM, et al. Electrostatic anchoring precedes stable membrane attachment of SNAP25/SNAP23 to the plasma membrane. Brunger AT, ed. eLife. 2017;6:e19394. doi:10.7554/eLife.19394.

SNRPN: Thompson LW, Morrison KD, Shirran SL, et al. Neurochondrin interacts with the SMN protein suggesting a novel mechanism for spinal muscular atrophy pathology. Journal of Cell Science. 2018;131(8):jcs211482. doi:10.1242/jcs.211482.

SOCS3: Polymorphisms near SOCS3 are associated with obesity and glucose homeostasis traits in Hispanic Americans from the Insulin Resistance Atherosclerosis Family Study. (PMID: 19083014) Talbert ME . Bowden DW Human genetics 2009 3 23 43 56 Common polymorphisms in SOCS3 are not associated with body weight, insulin sensitivity or lipid profile in normal female twins. (PMID: 16402267) Jamshidi Y . O'Dell SD Diabetologia 2006 3 23 43 56 A new high affinity binding site for suppressor of cytokine sig

SOD1: Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. (PMID: 8446170) Rosen DR ... Deng HX Nature 1993 2 3 4 39 69 Age and founder effect of SOD1 A4V mutation causing ALS. (PMID: 19176896) Saeed M ... Siddique T Neurology 2009 3 21 39 69 Sixteen novel mutations in the Cu/Zn superoxide dismutase gene in amyotrophic lateral sclerosis: a decade of discoveries, defects and disputes. (PMID: 14506936) Andersen PM ... Brown RH Amyotrophic lateral sclerosis and other motor neuron disorders : official publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases 2003 3 4 21 69 Familial ALS is associated with mutations in all exons of SOD1: a novel mutation in exon 3 (Gly72Ser). (PMID: 9455977) Orrell RW ... deBelleroche JS Journal of the neurological sciences 1997 3 4 21 69 Screening of SOD1, FUS and TARDBP genes in patients with amyotrophic lateral sclerosis in central-southern China. (PMID: 27604643) Hou L ... Shen L Scientific reports 2016



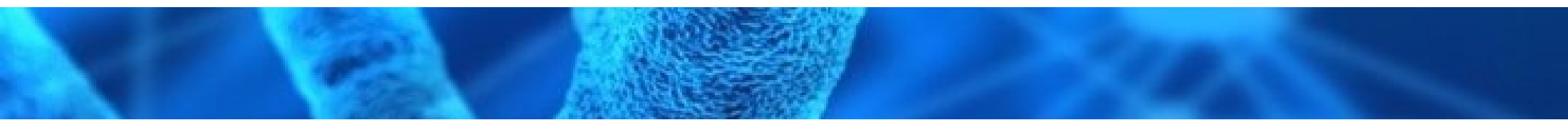
SOD2: The polymorphism of manganese superoxide dismutase is associated with diabetic nephropathy in Japanese type 2 diabetic patients. (PMID: 12624725) Nomiyama T ... Kawamori R Journal of human genetics 2003 3 4 21 39 69 A functional polymorphism in the manganese superoxide dismutase gene and diabetic nephropathy. (PMID: 17192491) Möllsten A ... Dahlquist G Diabetes 2007 3 21 39 69 Genotype-activity relationship for Mn-superoxide dismutase, glutathione peroxidase 1 and catalase in humans. (PMID: 16538174) Bastaki M ... Holland N Pharmacogenetics and genomics 2006 3 21 39 69 The mitochondrial superoxide dismutase A16V polymorphism in the cardiomyopathy associated with hereditary haemochromatosis. (PMID: 15591282) Valenti L ... Fargion S Journal of medical genetics 2004 3 21 39 69 Missense polymorphisms in three oxidative-stress enzymes (GSTP1, SOD2, and GPX1) and dyskinesias in Russian psychiatric inpatients from Siberia. (PMID: 20041472) Al Hadithy AF ... Loonen AJ Human psychopharmacology 2010 The mitochondrial superoxide dismutase A16V polymorphism in the cardiomyopathy associated with hereditary haemochromatosis. (PMID: 15591282) Valenti L ... Fargion S Journal of medical genetics 2004 3 21 39 69 The Ala-9Val polymorphism in the mitochondrial targeting sequence (MTS) of the manganese superoxide dismutase gene is not associated with juvenile-onset asthma. (PMID: 15589819) Gurel A ... Elyas H Clinical biochemistry 2004 3 21 39 The -9Ala/-9Val polymorphism in the mitochondrial targeting sequence of the manganese superoxide dismutase gene (MnSOD) is associated with age among Hispanics with colorectal carcinoma. (PMID: 11836586) Stoehlmacher J ... Lenz HJ Oncology reports 2002 3 21 39 Protein stability of mitochondrial superoxide dismutase SOD2 is regulated by USP36. (PMID: 21268071) Kim MS ... Baek KH Journal of cellular biochemistry 2011 3 4 Macrophages that survive hyperoxia exposure have higher superoxide dismutase activities in their mitochondria. (PMID: 20204772) Kokubo K ... Kobayashi H Advances in experimental medicine and biology 2010 Regulation of Anti-Apoptotic SOD2 and BIRC3 in Periodontal Cells and Tissues. (PMID: 33435582) Rath-Deschner B ... Damanaki A International journal of molecular sciences 2021 3 General Control of Amino Acid Synthesis 5-Like 1-Mediated Acetylation of Manganese Superoxide Dismutase Regulates Oxidative Stress in Diabetic Kidney Disease. (PMID: 33680286) Lv T ... Wan Q Oxidative medicine and cellular longevity 2021 3 LncRNA MAFG-AS1 affects the tumorigenesis of breast cancer cells via the miR-574-5p/SOD2 axis. (PMID: 33989902) Dai J ... Yan M Biochemical and biophysical research communications 2021 3 Rewiring of the Human Mitochondrial Interactome during Neuronal Reprogramming Reveals Regulators of the Respirasome and Neurogenesis. (PMID: 31536960) Moutaoufik MT ... Babu M iScience 2019 3 Associations Between Genetic Polymorphisms in the VEGFA, ACE, and SOD2 Genes and Susceptibility to Diabetic Nephropathy in the Han Chinese. (PMID: 31524543) Luo Y ... Peng H Genetic testing and molecular biomarkers 2019 3 USP36 protects proximal tubule cells from ischemic injury by stabilizing c-Myc and SOD2. (PMID: 30975468) Liu Q ... Wan Q Biochemical and biophysical research communications 2019 3 PARP1-LSD1 functional interplay controls transcription of SOD2 that protects human pro-inflammatory macrophages from death under an oxidative condition. (PMID: 30529301) Tokarz P ... Robaszkiewicz A Free radical biology & medicine 2019 3 miR-330-3p promotes lung cancer cells invasion, migration, and metastasis by directly targeting hSOD2b. (PMID: 30192404) Shen L ... Wang F Biotechnology and applied biochemistry 2019

SOD3: Manganese and extracellular superoxide dismutase polymorphisms and risk for asbestosis. (PMID: 19636420) Franko A ... Dolzan V Journal of biomedicine & biotechnology 2009 3 21 39 The structure of human extracellular copper-zinc superoxide dismutase at 1.7 A resolution: insights into heparin and collagen binding. (PMID: 19289127) Antonyuk SV ... Hasnain SS Journal of molecular biology 2009 3 4 21 Genetic polymorphisms modifying oxidative stress are associated with disease activity in rheumatoid arthritis patients. (PMID: 19242068) Bohanec Grabar P ... Dolzan V Disease markers 2009 3 21 39 Extracellular superoxide dismutase haplotypes are associated with acute lung injury and mortality. (PMID: 18948423) Arcaroli JJ ... Crapo JD American journal of respiratory and critical care medicine 2009 3 21 39 Association of extracellular superoxide dismutase (SOD3) Ala40Thr gene polymorphism with pre-eclampsia complicated by severe fetal growth restriction. (PMID: 19108943) Rosta K ... Vér A European journal of obstetrics, gynecology, and reproductive biology 2009

SORBS1: Song L, Chang R, Dai C, et al. SORBS1 suppresses tumor metastasis and improves the sensitivity of cancer to chemotherapy drug. Oncotarget. 2017;8(6):9108-9122. doi:10.18632/oncotarget.12851.

SORL1: Thonberg H, Chiang H-H, Lilius L, et al. Identification and description of three families with famílial Alzheimer disease that segregate variants in the SORL1gene. Acta Neuropathologica Communications. 2017;5:43. doi:10.1186/s40478-017-0441-9.

SPAG16: Mehra S, Kapur S, Ganesh S. Association between a Tetranucleotide Repeat Polymorphism of SPAG16 Gene and Cataract in Male Children. Journal of Biomarkers. 2013;2013:810395. doi:10.1155/2013/810395.



SPATA2L: A "double adaptor" method for improved shotgun library construction. (PMID: 8619474) Andersson B . Gibbs RA Analytical biochemistry 1996 2 3 56 Architecture of the human interactome defines protein communities and disease networks. (PMID: 28514442) Huttlin EL . Harper JW Nature 2017 3 56 RNA-binding activity of TRIM25 is mediated by its PRY/SPRY domain and is required for ubiquitination. (PMID: 29117863) Choudhury NR . Michlewski G BMC biology 2017 3 56 SPATA2 Links CYLD to LUBAC, Activates

SPHKAP: GWAS of Osteoarthritis Biomarkers Serum Hyaluronic Acid and Cartilage Oligomeric Matrix Protein Implicates FOXN4, ETV6, KIAA1217, ZNF521, and CSGALNACT1 +1 Coan HB,1 Choudary A,2 Nicolella DP,1 Dyer TD, 1 Curran JE,1 Carless MC,1 Kumar S,1 Kent Jr. JW,1 Almasy LA,1 Goring HHH,1 Blangero J,1 Mahaney MC, 1 Havill LM Sphingosine kinase interacting protein is an A-kinase anchoring protein specific for type I cAMP-dependent protein kinase. (PMID: 20394097) Kovanich D ... Scholten A Chembiochem : a European journal of chemical biology 2010 3 4 22 Cloning and characterization of a protein kinase A anchoring protein (AKAP)-related protein that interacts with and regulates sphingosine kinase 1 activity. (PMID: 12080051) Lacaná E ... Spiegel S The Journal of biological chemistry 2002 2 3 4 Prediction of the coding sequences of unidentified human genes. XIX. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. (PMID: 11214970) Nagase T ... Ohara O DNA research : an international journal for rapid publication of reports on genes and genomes 2000 2 3 4 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 40 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020

SPINK5: Furio L, Pampalakis G, Michael IP, Nagy A, Sotiropoulou G, Hovnanian A. KLK5 Inactivation Reverses Cutaneous Hallmarks of Netherton Syndrome. Gallo RL, ed. PLoS Genetics. 2015;11(9):e1005389. doi:10.1371/journal.pgen.1005389.

SPIRE2: Prediction of the coding sequences of unidentified human genes. XX. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. (PMID: 11347906) Nagase T . Ohara O DNA research : an international journal for rapid publication of reports on genes and genomes 2001 2 3 4 58 DNA damage induces nuclear actin filament assembly by Formin -2 and Spire-½ that promotes efficient DNA repair. [corrected]. (PMID: 26287480) Belin BJ . Mullins RD eLife 2015 3 4 58 Spir

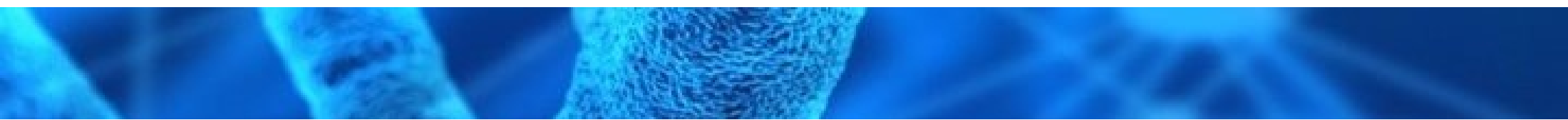
SPOCK3: Wang L-Y, Cui J-J, Zhu T, et al. Biomarkers identified for prostate cancer patients through genome-scale screening. Oncotarget. 2017;8(54):92055-92063. doi:10.18632/oncotarget.20739.

SPSB4: Okumura F, Joo-Okumura A, Obara K, et al. Ubiquitin ligase SPSB4 diminishes cell repulsive responses mediated by EphB2. Chernoff J, ed. Molecular Biology of the Cell. 2017;28(24):3532-3541. doi:10.1091/mbc.E17-07-0450.

SPTLC3: Cloning and initial characterization of a new subunit for mammalian serine-palmitoyltransferase. (PMID: 17023427) Hornemann T ... von Eckardstein A The Journal of biological chemistry 2006 2 3 4 54 Genome-wide association study of personality traits in bipolar patients. (PMID: 21368711) Alliey-Rodriguez N ... Gershon ES Psychiatric genetics 2011 3 41 54 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 41 54 Variation at the NFATC2 locus increases the risk of thiazolidinedione-induced edema in the Diabetes REduction Assessment with ramipril and rosiglitazone Medication (DREAM) study. (PMID: 20628086) Bailey SD ... DREAM investigators Diabetes care 2010 3 41 54 A genome-wide perspective of genetic variation in human metabolism. (PMID: 20037589) Illig T ... Suhre K Nature genetics 2010

SRPX: A gene (SRPX) encoding a sushi-repeat-containing protein is deleted in patients with X-linked retinitis pigmentosa. (PMID: 8634708) Meindl A . Meitinger T Human molecular genetics 1995 2 3 4 23 56 Identification of a novel gene, ETX1 from Xp21.1, a candidate gene for X-linked retintis pigmentosa (RP3). (PMID: 8634709) Dry KL . Thomson K Human molecular genetics 1995 2 3 4 56 The DNA sequence of the human X chromosome. (PMID: 15772651) Ross MT . Bentley DR Nature 2005 3 4 56 The status, qual

STAT2: Arimoto K, Löchte S, Stoner SA, et al. STAT2 is an essential adaptor in USP18-mediated suppression of type I interferon signaling. Nature structural & molecular biology. 2017;24(3):279-289. doi:10.1038/nsmb.3378.



STAT3: Ethan L. Morgan, Christopher W. Wasson, Lucy Hanson, David Kealy, Ieisha Pentland, Victoria McGuire, Cinzia Scarpini, Nicholas Coleman, J. Simon C. Arthur, Joanna L. Parish, Sally Roberts, Andrew MacdonaldPLoS Pathog. 2018 Apr; 14(4): e1006975. Published online 2018 Apr 9. doi: 10.1371/journal.ppat.1006975PMCID: PMC5908086Michaela Galoczova, Philip Coates, Borivoj VojtesekCell Mol Biol Lett. 2018; 23: 12. Published online 2018 Mar 22. doi: 10.1186/s11658-018-0078-0PMCID: PMC5863838Teresa A.

STAT4: McWilliams IL, Rajbhandari R, Nozell S, Benveniste E, Harrington LE. STAT4 controls GM-CSF production by both Th1 and Th17 cells during EAE. Journal of Neuroinflammation. 2015;12:128. doi:10.1186/s12974-015-0351-3.

STAT6: Wang C, Zhu C, Wei F, et al. Nuclear Localization and Cleavage of STAT6 Is Induced by Kaposi's Sarcoma-Associated Herpesvirus for Viral Latency. Ling PD, ed. PLoS Pathogens. 2017;13(1):e1006124. doi:10.1371/journal.ppat.1006124.

STK39: Bazot Q, Paschos K, Allday MJ. Epstein-Barr Virus (EBV) Latent Protein EBNA3A Directly Targets and Silences the STK39 Gene in B Cells Infected by EBV. Sandri-Goldin RM, ed. Journal of Virology. 2018;92(7):e01918-17. doi:10.1128/JVI.01918-17.

STN1: OB fold-containing protein 1 (OBFC1), a human homolog of yeast Stn1, associates with TPP1 and is implicated in telomere length regulation. (PMID: 19648609) Wan M ... Liu D The Journal of biological chemistry 2009 2 3 4 23 Mutations in STN1 cause Coats plus syndrome and are associated with genomic and telomere defects. (PMID: 27432940) Simon AJ ... Somech R The Journal of experimental medicine 2016 3 4 74 CST for the grand finale of telomere replication. (PMID: 23851344) Chen LY ... Lingner J Nucleus (Austin, Tex.) 2013 3 4 Human CST promotes telomere duplex replication and general replication restart after fork stalling. (PMID: 22863775) Stewart JA ... Price CM The EMBO journal 2012 3 4 The human CST complex is a terminator of telomerase activity. (PMID: 22763445) Chen LY ... Lingner J Nature 2012

STON2: Sun X, Zhang W, Li H, et al. Stonin 2 Overexpression is Correlated with Unfavorable Prognosis and Tumor Invasion in Epithelial Ovarian Cancer. International Journal of Molecular Sciences. 2017;18(8):1653. doi:10.3390/ijms18081653.

STXBP5L: Ehrmann I, Dalgliesh C, Liu Y, et al. The Tissue-Specific RNA Binding Protein T-STAR Controls Regional Splicing Patterns of Neurexin Pre-mRNAs in the Brain. Bickmore WA, ed. PLoS Genetics. 2013;9(4):e1003474. doi:10.1371/journal.pgen.1003474.

SUPT3H: A. Rasim Barutcu, Phillip W. L. Tai, Hai Wu, Jonathan A. R. Gordon, Troy W. Whitfield, Jason R. Dobson, Anthony N. Imbalzano, Jane B. Lian, André J. van Wijnen, Janet L. Stein, Gary S. SteinNucleic Acids Res. 2014 Sep 15; 42(16): 10360–10372. Published online 2014 Aug 12. doi: 10.1093/nar/gku712PMCID: PMC4176362Sarah J Rice, Guillaume Aubourg, Antony K Sorial, David Almarza, Maria Tselepi, David J Deehan, Louise N Reynard, John LoughlinHum Mol Genet. 2018 Oct 1; 27(19): 3464–3474. Published

SYK: Weis V, Königsberger S, Amler S, Wienands J, Kiefer F. Unperturbed Immune Function despite Mutation of C-Terminal Tyrosines in Syk Previously Implicated in Signaling and Activity Regulation. Molecular and Cellular Biology. 2017;37(21):e00216-17. doi:10.11

SYT1: Siao W, Wang P, Voigt B, Hussey PJ, Baluska F. Arabidopsis SYT1 maintains stability of cortical endoplasmic reticulum networks and VAP27-1-enriched endoplasmic reticulum–plasma membrane contact sites. Journal of Experimental Botany. 2016;67(21):6161-6171.

TAP2: Knox B, Wang Y, Rogers LJ, et al. A Functional SNP in the 3'-UTR of TAP2 Gene Interacts with MicroRNA Hsa-miR-1270 to Suppress the Gene Expression. Environmental and molecular mutagenesis. 2018;59(2):134-143. doi:10.1002/em.22159.

TBC1D1: Hargett SR, Walker NN, Keller SR. Rab GAPs AS160 and Tbc1d1 play nonredundant roles in the regulation of glucose and energy homeostasis in mice. American Journal of Physiology - Endocrinology and Metabolism. 2016;310(4):E276-E288. doi:10.1152/ajpendo.0034

TCF2: Quan X, Zhang L, Li Y, Liang C. TCF2 Attenuates FFA-Induced Damage in Islet ?-Cells by Regulating Production of Insulin and ROS. International Journal of Molecular Sciences. 2014;15(8):13317-13332. doi:10.3390/ijms150813317.

TCF4: Kennedy AJ, Rahn EJ, Paulukaitis BS, et al. Tcf4 Regulates Synaptic Plasticity, DNA Methylation, and Memory Function. Cell reports. 2016;16(10):2666-2685. doi:10.1016/j.celrep.2016.08.004. Elucidating the genetic basis of social interaction and isolation. Day FR1, Ong KK1, Perry JRB2. MRC Epidemiology Unit, Box 285 Institute of Metabolic Science, Cambridge Biomedical Campus, University of Cambridge School of Clinical Medicine, Cambridge, CB2 0QQ, UK.

TCF7L2: Variant of transcription factor 7-like 2 (TCF7L2) gene confers risk of type 2 diabetes. (PMID: 16415884) Grant SF ... Stefansson K Nature genetics 2006 3 4 21 39 69 Relationship of five type 2 diabetes candidate gene polymorphisms to the age at diagnosis of diabetes in the Slovakian population. (PMID: 20437825) Kozarova M ... Tkac I Bratislavske lekarske listy 2010 3 21 39 Transcription factor 7-like 2-gene polymorphism is related to fasting C peptide in latent autoimmune diabetes in adults (LADA). (PMID: 19533015) Szepietowska B ... Szelachowska M Acta diabetologica 2010 3 21 39 Interactions between TCF7L2 genotype and growth hormone-induced changes in glucose homeostasis in small for gestational age children. (PMID: 19473183) de Kort SW ... Hokken-Koelega AC Clinical endocrinology 2010 3 21 39 TCF7L2 rs7903146-macronutrient interaction in obese individuals' responses to a 10-wk randomized hypoenergetic diet. (PMID: 20032493) Grau K ... Sørensen TI The American journal of clinical nutrition 2010 De novo variants in TCF7L2 are associated with a syndromic neurodevelopmental disorder. (PMID: 34003604) Dias C ... Rodan LH American journal of medical genetics. Part A 2021 3 69 Evidence of interaction between type 2 diabetes susceptibility genes and dietary fat intake for adiposity and glucose homeostasis-related phenotypes. (PMID: 20215779) Ruchat SM ... Pérusse L Journal of nutrigenetics and nutrigenomics 2009 3 39 A proximity-dependent biotinylation map of a human cell. (PMID: 34079125) Go CD ... Gingras AC Nature 2021 3 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 Association of Combined TCF7L2 and KCNQ1 Gene Polymorphisms with Diabetic Micro- and Macrovascular Complications in Type 2 Diabetes Mellitus. (PMID: 33752320) Rattanatham R ... Settasatian C Diabetes & metabolism journal 2021 Genetic susceptibility to type 2 diabetes is associated with reduced prostate cancer risk. (PMID: 20203524) Pierce BL ... Ahsan H Human heredity 2010 3 39 RIG-I regulates myeloid differentiation by promoting TRIM25-mediated ISGylation. (PMID: 32513696) Wu SF ... Chen S Proceedings of the National Academy of Sciences of the United States of America 2020 3 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 T Cell Factor 4 Is Involved in Papillary Thyroid Carcinoma via Regulating Long Non-Coding RNA HCP5. (PMID: 33371788) Wang R ... Li G Technology in cancer research & treatment 2020 3 Molecular Characterization of the Oncogene BTF3 and Its Targets in Colorectal Cancer. (PMID: 33644029) Wang H ... Li X Frontiers in cell and developmental biology 2020

TCHH: Common variants in the trichohyalin gene are associated with straight hair in Europeans. (PMID: 19896111) Medland SE . Martin NG American journal of human genetics 2009 3 23 45 58 The structure of human trichohyalin. Potential multiple roles as a functional EF-hand-like calcium-binding protein, a cornified cell envelope precursor, and an intermediate filament-associated (cross-linking) protein. (PMID: 7685034) Lee SC . Steinert PM The Journal of biological chemistry 1993 3 4 23 58 Trichohya

TERC: TERC and TERT gene mutations in patients with bone marrow failure and the significance of telomere length measurements. (PMID: 18931339) Du HY . Bessler M Blood 2009 3 23 45 58 Cell cycle-dependent recruitment of telomerase RNA and Cajal bodies to human telomeres. (PMID: 16319170) Jády BE . Kiss T Molecular biology of the cell 2006 2 3 23 58 Genetic variants in eleven telomere-associated genes and the risk of incident cardio/cerebrovascular disease: The Women's Genome Health Study. (PMID: 2

TERF2: Human telomeres contain two distinct Myb-related proteins, TRF1 and TRF2. (PMID: 9326950) Broccoli D ... de Lange T Nature genetics 1997 2 3 4 22 Arginine methylation regulates telomere length and stability. (PMID: 19596784) Mitchell TR ... Zhu XD Molecular and cellular biology 2009 3 4 22 Polymorphisms in telomere-associated genes, breast cancer susceptibility and prognosis. (PMID: 19766477) Varadi V ... Försti A European journal of cancer (Oxford, England : 1990) 2009 3 22 40 Polymorphisms in telomere maintenance genes and risk of lung cancer. (PMID: 19773453) Choi JE ... Park JY Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 22 40 Genetic variation in five genes important in telomere biology and risk for breast cancer. (PMID: 17848914) Savage SA ... Garcia-Closas M British journal of cancer 2007

TERT: Bordeira Gaspar T, Sá A, Lopes JM, Sobrinho-Simões M, Soares P, Vinagre J. Telomere Maintenance Mechanisms in Cancer. Genes. 2018;9(5):241. doi:10.3390/genes9050241.

TET1: Jing Wang, Dawei Zhang, Juan Du, Chi Zhou, Zhi Li, Xing Liu, Gang Ouyang, Wuhan XiaoNucleic Acids Res. 2017 Dec 15; 45(22): 12700–12714. Published online 2017 Sep 28. doi: 10.1093/nar/gkx869PMCID: PMC5727443Jianing Zhong, Xianfeng Li, Wanshi Cai, Yan Wang, Shanshan Dong, Jie Yang, Jian'an Zhang, Nana Wu, Yuanyuan Li, Fengbiao Mao, Cheng Zeng, Jinyu Wu, Xingzhi Xu, Zhong Sheng SunNucleic Acids Res. 2017 Jan 25; 45(2): 672–684. Published online 2016 Oct 12. doi: 10.1093/nar/gkw919PMCID: PMC53

TFAP2B: Zainolabidin N, Kamath SP, Thanawalla AR, Chen AI. Distinct Activities of Tfap2A and Tfap2B in the Specification of GABAergic Interneurons in the Developing Cerebellum. Frontiers in Molecular Neuroscience. 2017;10:281. doi:10.3389/fnmol.2017.00281.

TGFB1: Ojiaku CA, Yoo EJ, Panettieri RA. Transforming Growth Factor ?1 Function in Airway Remodeling and Hyperresponsiveness. The Missing Link? American Journal of Respiratory Cell and Molecular Biology. 2017;56(4):432-442. doi:10.1165/rcmb.2016-0307TR.

TGFB2: Ahmed ASI, Bose GC, Huang L, Azhar M. Generation of mice carrying a knockout-first and conditional-ready allele of transforming growth factor beta2 gene. Genesis (New York, NY?: 2000). 2014;52(9):817-826. doi:10.1002/dvg.22795.

TGFBR3: Julian T. Schwartze, Simone Becker, Elpidoforos Sakkas, ?ukasz A. Wujak, Gero Niess, Jakob Usemann, Frank Reichenberger, Susanne Herold, István Vadász, Konstantin Mayer, Werner Seeger, Rory E. MortyJ Biol Chem. 2014 Feb 7; 289(6): 3262–3275. Published online 2013 Dec 17. doi: 10.1074/jbc.M113.541052PMCID: PMC3916529 Daniel M. DeLaughter, Cynthia R. Clark, Danos C. Christodoulou, Christine E. Seidman, H. Scott Baldwin, J. G. Seidman, Joey V. BarnettPLoS One. 2016; 11(8): e0159710.

THADA: Moraru A, Cakan-Akdogan G, Strassburger K, et al. THADA Regulates the Organismal Balance between Energy Storage and Heat Production. Developmental Cell. 2017;41(1):72-81.e6. doi:10.1016/j.devcel.2017.03.016.

THBD: Van Mens TE, Liang H-PH, Basu S, et al. Variable phenotypic penetrance of thrombosis in adult mice after tissue-selective and temporally controlled Thbd gene inactivation. Blood Advances. 2017;1(15):1148-1158. doi:10.1182/bloodadvances.2017005058.

THBS2: Wang X, Zhang L, Li H, Sun W, Zhang H, Lai M. THBS2 is a Potential Prognostic Biomarker in Colorectal Cancer. Scientific Reports. 2016;6:33366. doi:10.1038/srep33366.

TKT: Xu IM-J, Lai RK-H, Lin S-H, et al. Transketolase counteracts oxidative stress to drive cancer development. Proceedings of the National Academy of Sciences of the United States of America. 2016;113(6):E725-E734. doi:10.1073/pnas.1508779113.

TKTL1: Molecular cloning of tissue-specific transcripts of a transketolase-related gene: implications for the evolution of new vertebrate genes. (PMID: 8838793) Coy JF . Poustka A Genomics 1996 2 3 4 23 58 Mutations in the transketolase-like gene TKTL1: clinical implications for neurodegenerative diseases, diabetes and cancer. (PMID: 15991799) Coy JF . Schubert P Clinical laboratory 2005 3 4 23 58 Lactate-dehydrogenase 5 is overexpressed in non-small cell lung cancer and correlates with the expres

TLR3: Zinngrebe J, Rieser E, Taraborrelli L, et al. LUBAC deficiency perturbs TLR3 signaling to cause immunodeficiency and autoinflammation. The Journal of Experimental Medicine. 2016;213(12):2671-2689. doi:10.1084/jem.20160041.

TLR4: Iotzova-Weiss G, Freiberger SN, Johansen P, et al. TLR4 as a negative regulator of keratinocyte proliferation. Brandner JM, ed. PLoS ONE. 2017;12(10):e0185668. doi:10.1371/journal.pone.0185668.

TM2D3: Jakobsdottir J, van der Lee SJ, Bis JC, et al. Rare Functional Variant in TM2D3is Associated with Late-Onset Alzheimer's Disease. Haines JL, ed. PLoS Genetics. 2016;12(10):e1006327. doi:10.1371/journal.pgen.1006327.

TM9SF2: Pacheco AR, Lazarus JE, Sit B, et al. CRISPR Screen Reveals that EHEC's T3SS and Shiga Toxin Rely on Shared Host Factors for Infection. Rubin EJ, ed. mBio. 2018;9(3):e01003-18. doi:10.1128/mBio.01003-18.

TMEM18: Larder R, Sim MFM, Gulati P, et al. Obesity-associated gene TMEM18 has a role in the central control of appetite and body weight regulation. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(35):9421-9426. doi:10.10

TMEM229B: Popova EY, Salzberg AC, Yang C, Zhang SS-M, Barnstable CJ. Identification and prediction of alternative transcription start sites that generate rod photoreceptor-specific transcripts from ubiquitously expressed genes. Lewin AS, ed. PLoS ONE. 2017;12(6):e0

TMEM241: Rodríguez A, Gonzalez L, Ko A, et al. Molecular characterization of the lipid GWAS signal on chromosome 18q11.2 implicates HNF4A-mediated regulation of the TMEM241 gene. Arteriosclerosis, thrombosis, and vascular biology. 2016;36(7):1350-1355. doi:10.1161

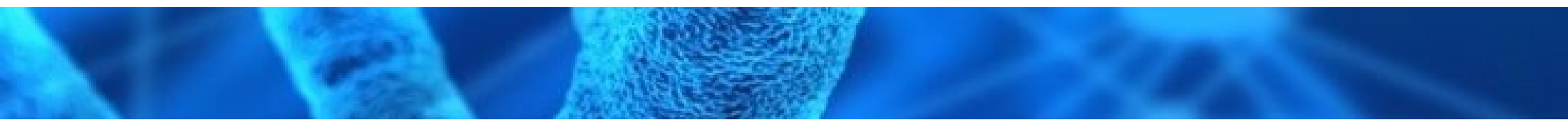
TMEM45B: Metri R, Mohan A, Nsengimana J, et al. Identification of a gene signature for discriminating metastatic from primary melanoma using a molecular interaction network approach. Scientific Reports. 2017;7:17314. doi:10.1038/s41598-017-17330-0.

TMOD1: Cheng C, Nowak RB, Biswas SK, Lo W-K, FitzGerald PG, Fowler VM. Tropomodulin 1 Regulation of Actin Is Required for the Formation of Large Paddle Protrusions Between Mature Lens Fiber Cells. Investigative Ophthalmology & Visual Science. 2016;57(10):4084-40

TMPO: M J Davies, T F Slater Biochem J. 1986 Dec 15; 240(3): 789–795.PMCID: PMC1147488 Lin Zhang, Gan Wang, Shiwen Chen, Jun Ding, Shiming Ju, Heli Cao, Hengli TianWorld J Surg Oncol. 2016; 14: 267. Published online 2016 Oct 19. doi: 10.1186/s12957-016-1018-yPMCID: PMC5069786 Federico Abascal, Michael L. Tress, Alfonso Valencia Bioinformatics. 2015 Jul 15; 31(14): 2257–2261. Published online 2015 Mar 2. doi: 10.1093/bioinformatics/btv132PMCID: PMC4495291

TMPRSS2: Identification of TMPRSS2 as a Susceptibility Gene for Severe 2009 Pandemic A(H1N1) Influenza and A(H7N9) Influenza. Cheng Z1, Zhou J2, To KK3, Chu H2, Li C1, Wang D1, Yang D1, Zheng S4, Hao K5, Bossé Y6, Obeidat M7, Brandsma CA8, Song YQ9, Chen Y4, Zheng BJ2, Li L4, Yuen KY3. Kim H, Datta A, Talwar S, Saleem SN, Mondal D, Abdel-Mageed AB. Estradiol-ER?2 signaling axis confers growth and migration of CRPC cells through TMPRSS2-ETV5 gene fusion. Oncotarget. 2017;8(38):62820-62833. doi:10.18632/oncotarget.11355.

TNF: Association of tumor necrosis factor polymorphisms with asthma and serum total IgE. (PMID: 14681301) Shin HD ... Park CS Human molecular genetics 2004 3 21 39 69 Cytokine gene polymorphisms: association with psoriatic arthritis susceptibility and severity. (PMID: 12746914) Balding J ... FitzGerald O Arthritis and rheumatism 2003 3 4 39 69 A polymorphism that affects OCT-1 binding to the TNF promoter region is associated with severe malaria. (PMID: 10369255) Knight JC ... Kwiatkowski D Nature genetics 1999 3 4 39 69 Interleukin (IL)-1beta, IL-1 receptor antagonist, IL-6, IL-8, IL-10, and tumor necrosis factor alpha gene polymorphisms in patients with febrile seizures. (PMID: 20486195) Chou IC ... Tsai FJ Journal of clinical laboratory analysis 2010 3 21 39 Genetic susceptibility to different clinical forms of tuberculosis in the Peruvian population. (PMID: 20188863) Taype CA ... Shaw MA Infection, genetics and evolution : journal of molecular epidemiology and evolutionary genetics in infectious diseases 2010 3 21 39 Genetic variation in chromosomal translocation breakpoint and immune function genes and risk of non-Hodgkin lymphoma. (PMID: 20087644) Fernberg P ... Ekström Smedby K Cancer causes & control : CCC 2010 3 21 39 Association between polymorphisms in human tumor necrosis factor-alpha (--308) and -beta (252) genes and development of gestational diabetes mellitus. (PMID: 20189261) Montazeri S ... Radhakrishnan AK Diabetes research and clinical practice 2010 3 21 39 Tumor necrosis factor alpha (-308), interleukin-6 (-174) and interleukin-10 (-1082) gene polymorphisms in polycystic ovary syndrome. (PMID: 20189706) Vural P ... Akgül C European journal of obstetrics, gynecology, and reproductive biology 2010 3 21 39 Genetic association of Toll-like-receptor 4 and tumor necrosis factor-alpha polymorphisms with Plasmodium falciparum blood infection levels. (PMID: 20307689) Basu M ... Sengupta S Infection, genetics and evolution : journal of molecular epidemiology and evolutionary genetics in infectious diseases 2010 3 21 39 The tumor necrosis factor polymorphism TNF (-308) is associated with susceptibility to meningococcal sepsis, but not with lethality. (PMID: 19242354) Read RC ... Wilson AG Critical care medicine 2009 3 21 39 The relation of adiponectin and tumor necrosis factor alpha levels between endothelial nitric oxide synthase, angiotensin-converting enzyme, transforming growth factor beta, and tumor necrosis factor alpha gene polymorphism in adrenal incidentalomas. (PMID: 19498318) Harman E ... Yilmaz C Journal of endocrinological investigation 2009 3 21 39 Association of IL10 and other immune response- and obesity-related genes with prostate cancer in CLUE II. (PMID: 19267370) Wang MH ... Platz EA The Prostate 2009 3 21 39 Analysis of inflammation- and atherosclerosis-related gene polymorphisms in branch retinal vein occlusion. (PMID: 19347053) Steinbrugger I ... Weger M Molecular vision 2009 3 21 39 Single nucleotide polymorphisms in TNF-alpha, TNFR2 gene and TNF-alpha production in Asian Indians. (PMID: 19811435) Gupta R ... Das SN Immunological investigations 2009 3 21 39 Genetic variants in immunoregulatory genes and risk for childhood lymphomas. (PMID: 19508433) Andrie E ...



Petridou ET European journal of haematology 2009 3 21 39 Cytokine gene polymorphisms in Behçet's disease and their association with clinical and laboratory findings. (PMID: 19796538) Dilek K ... Oral HB Clinical and experimental rheumatology 2009 3 21 39 Effect of desensitization in solid organ transplant recipients depends on some cytokines genes polymorphism. (PMID: 19332120) Lobashevsky AL ... Turrentine MW Transplant immunology 2009 3 21 39 Interleukin-1 receptor antagonist and tumour necrosis factor-alpha gene polymorphisms in Turkish patients with allergic contact dermatitis. (PMID: 19706048) Ertam I ... Ozkinay F Contact dermatitis 2009 3 21 39 TNFA and IL10 gene polymorphisms are not associated with periodontitis in Brazilians. (PMID: 19771178) Moreira PR ... Dutra WO The open dentistry journal 2009 3 21 39 [Effect of TNF-alpha gene polymorphism on outcome of thalidomide-based regimens for multiple myeloma]. (PMID: 19954656) DU J ... Hou J Zhonghua xue ye xue za zhi = Zhonghua xueyexue zazhi 2009 3 21 39 Long-range linkage on chromosome 6p of VEGF, FKBP5, HLA and TNF alleles associated with transplant rejection. (PMID: 19233472) Chen Y ... Hutchinson IV Molecular immunology 2009 3 21 39 Association of polymorphisms in Tumor Necrosis Factor Alpha and Beta genes with increased risk for oral cancer. (PMID: 19528505) Yapijakis C ... Vairaktaris E Anticancer research 2009 3 21 39 Association between inflammatory gene polymorphisms and coronary artery disease in an Indian population. (PMID: 18157711) Banerjee I ... Ganesh S Journal of thrombosis and thrombolysis 2009 3 21 39 Association of gastric disease with polymorphisms in the inflammatory-related genes IL-1B, IL-1RN, IL-10, TNF and TLR4. (PMID: 19295440) Murphy G ... O'Sullivan M European journal of gastroenterology & hepatology 2009 3 21 39 The role of tumor necrosis factor (TNF)-alpha and TNF receptor polymorphisms in susceptibility to ankylosing spondylitis. (PMID: 19772798) Chatzikyriakidou A ... Drosos AA Clinical and experimental rheumatology 2009 3 21 39 Variation in inflammation-related genes and risk of incident nonfatal myocardial infarction or ischemic stroke. (PMID: 17981284) Bis JC ... Psaty BM Atherosclerosis 2008 3 21 39 Association of cystic fibrosis transmembrane conductance regulator (CFTR) mutation/variant/haplotype and tumor necrosis factor (TNF) promoter polymorphism in hyperlipidemic pancreatitis. (PMID: 17981921) Chang YT ... Wong JM Clinical chemistry 2008 3 21 39 Polymorphisms of the TNF, CD14, and HSPA1B genes in patients with acute alcohol-induced pancreatitis. (PMID: 18580445) Tukiainen E ... Repo H Pancreas 2008 3 21 39 TNF-alpha gene promoter polymorphism at nucleotide -308 and the inflammatory response and oxidative stress induced by cardiac surgery: role of heart failure and medical treatment. (PMID: 18450464) Galiñanes M ... Hadjinikolaou L European journal of cardio-thoracic surgery : official journal of the European Association for Cardio-thoracic Surgery 2008 3 21 39 New approach reveals CD28 and IFNG gene interaction in the susceptibility to cervical cancer. (PMID: 18337305) Guzman VB ... Morgun A Human molecular genetics 2008 3 21 39 Impact of genetic polymorphisms of leptin and TNF-alpha on rosiglitazone response in Chinese patients with type 2 diabetes. (PMID: 18438653) Liu HL ... Liu ZQ European journal of clinical pharmacology 2008 3 21 39 Association between the level of circulating bioactive tumor necrosis factor alpha and the tumor necrosis factor alpha gene polymorphism at -308 in patients with rheumatoid arthritis treated with a tumor necrosis factor alpha inhibitor. (PMID: 18438841) Marotte H ... Miossec P Arthritis and rheumatism 2008 3 21 39 TNF, IFNG, IL6, IL10 and TGFB1 gene polymorphisms in South and Southeast Brazil. (PMID: 18680515) Visentainer JE ... de Souza CA International journal of immunogenetics 2008 3 21 39 Cytokine Genes TNF, IL1A, IL1B, IL6, IL1RN and IL10, and childhood-onset mood disorders. (PMID: 18832862) Misener VL ... International Consortium for Childhood-Onset Mood Disorders Neuropsychobiology 2008 3 21 39 [Association of immune system gene polymorphisms with quantitative features which are pathogenetically important in chronic viral hepatitis]. (PMID: 18610832) Goncharova IA ... pyzurev VP Molekuliarnaia biologiia 2008 3 21 39 IL10 polymorphisms are associated with airflow obstruction in severe alpha1-antitrypsin deficiency. (PMID: 17690329) Demeo DL ... Silverman EK American journal of respiratory cell and molecular biology 2008 3 21 39 Polymorphisms in cytokine genes and their association with acute rejection and recurrence of hepatitis B in Chinese liver transplant recipients. (PMID: 18375254) Xie HY ... Zheng SS Archives of medical research 2008 3 21 39 Tumor necrosis factor-alpha and interleukin-10 gene promoter polymorphisms in Turkish rheumatoid arthritis patients. (PMID: 18427872) Ates O ... Topal-Sarikaya A Clinical rheumatology 2008 3 21 39 Interleukin-10 (IL10) and tumor necrosis factor alpha (TNF) gene polymorphisms in Parkinson's disease patients. (PMID: 18362084) Bialecka M ... Droździk M Parkinsonism & related disorders 2008 3 21 39 Interleukin-10 and tumor necrosis factor-alpha gene polymorphisms in tuberculosis. (PMID: 18071881) Ates O ... Topal-Sarikaya A Journal of clinical immunology 2008 3 21 39 A single tumour necrosis factor haplotype influences the response to adalimumab in rheumatoid arthritis. (PMID: 17673491) Miceli-Richard C ... Mariette X Annals of the rheumatic diseases 2008 3 21 39 Genetic polymorphisms of RANTES, IL1-A, MCP-1 and TNF-A genes in patients with prostate cancer. (PMID: 19099590) Sáenz-López P ... Ruiz-Cabello F BMC cancer 2008 3 21 39 [Association between susceptibility of endometriosis and the gene polymorphism of tumor necrosis factor]. (PMID: 18754435) Lu DH ... Deng L Zhonghua yi xue za zhi 2008 3 21 39 Inflammatory system gene polymorphism and the risk of stroke: a case-control study in an Indian population. (PMID: 18158110) Banerjee I ... Ganesh S Brain research bulletin 2008 3 21 39 Influence of functional interleukin 10/tumor necrosis factor-alpha polymorphisms on interferon-alpha, IL-10, and regulatory T cell population in patients with systemic lupus erythematosus receiving antimalarial treatment. (PMID: 18597405) López P ... Suárez A The Journal of rheumatology 2008 3 21 39 Association

between LTA, TNF and AGER polymorphisms and late diabetic complications. (PMID: 18575614) Lindholm E ... Agardh CD PloS one 2008 3 21 39 Polymorphisms of tumor-necrosis factor-alpha - 308 and lymphotoxin-alpha + 250: possible modulation of susceptibility to apoptosis in chronic lymphocytic leukemia and non-Hodgkin lymphoma mononuclear cells. (PMID: 19021060) Jevtovic-Stoimenov T ... Pavlović S Leukemia & lymphoma 2008 3 21 39 Genetic polymorphisms of adiponectin and tumor necrosis factor-alpha and nonalcoholic fatty liver disease in Chinese people. (PMID: 18565022) Wong VW ... Chan HL Journal of gastroenterology and hepatology 2008 3 21 39 Cytokine gene polymorphisms in heavy drinkers with and without decompensated liver disease: a case-control study. (PMID: 19086955) Gleeson D ... Duff GW The American journal of gastroenterology 2008 3 21 39 Combined effect of tumour necrosis factor-alpha and interleukin-13 polymorphisms on bronchial hyperresponsiveness in Korean children with asthma. (PMID: 18341619) Kim HB ... Hong SJ Clinical and experimental allergy : journal of the British Society for Allergy and Clinical Immunology 2008 3 21 39 Influence of cytokine gene polymorphisms on IgA nephropathy. (PMID: 18300111) Bantis C ... Ivens K Renal failure 2008 3 21 39 Cytokine gene polymorphism in kidney transplantation--impact of TGF-beta 1, TNF-alpha and IL-6 on graft outcome. (PMID: 18158121) Nikolova PN ... Naumova EJ Transplant immunology 2008 3 21 39 Epistatic interactions are critical to gene-association studies: PAI-1 and risk for mortality after burn injury. (PMID: 18182917) Barber RC ... Horton JW Journal of burn care & research : official publication of the American Burn Association 2008 3 21 39 TNF and sTNFR1/2 plasma levels in ALS patients. (PMID: 18083240) Cereda C ... Ceroni M Journal of neuroimmunology 2008 3 21 39 Associations between tumor necrosis factor-alpha and lymphotoxin-alpha polymorphisms and idiopathic recurrent miscarriage. (PMID: 18299433) Zammiti W ... Mahjoub T Reproduction (Cambridge, England) 2008 3 21 39 Polymorphisms of tumor necrosis factor-alpha, interleukin-10, cytochrome P450 3A5 and ABCB1 in Chinese liver transplant patients treated with immunosuppressant tacrolimus. (PMID: 17568575) Li D ... Zhang GL Clinica chimica acta; international journal of clinical chemistry 2007 3 21 39 Frequency distribution of cytokine gene polymorphisms in the healthy North Indian population. (PMID: 17257312) Kaur G ... Mehra NK Tissue antigens 2007 3 21 39 Effect of the 252A>G polymorphism of the lymphotoxin-alpha gene on inflammatory markers of response to cigarette smoking in Korean healthy men. (PMID: 17113059) Jang Y ... Lee JH Clinica chimica acta; international journal of clinical chemistry 2007 3 21 39 Genetic polymorphisms in immunoresponse genes TNFA, IL6, IL10, and TLR4 are associated with recurrent acute otitis media. (PMID: 17908769) Emonts M ... Sanders EA Pediatrics 2007 3 21 39 Association between human African trypanosomiasis and the IL6 gene in a Congolese population. (PMID: 16720107) Courtin D ... Garcia A Infection, genetics and evolution : journal of molecular epidemiology and evolutionary genetics in infectious diseases 2007 3 21 39 TNF polymorphism and bronchoalveolar lavage cell TNF-alpha levels in chronic beryllium disease and beryllium sensitization. (PMID: 17208287) Sato H ... Maier LA The Journal of allergy and clinical immunology 2007 3 21 39 IL-10 and IL-6 gene polymorphisms as potential host susceptibility factors in Brucellosis. (PMID: 17544674) Budak F ... Oral HB Cytokine 2007 3 21 39 Cytokine gene polymorphisms in Colombian patients with systemic lupus erythematosus. (PMID: 17711410) Guarnizo-Zuccardi P ... Vasquez G Tissue antigens 2007 3 21 39 Tumour necrosis factor alpha promoter polymorphisms and etanercept therapy in juvenile idiopathic arthritis. (PMID: 16951943) Schmeling H ... Horneff G Rheumatology international 2007 Dual proteome-scale networks reveal cell-specific remodeling of the human interactome. (PMID: 33961781) Huttlin EL ... Gygi SP Cell 2021 3 MIND bomb 2 prevents RIPK1 kinase activity-dependent and -independent apoptosis through ubiquitylation of cFLIPL. (PMID: 33469115) Nakabayashi O ... Nakano H Communications biology 2021 3 An NF-κB-responsive long noncoding RNA, PINT, regulates TNF-α gene transcription by scaffolding p65 and EZH2. (PMID: 34405442) Ye M ... Zhou R FASEB journal : official publication of the Federation of American Societies for Experimental Biology 2021 3 Polymorphisms of the TNF, LTA, and TNFRSF1B genes are associated with onsets of menarche and menopause in US women of European ancestry. (PMID: 34595982) Dvornyk V ... Deng HW Annals of human biology 2021 3 Meta-analysis for association of TNFA-308(G > A) SNP with vitiligo susceptibility. (PMID: 34673212) Giri PS ... Dwivedi M Gene 2022

TNFAIP3: Das T, Chen Z, Hendriks RW, Kool M. A20/Tumor Necrosis Factor ?-Induced Protein 3 in Immune Cells Controls Development of Autoinflammation and Autoimmunity: Lessons from Mouse Models. Frontiers in Immunology. 2018;9:104. doi:10.3389/fimmu.2018.00104.

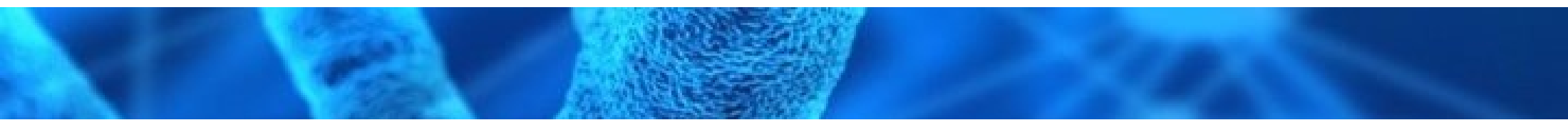
TNFRSF11A: Mutations in TNFRSF11A, affecting the signal peptide of RANK, cause familial expansile osteolysis. (PMID: 10615125) Hughes AE ... Anderson DM Nature genetics 2000 2 3 4 21 70 Human osteoclast-poor osteopetrosis with hypogammaglobulinemia due to TNFRSF11A (RANK) mutations. (PMID: 18606301) Guerrini MM ... Frattini A American journal of human genetics 2008 3 4 21 70 A homologue of the TNF receptor and its ligand enhance T-cell growth and dendritic-cell function. (PMID: 9367155) Anderson DM ... Galibert L Nature 1997 2 3 4 21 Gene-gene interactions in RANK/RANKL/OPG system influence bone mineral density in postmenopausal women. (PMID: 19896533) Zupan J ... Marc J The Journal of steroid biochemistry and molecular biology 2010 3 21 39 Genetic variations in genes encoding RANK, RANKL, and OPG in rheumatoid arthritis: a case-control study. (PMID: 20231205) Assmann G ... Wieczorek S The Journal of rheumatology 2010

TNFRSF1B: Li H, Anderson SK. Association of TNFRSF1B Promoter Polymorphisms with Human Disease: Further Studies Examining T-Regulatory Cells Are Required. Frontiers in Immunology. 2018;9:443. doi:10.3389/fimmu.2018.00443.

TNFSF14: LIGHT/LTβR signaling regulates self-renewal and differentiation of hematopoietic and leukemia stem cells. (PMID: 33594067) Höpner SS ... Ochsenbein AF Nature communications 2021 3 Increased level of LIGHT/TNFSF14 is associated with survival in aneurysmal subarachnoid hemorrhage. (PMID: 33492677) Schranz D ... Csecsei P Acta neurologica Scandinavica 2021 Genomic characterization of LIGHT reveals linkage to an immune response locus on chromosome 19p13.3 and distinct isoforms generated by alternate splicing or proteolysis. (PMID: 11673523) Granger SW ... Ware CF Journal of immunology (Baltimore, Md. : 1950) 2001 3 4 21 LIGHT, a new member of the TNF superfamily, and lymphotoxin alpha are ligands for herpesvirus entry mediator. (PMID: 9462508) Mauri DN ... Ware CF Immunity 1998 2 3 4 Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. (PMID: 21833088) International Multiple Sclerosis Genetics Consortium ... Compston A Nature 2011 3 39 CXCR3 binding chemokine and TNFSF14 over expression in bladder urothelium of patients with ulcerative interstitial cystitis. (PMID: 20096889) Ogawa T ... Nishizawa O The Journal of urology 2010 3 21 An approach based on a genome-wide association study reveals candidate loci for narcolepsy. (PMID: 20677014) Shimada M ... Tokunaga K Human genetics 2010

TNFSF4: Manku H, Langefeld CD, Guerra SG, et al. Trans-Ancestral Studies Fine Map the SLE-Susceptibility Locus TNFSF4 . McCarthy MI, ed. PLoS Genetics. 2013;9(7):e1003554. doi:10.1371/journal.pgen.1003554.

TNKS: Two new Loci for body-weight regulation identified in a joint analysis of genome-wide association studies for early-onset extreme obesity in French and german study groups. (PMID: 20421936) Scherag A ... Meyre D PLoS genetics 2010 3 22 40 Polymorphisms in telomere-associated genes, breast cancer susceptibility and prognosis. (PMID: 19766477) Varadi V ... Försti A European journal of cancer (Oxford, England : 1990) 2009 3 22 40 Polymorphisms in telomere maintenance genes and risk of lung cancer. (PMID: 19773453) Choi JE ... Park JY Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 22 40 Zinc binding catalytic domain of human tankyrase 1. (PMID: 18436240) Lehtiö L ... Weigelt J Journal of molecular biology 2008 3 4 22 NuMA is a major acceptor of poly(ADP-ribosyl)ation by tankyrase 1 in mitosis. (PMID: 16076287) Chang W ... Smith S The Biochemical journal 2005



TP53: Pathway-based approaches to imaging genetics association studies: Wnt signaling, GSK3beta substrates and major depression. (PMID: 20219685) Inkster B ... Matthews PM NeuroImage 2010 3 39 Effect of MDM2 SNP309 and p53 codon 72 polymorphisms on lung cancer risk and survival among non-smoking Chinese women in Singapore. (PMID: 20219101) Chua HW ... Seow A BMC cancer 2010 3 39 Critical role for Ser20 of human p53 in the negative regulation of p53 by Mdm2. (PMID: 10202144) Unger T ... Haupt Y The EMBO journal 1999 3 21 Multilevel proteomics reveals host perturbations by SARS-CoV-2 and SARS-CoV. (PMID: 33845483) Stukalov A ... Pichlmair A Nature 2021 3 Metformin induces apoptosis and inhibits migration by activating the AMPK/p53 axis and suppressing PI3K/AKT signaling in human cervical cancer cells. (PMID: 33236135) Chen YH ... Hsiao YH Mo The RNF20/40 complex regulates p53-dependent gene transcription and mRNA splicing. (PMID: 31152661) Wu C ... Yu X Journal of molecular cell biology 2020 3 Human colorectal cancer derived-MSCs promote tumor cells escape from senescence via P53/P21 pathway. (PMID: 31218648) Li G ... Feng Y Clinical & translational oncology : official publication of the Federation of Spanish Oncology Societies and of the National Cancer Institute of Mexico 2020 3 Tumor-derived CK1α mutations enhance MDMX inhibition of p53. (PMID: 31462704) Liu X ... Chen J Oncogene 2020 TP53 mutations predict disease control in metastatic colorectal cancer treated with cetuximab-based chemotherapy. (PMID: 19367287) Oden-Gangloff A ... Frebourg T British journal of cancer 2009 3 21 39 70 Germline TP53 mutations in BRCA1 and BRCA2 mutation-negative French Canadian breast cancer families. (PMID: 17541742) Arcand SL ... Tonin PN Breast cancer research and treatment 2008 3 21 39 70 BRCA1, BRCA2 and TP53 mutations in very early-onset breast cancer with associated risks to relatives. (PMID: 16644204) Lalloo F ... Evans DG European journal of cancer (Oxford, England : 1990) 2006 3 21 39 70 TP53, BRCA1, and BRCA2 tumor suppressor genes are not commonly mutated in survivors of Hodgkin's disease with second primary neoplasms. (PMID: 14673037) Nichols KE ... Diller L Journal of clinical oncology : official journal of the American Society of Clinical Oncology 2003 3 21 39 70 Hereditary TP53 codon 292 and somatic P16INK4A codon 94 mutations in a Li-Fraumeni syndrome family. (PMID: 10484981) Güran S ... Imirzalioğlu N Cancer genetics and cytogenetics 1999

TPH1: Wigner P, Czarny P, Synowiec E, et al. Association between single nucleotide polymorphisms of TPH1 and TPH2 genes, and depressive disorders. Journal of Cellular and Molecular Medicine. 2018;22(3):1778-1791. doi:10.1111/jcmm.13459.

TPH2: Donner NC, Kubala KH, Hassell Jr. JE, et al. Two models of inescapable stress increase tph2 mRNA expression in the anxiety-related dorsomedial part of the dorsal raphe nucleus. Neurobiology of Stress. 2018;8:68-81. doi:10.1016/j.ynstr.2018.01.003.

TPTE2P1: Xu S, Zhan M, Wang J. Epithelial-to-mesenchymal transition in gallbladder cancer: from clinical evidence to cellular regulatory networks. Cell Death Discovery. 2017;3:17069-. doi:10.1038/cddiscovery.2017.69.

TRABD2B: Yamada Y, Sakuma J, Takeuchi I, et al. Identification of eight genetic variants as novel determinants of dyslipidemia in Japanese by exome-wide association studies. Oncotarget. 2017;8(24):38950-38961. doi:10.18632/oncotarget.17159.

TRAPPC9: Li C, Luo X, Zhao S, et al. COPI–TRAPPII activates Rab18 and regulates its lipid droplet association. The EMBO Journal. 2017;36(4):441-457. doi:10.15252/embj.201694866.

TREM2: Jay TR, von Saucken VE, Landreth GE. TREM2 in Neurodegenerative Diseases. Molecular Neurodegeneration. 2017;12:56. doi:10.1186/s13024-017-0197-5.

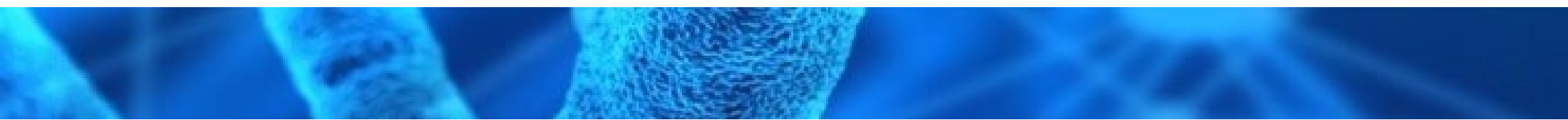
TRIB3: Aimé P, Sun X, Zareen N, et al. Trib3 Is Elevated in Parkinson’s Disease and Mediates Death in Parkinson’s Disease Models. The Journal of Neuroscience. 2015;35(30):10731-10749. doi:10.1523/JNEUROSCI.0614-15.2015.

TRIM29: Li Q, Lin L, Tong Y, et al. TRIM29 negatively controls antiviral immune response through targeting STING for degradation. Cell Discovery. 2018;4:13. doi:10.1038/s41421-018-0010-9.

TRIM47: Han Y, Tian H, Chen P, Lin Q. TRIM47 overexpression is a poor prognostic factor and contributes to carcinogenesis in non-small cell lung carcinoma. Oncotarget. 2017;8(14):22730-22740. doi:10.18632/oncotarget.15188.

TRIM66: Chen Y, Guo Y, Yang H, et al. TRIM66 overexpresssion contributes to osteosarcoma carcinogenesis and indicates poor survival outcome. Oncotarget. 2015;6(27):23708-23719.

TRPM6: Chubanov V, Ferioli S, Wisnowsky A, et al. Epithelial magnesium transport by TRPM6 is essential for prenatal development and adult survival. Lewis RS, ed. eLife. 2016;5:e20914. doi:10.7554/eLife.20914.



TRPM7: Abiria SA, Krapivinsky G, Sah R, et al. TRPM7 senses oxidative stress to release Zn2+ from unique intracellular vesicles. Proceedings of the National Academy of Sciences of the United States of America. 2017;114(30):E6079-E6088. doi:10.1073/pnas.170738011

TRPS1: Dana Elster, Marie Tollot, Karin Schlegelmilch, Alessandro Ori, Andreas Rosenwald, Erik Sahai, Björn von EyssNat Commun. 2018; 9: 3115. Published online 2018 Aug 6. doi: 10.1038/s41467-018-05370-7Correction in: Nat Commun. 2018; 9: 3781.PMCID: PMC6079100Anne-Sophie Bach, Danielle Derocq, Valérie Laurent-Matha, Philippe Montcourrier, Salwa Salwa Sebti, Béatrice Orsetti, Charles Theillet, Céline Gongora, Sophie Pattingre, Eva Ibing, Pascal Roger, Laetitia K. Linares, Thomas Reinheckel, Guilla

TSHZ2: Analysis of TSHZ2 and TSHZ3 genes in congenital pelvi-ureteric junction obstruction. (PMID: 19745106) Jenkins D ... Woolf AS Nephrology, dialysis, transplantation : official publication of the European Dialysis and Transplant Association - European Renal Association 2010 3 4 40 Personalized smoking cessation: interactions between nicotine dose, dependence and quit-success genotype score. (PMID: 20379614) Rose JE ... Uhl GR Molecular medicine (Cambridge, Mass.) 2010 3 40 Variation at the NFATC2 locus increases the risk of thiazolidinedione-induced edema in the Diabetes REduction Assessment with ramipril and rosiglitazone Medication (DREAM) study. (PMID: 20628086) Bailey SD ... DREAM investigators Diabetes care 2010 3 40 FE65 binds Teashirt, inhibiting expression of the primate-specific caspase-4. (PMID: 19343227) Kajiwara Y ... Buxbaum JD PloS one 2009 3 4 Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. (PMID: 19862010) Ganesh SK ... Lin JP Nature genetics 2009

TSLP: Verstraete K, Peelman F, Braun H, et al. Structure and antagonism of the receptor complex mediated by human TSLP in allergy and asthma. Nature Communications. 2017;8:14937. doi:10.1038/ncomms14937.

TTN: Yu-Huan Shih, Alexey V. Dvornikov, Ping Zhu, Xiao Ma, Maengjo Kim, Yonghe Ding, Xiaolei XuDevelopment. 2016 Dec 15; 143(24): 4713–4722. doi: 10.1242/dev.139246PMCID: PMC5201027Ali M. Tabish, Valerio Azzimato, Aris Alexiadis, Byambajav Buyandelger, Ralph KnöllBiophys Rev. 2017 Jun; 9(3): 207–223. Published online 2017 May 5. doi: 10.1007/s12551-017-0265-7PMCID: PMC5498329 Maria Franaszczyk, Przemyslaw Chmielewski, Grazyna Truszkowska, Piotr Stawinski, Ewa Michalak, Malgorzata Rydzanicz.

TTPA: Adivitiya, Khasa YP. The evolution of recombinant thrombolytics: Current status and future directions. Bioengineered. 2017;8(4):331-358. doi:10.1080/21655979.2016.1229718.

TTR: Offenburger S-L, Ho XY, Tachie-Menson T, Coakley S, Hilliard MA, Gartner A. 6-OHDA-induced dopaminergic neurodegeneration in Caenorhabditis elegansis promoted by the engulfment pathway and inhibited by the transthyretin-related protein TTR-33. Caldwell K,

TUB: Von Loeffelholz O, Venables NA, Drummond DR, Katsuki M, Cross R, Moores CA. Nucleotide– and Mal3-dependent changes in fission yeast microtubules suggest a structural plasticity view of dynamics. Nature Communications. 2017;8:2110. doi:10.1038/s41467-017-0

TWIST1: Rajendra P. Gajula, Sivarajan T. Chettiar, Russell D. Williams, Katriana Nugent, Yoshinori Kato, Hailun Wang, Reem Malek, Kekoa Taparra, Jessica Cades, Anvesh Annadanam, A-Rum Yoon, Elana Fertig, Beth A. Firulli, Lucia Mazzacurati, Timothy F. Burns, Anthony B. Firulli, Steven S. An, Phuoc T. Tran Neoplasia. 2015 Jan; 17(1): 16–31. Published online 2015 Jan 23. doi: 10.1016/j.neo.2014.10.009PMCID: PMC4309734 Jing Wang, Kumar Nikhil, Keith Viccaro, Lei Chang, Max Jacobsen, George Sandusky.

TXNRD2: Kiermayer C, Northrup E, Schrewe A, et al. Heart-Specific Knockout of the Mitochondrial Thioredoxin Reductase (Txnrd2) Induces Metabolic and Contractile Dysfunction in the Aging Myocardium. Journal of the American Heart Association: Cardiovascular and Cer

TYK2: Kreins AY, Ciancanelli MJ, Okada S, et al. Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. The Journal of Experimental Medicine. 2015;212(10):1641-1662. doi:10.1084/jem.20140280.

TYR: Evaluation of the effect of narrow band-ultraviolet B on the expression of tyrosinase, TYRP-1, and TYRP-2 mRNA in vitiligo skin and their correlations with clinical improvement: A retrospective study. (PMID: 33314655) Awad SS ... Telep RAA Dermatologic therapy 2021 3 Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. (PMID: 33223529) Mor-Shaked H ... Harel T European journal of human genetics : EJHG 2021 Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. (PMID: 32966289) Rayner JE ... Sturm RA PloS one 2020 3 Association between brown eye colour in rs12913832:GG individuals and SNPs in TYR, TYRP1, and SLC24A4. (PMID: 32915910) Meyer OS ... Andersen JD PloS one 2020 3 UP256 Inhibits Hyperpigmentation by Tyrosinase Expression/Dendrite Formation via Rho-Dependent Signaling and by Primary Cilium Formation in Melanocytes. (PMID: 32731326) Kang MC ... Kim SY International journal of molecular sciences 2020 3 Mapping the TYR gene reveals novel and previously reported variants in Eastern Indian patients highlighting preponderance of the same changes in multiple unrelated ethnicities. (PMID: 32115698) Ganguly K ... Sengupta M Annals of human genetics 2020 Comprehensive analysis reveals mutational spectra and common alleles in Chinese patients with oculocutaneous albinism. (PMID: 19865097) Wei A ... Li W The Journal of investigative dermatology 2010 3 21 39 70 Birth prevalence and mutation spectrum in danish patients with autosomal recessive albinism. (PMID: 19060277) Grønskov K ... Rosenberg T Investigative ophthalmology & visual science 2009 3 21 39 70 Contribution of genetic factors for melanoma susceptibility in sporadic US melanoma patients. (PMID: 19320745) Council ML ... Bowcock AM Experimental dermatology 2009 3 21 39 70 Comprehensive analysis of oculocutaneous albinism among non-Hispanic caucasians shows that OCA1 is the most prevalent OCA type. (PMID: 18463683) Hutton SM ... Spritz RA The Journal of investigative dermatology 2008 3 21 39 70 Genetic determinants of hair, eye and skin pigmentation in Europeans. (PMID: 17952075) Sulem P ... Stefansson K Nature genetics 2007

UBAC2: Yamazoe K, Meguro A, Takeuchi M, Shibuya E, Ohno S, Mizuki N. Comprehensive analysis of the association between UBAC2 polymorphisms and Behçet's disease in a Japanese population. Scientific Reports. 2017;7:742. doi:10.1038/s41598-017-00877-3.

UBASH3A: Yan Ge, Taylor K. Paisie, Jeremy R.B. Newman, Lauren M. McIntyre, Patrick ConcannonDiabetes. 2017 Jul; 66(7): 2033–2043. Published online 2017 Jun 15. doi: 10.2337/db16-1023PMCID: PMC5482087 Lina-Marcela Diaz-Gallo, Elena Sánchez, Norberto Ortego-Centeno, Jose Mario Sabio, Francisco J. García-Hernández, Enrique de Ramón, Miguel A. González-Gay, Torsten Witte, Hans-Joachim Anders, María F. González-Escribano, Javier MartinPLoS One. 2013; 8(4): e60646. Published online 2013 Apr 2. doi: 10.137

UBE2E2: Nattachet Plengvidhya, Chutima Chanprasert, Nalinee Chongjaroen, Pa-thai Yenchitsomanus, Mayuree Homsanit, Watip TangjittipokinBMC Med Genet. 2018; 19: 93. Published online 2018 Jun 5. doi: 10.1186/s12881-018-0614-9PMCID: PMC5989367 Kuanfeng Xu, Lin Jiang, Mei Zhang, Xuqin Zheng, Yong Gu, Zhixiao Wang, Yun Cai, Hao Dai, Yun Shi, Shuai Zheng, Yang Chen, Li Ji, Xinyu Xu, Heng Chen, Min Sun, Tao YangMedicine (Baltimore) 2016 May; 95(19): e3604. Published online 2016 May 13.

UCP2: Ježek P, Holendová B, Garlid KD, Jab?rek M. Mitochondrial Uncoupling Proteins: Subtle Regulators of Cellular Redox Signaling . Antioxidants & Redox Signaling. 2018;29(7):667-714. doi:10.1089/ars.2017.7225.

UGT1A1: Ramírez J, Mirkov S, House LK, Ratain MJ. Glucuronidation of OTS167 in Humans Is Catalyzed by UDP-Glucuronosyltransferases UGT1A1, UGT1A3, UGT1A8, and UGT1A10. Drug Metabolism and Disposition. 2015;43(7):928-935. doi:10.1124/dmd.115.063271.

UGT2B7: Qiong Lu, Yuan-Tao Huang, Yi Shu, Ping Xu, Da-Xiong Xiang, Qiang Qu, Jian QuMedicine (Baltimore) 2018 Jul; 97(30): e11662. Published online 2018 Jul 27. doi: 10.1097/MD.0000000000011662PMCID: PMC6078657Zi-zhao Yang, Li Li, Lu Wang, Ling-min Yuan, Ming-cheng Xu, Jing-kai Gu, Hui-di Jiang, Lu-shan Yu, Su ZengActa Pharmacol Sin. 2017 Aug; 38(8): 1184–1194. Published online 2017 May 29. doi: 10.1038/aps.2016.157 PMCID: PMC5547550Zi-Zhao Yang, Li Li, Ming-Cheng Xu, Hai-Xing Ju, Miao Hao, Jing-Ka

UNC13A: Benjamin Gaastra, Aleksey Shatunov, Sara Pulit, Ashley R. Jones, William Sproviero, Alexandra Gillett, Zhongbo Chen, Janine Kirby, Isabella Fogh, John F. Powell, P. Nigel Leigh, Karen E. Morrison, Pamela J. Shaw, Christopher E. Shaw, Leonard H. van den Berg, Jan H. Veldink, Cathryn M. Lewis, Ammar Al-ChalabiAmyotroph Lateral Scler Frontotemporal Degener. 2016 Nov 16; 17(7-8): 593–599. Published online 2016 Sep 1. doi: 10.1080/21678421.2016.1213852PMCID: PMC5125285Noa Lipstein. UNC13A in amyotrophic lateral sclerosis: from genetic association to therapeutic target Sean W Willemse ,1 Peter Harley,2 Ruben P A van Eijk ,1,3 Koen C Demaegd,1 Pavol Zelina,4 R Jeroen Pasterkamp,4 Philip van Damme ,5,6 Caroline Ingre,7 Wouter van Rheenen,1 Jan H Veldink ,1 Matthew C Kiernan ,8,9 Ammar Al- Chalabi ,10 Leonard H van den Berg,1 Pietro Fratta,2 Michael A van Es

UNC5C: Qiangqiang Shao, Tao Yang, Huai Huang, Farrah Alarmanazi, Guofa LiuJ Neurosci. 2017 Jun 7; 37(23): 5620–5633. doi: 10.1523/JNEUROSCI.2617-16.2017PMCID: PMC5469302 Sebastian Poliak, Daniel Morales, Louis-Philippe Croteau, Dayana Krawchuk, Elena Palmesino, Susan Morton, Jean-François Cloutier, Frederic Charron, Matthew B Dalva, Susan L Ackerman, Tzu-Jen Kao, Artur KaniaeLife. 2015; 4: e10841. Published online 2015 Dec 3. doi: 10.7554/eLife.10841PMCID: PMC4764565Quan Li, Bai-Ling Wang, Fu-Rong

UROS: Sandoval JR, Lacerda DR, Jota MSA, et al. The Genetic History of Indigenous Populations of the Peruvian and Bolivian Altiplano: The Legacy of the Uros. Caramelli D, ed. PLoS ONE. 2013;8(9):e73006. doi:10.1371/journal.pone.0073006.

VCAN: Mitsui Y, Shiina H, Kato T, et al. Versican Promotes Tumor Progression, Metastasis and Predicts Poor Prognosis in Renal Carcinoma. Molecular cancer research?: MCR. 2017;15(7):884-895. doi:10.1158/1541-7786.MCR-16-0444.

VDR: Vitamin D receptor (VDR) gene polymorphisms and haplotypes, interactions with plasma 25-hydroxyvitamin D and 1,25-dihydroxyvitamin D, and prostate cancer risk. (PMID: 17440943) Mikhak B … Giovannucci E The Prostate 2007 3 21 24 39 Hereditary vitamin D resistant rickets caused by a novel mutation in the vitamin D receptor that results in decreased affinity for hormone and cellular hyporesponsiveness. (PMID: 9005998) Malloy PJ … Feldman D The Journal of clinical investigation 1997 3 4 21 69 A novel mutation in the deoxyribonucleic acid-binding domain of the vitamin D receptor causes hereditary 1,25-dihydroxyvitamin D-resistant rickets. (PMID: 8675579) Lin NU … Feldman D The Journal of clinical endocrinology and metabolism 1996 3 4 21 69 Two mutations in the hormone binding domain of the vitamin D receptor cause tissue resistance to 1,25 dihydroxyvitamin D3. (PMID: 8392085) Kristjansson K … Hughes MR The Journal of clinical investigation 1993 3 4 21 69 A unique point mutation in the human vitamin D receptor chromosomal gene confers hereditary resistance to 1,25-dihydroxyvitamin D3. (PMID: 2177843) Sone T … Pike JW Molecular endocrinology (Baltimore, Md.) 1990 3 4 21 69 Detection of VDR gene ApaI and TaqI polymorphisms in patients with type 2 diabetes mellitus using PCR-RFLP method in a Turkish population. (PMID: 19186074) Dilmec F … van Kuilenburg AB Journal of diabetes and its complications 2010 3 21 39 Rubella vaccine-induced cellular immunity: evidence of associations with polymorphisms in the Toll-like, vitamin A and D receptors, and innate immune response genes. (PMID: 19902255) Ovsyannikova IG … Poland GA Human genetics 2010 3 21 39 Pharmacogenetic risk factors for altered bone mineral density and body composition in pediatric acute lymphoblastic leukemia. (PMID: 20015871) te Winkel ML … van den Heuvel-Eibrink MM Haematologica 2010 3 21 39 Calcium, vitamin D, VDR genotypes, and epigenetic and genetic changes in rectal tumors. (PMID: 20432164) Slattery ML … Samowitz W Nutrition and cancer 2010 3 21 39 Genetic variation in the vitamin D receptor (VDR) and the vitamin D-binding protein (GC) and risk for colorectal cancer: results from the Colon Cancer Family Registry. (PMID: 20086113) Poynter JN … Haile RW Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2010 3 21 39 Vitamin d pathway genes, diet, and risk of renal cell carcinoma. (PMID: 20049159) Karami S … Moore LE International journal of endocrinology 2010 3 21 39 Maternal serum 25-hydroxyvitamin D concentrations are associated with small-for-gestational age births in white women. (PMID: 20200114) Bodnar LM … Simhan HN The Journal of nutrition 2010 3 21 39 Vitamin D receptor gene polymorphisms and distinct clinical phenotypes of hepatitis B carriers in Taiwan. (PMID: 19693091) Huang YW … Kao JH Genes and immunity 2010 3 21 39 Vitamin D receptor genetic polymorphisms and tuberculosis: updated systematic review and meta-analysis. (PMID: 20003690) Gao L … Jin Q The international journal of tuberculosis and lung disease : the official journal of the International Union against Tuberculosis and Lung Disease 2010 3 21 39 Influence of vitamin D receptor haplotypes on blood lead concentrations in environmentally exposed children of Uygur and Han populations. (PMID: 20100036) Chen Y … Tian W Biomarkers : biochemical indicators of exposure, response, and susceptibility to chemicals 2010 3 21 39 A rare haplotype of the vitamin D receptor gene is protective against diabetic nephropathy. (PMID: 19783860) Martin RJ … Warren 3/UK GoKinD Study Group Nephrology, dialysis, transplantation : official publication of the European Dialysis and Transplant Association - European Renal Association 2010 3 21 39 Polymorphisms in vitamin D metabolism related genes and risk of multiple sclerosis. (PMID: 20007432) Simon KC … Ascherio A Multiple sclerosis

(Houndmills, Basingstoke, England) 2010 3 21 39 Maternal 25-hydroxyvitamin D concentration and offspring birth size: effect modification by infant VDR genotype. (PMID: 19018272) Morley R ... Ponsonby AL European journal of clinical nutrition 2009 3 21 39 Staphylococcus aureus nasal carriage might be associated with vitamin D receptor polymorphisms in type 1 diabetes. (PMID: 19411183) Panierakis C ... Galanakis E International journal of infectious diseases : IJID : official publication of the International Society for Infectious Diseases 2009 3 21 39 Genetic and environmental factors in human osteoporosis from Sub-Saharan to Mediterranean areas. (PMID: 19255718) Musumeci M ... Musumeci S Journal of bone and mineral metabolism 2009 3 21 39 Plasma 1,25 dihydroxy vitamin D3 level and expression of vitamin d receptor and cathelicidin in pulmonary tuberculosis. (PMID: 19219539) Selvaraj P ... Alagarasu K Journal of clinical immunology 2009 3 21 39 Vitamin D binding protein genotype and osteoporosis. (PMID: 19488670) Fang Y ... Uitterlinden AG Calcified tissue international 2009 3 21 39 Analysis of SNPs and haplotypes in vitamin D pathway genes and renal cancer risk. (PMID: 19753122) Karami S ... Moore LE PloS one 2009 3 21 39 Association of a single nucleotide polymorphism in the constitutive androstane receptor gene with bone mineral density. (PMID: 19702932) Urano T ... Inoue S Geriatrics & gerontology international 2009 3 21 39 Association of oestrogen receptor gene polymorphism with the long-term results of rotational acetabular osteotomy. (PMID: 19219433) Yamanaka M ... Kurosawa H International orthopaedics 2009 3 21 39 Associations of vitamin D receptor, calcium-sensing receptor and parathyroid hormone gene polymorphisms with calcium homeostasis and peripheral bone density in adult Finns. (PMID: 19690432) Laaksonen MM ... Lamberg-Allardt CJ Journal of nutrigenetics and nutrigenomics 2009 3 21 39 Vitamin D pathway gene variants and prostate cancer risk. (PMID: 19454612) Holt SK ... Stanford JL Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 21 39 Vitamin D receptor TaqI, BsmI and ApaI polymorphisms and osteoarthritis susceptibility: a meta-analysis. (PMID: 19073371) Lee YH ... Song GG Joint bone spine 2009 3 21 39 Association between vitamin D receptor gene polymorphisms and severe chronic periodontitis in a Chinese population. (PMID: 19335080) Wang C ... Zhang J Journal of periodontology 2009 3 21 39 Vitamin D receptor polymorphisms and breast cancer risk: results from the National Cancer Institute Breast and Prostate Cancer Cohort Consortium. (PMID: 19124512) McKay JD ... Thun MJ Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 21 39 Polymorphisms of the vitamin D receptor gene and stress fractures. (PMID: 19391078) Chatzipapas C ... Stratakis CA Hormone and metabolic research = Hormon- und Stoffwechselforschung = Hormones et metabolisme 2009 3 21 39 Vitamin D receptor polymorphisms are associated with increased susceptibility to primary biliary cirrhosis in Japanese and Italian populations. (PMID: 19376604) Tanaka A ... Takikawa H Journal of hepatology 2009 3 21 39 5' regulatory and 3' untranslated region polymorphisms of vitamin D receptor gene in south Indian HIV and HIV-TB patients. (PMID: 18712587) Alagarasu K ... Narayanan PR Journal of clinical immunology 2009 3 21 39 The bone mass density in postmenopausal women using hormonal replacement therapy in relation to polymorphism in vitamin D receptor and estrogen receptor genes. (PMID: 19903038) Brodowska A ... Nawrocka-Rutkowska J Gynecological endocrinology : the official journal of the International Society of Gynecological Endocrinology 2009 3 21 39 Vitamin D receptor and calcium sensing receptor polymorphisms and the risk of colorectal cancer in European populations. (PMID: 19706842) Jenab M ... Riboli E Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology 2009 3 21 39 Genetic determinants of extracellular magnesium concentration: analysis of multiple candidate genes, and evidence for association with the estrogen receptor alpha (ESR1) locus. (PMID: 19695239) Shuen AY ... Cole DE Clinica chimica acta; international journal of clinical chemistry 2009 3 21 39 Association between VDR and ESR1 gene polymorphisms with bone and obesity phenotypes in Chinese male nuclear families. (PMID: 19960008) Gu JM ... Zhang ZL Acta pharmacologica Sinica 2009

VEGFA: Chesnokov MS, Khesina PA, Shavochkina DA, et al. Shift in VEGFA isoform balance towards more angiogenic variants is associated with tumor stage and differentiation of human hepatocellular carcinoma. Singh SR, ed. PeerJ. 2018;6:e4915. doi:10.7717/peerj.491

VN1R85P: Genetic polymorphisms associated with telomere length and risk of developing myeloproliferative neoplasms Matteo Giaccherini, Angelica Macauda, Nicola Sgherza, Juan Sainz, Federica Gemignani, Josè Manuel Sanchez Maldonado, Manuel Jurado, Francesca Tavano, Grzegorz Mazur, Andrés Jerez, Joanna Góra-Tybor, Aleksandra Gołos, Francisca Hernández Mohedo, Joaquin Martinez Lopez, Judit Várkonyi, Raffaele Spadano, Aleksandra Butrym, Federico Canzian & Daniele Campa

VPS26A: Kirsty J. McMillan, Matthew Gallon, Adam P. Jellett, Thomas Clairfeuille, Frances C. Tilley, Ian McGough, Chris M. Danson, Kate J. Heesom, Kevin A. Wilkinson, Brett M. Collins, Peter J. CullenJ Cell Biol. 2016 Aug 15; 214(4): 389–399. doi: 10.1083/jcb.201604057PMCID: PMC4987296 Matthew Gallon, Thomas Clairfeuille, Florian Steinberg, Caroline Mas, Rajesh Ghai, Richard B. Sessions, Rohan D. Teasdale, Brett M. Collins, Peter J. CullenProc Natl Acad Sci U S A. 2014 Sep 2; 111(35): E3604–E3613.

VPS33B: Binggang Xiang, Guoying Zhang, Shaojing Ye, Rui Zhang, Cai Huang, Jun Liu, Min Tao, Changgeng Ruan, Susan S. Smyth, Sidney W. Whiteheart, Zhenyu Li Circulation. Author manuscript; available in PMC 2016 Dec 15. Published in final edited form as: Circulation. 2015 Dec 15; 132(24): 2334–2344. Published online 2015 Sep 23. doi: 10.1161/CIRCULATIONAHA.115.018361PMCID: PMC4679702 Hao Gu, Chiqi Chen, Xiaoxin Hao, Conghui Wang, Xiaocui Zhang, Zhen Li, Hongfang Shao, Hongxiang Zeng, Zhuo Yu, Li Xie.

VPS9D1: Isolation and mapping of a putative b subunit of human ATP synthase (ATP-BL) from human leukocytes. (PMID: 10231027) Sugimoto J . Isobe M DNA research : an international journal for rapid publication of reports on genes and genomes 1999 2 3 4 56 A human interactome in three quantitative dimensions organized by stoichiometries and abundances. (PMID: 26496610) Hein MY . Mann M Cell 2015 3 56 Toward a confocal subcellular atlas of the human proteome. (PMID: 18029348) Barbe L . Andersson-Svahn

VSIG10: Andrew C. Nelson, Arne W. Mould, Elizabeth K. Bikoff, Elizabeth J. RobertsonSci Rep. 2017; 7: 6793. Published online 2017 Jul 28. doi: 10.1038/s41598-017-06859-9PMCID: PMC5533796 Aileen Marshall, Margus Lukk, Claudia Kutter, Susan Davies, Graeme Alexander, Duncan T. OdomPLoS One. 2013; 8(3): e59459. Published online 2013 Mar 18. doi: 10.1371/journal.pone.0059459 PMCID: PMC3601070Scott D. Cook-Sather, Jin Li, Theodora K. Goebel, Emily M. Sussman, Mohamed A. Rehman, Hakon Hakonarson Pain.

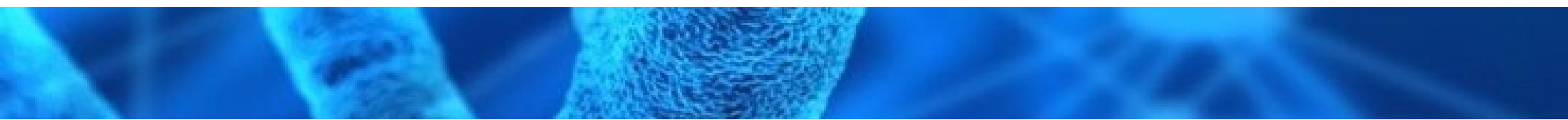
VSTM1: Wang D, Li Y, Liu Y, He Y, Shi G. Expression of VSTM1-v2 Is Increased in Peripheral Blood Mononuclear Cells from Patients with Rheumatoid Arthritis and Is Correlated with Disease Activity. Proost P, ed. PLoS ONE. 2016;11(1):e0146805. doi:10.1371/journal.p

WASF5P: Genomewide association study of an AIDS-nonprogression cohort emphasizes the role played by HLA genes (ANRS Genomewide Association Study 02). (PMID: 19115949) Limou S … ANRS Genomic Group The Journal of infectious diseases 2009 3 39 Common genetic variation and the control of HIV-1 in humans. (PMID: 20041166) Fellay J … NIAID Center for HIV/AIDS Vaccine Immunology (CHAVI) PLoS genetics 2009 3 39 A genome-wide association study of nasopharyngeal carcinoma identifies three new susceptibility loci. (PMID: 20512145) Bei JX … Zeng YX Nature genetics 2010 39 Genome-wide association study for vitiligo identifies susceptibility loci at 6q27 and the MHC. (PMID: 20526339) Quan C … Zhang XJ Nature genetics 2010 39 Genome-wide association study identifies variants in the MHC class I, IL10, and IL23R-IL12RB2 regions associated with Behçet's disease. (PMID: 20622878) Remmers EF … Gül A Nature genetics 2010 39

WDPCP: Cheng Cui, Bishwanath Chatterjee, Thomas P. Lozito, Zhen Zhang, Richard J. Francis, Hisato Yagi, Lisa M. Swanhart, Subramaniam Sanker, Deanne Francis, Qing Yu, Jovenal T. San Agustin, Chandrakala Puligilla, Tania Chatterjee, Terry Tansey, Xiaoqin Liu, Matthew W. Kelley, Elias T. Spiliotis, Adam V. Kwiatkowski, Rocky Tuan, Gregory J. Pazour, Neil A. Hukriede, Cecilia W. LoPLoS Biol. 2013 Nov; 11(11): e1001720. Published online 2013 Nov 26.

WDR11-AS1: Timothy J. Hohman, Mary Ellen I. Koran, Tricia A. Thornton-Wells, for the Alzheimer's Neuroimaging Initiative Front Aging Neurosci. 2014; 6: 183. Published online 2014 Aug 4. doi: 10.3389/fnagi.2014.00183PMCID: PMC4121544 Ho-Young Son, Yul Hwangbo, Seong-Keun Yoo, Sun-Wha Im, San Duk Yang, Soo-Jung Kwak, Min Seon Park, Soo Heon Kwak, Sun Wook Cho, Jun Sun Ryu, Jeongseon Kim, Yuh-Seog Jung, Tae Hyun Kim, Su-jin Kim, Kyu Eun Lee, Do Joon Park, Nam Han Cho, Joohon Sung, Jeong-Sun Seo, Eun Kyun

WDR12: Anne-Mari Moilanen, Jaana Rysä, Leena Kaikkonen, Teemu Karvonen, Erja Mustonen, Raisa Serpi, Zoltán Szabó, Olli Tenhunen, Zsolt Bagyura, Juha Näpänkangas, Pauli Ohukainen, Pasi Tavi, Risto Kerkelä, Margrét Leósdóttir, Björn Wahlstrand, Thomas Hedner, Olle Melander, Heikki RuskoahoPLoS One. 2015; 10(4): e0124907. Published online 2015 Apr 27. doi: 10.1371/journal.pone.0124907PMCID: PMC4411154 Yancun Yin, Ling Zhou, Renhui Zhan, Qiang Zhang, Minjing LiCancer Manag Res. 2018; 10: 3985–3993.



WFS1: Yasufumi Sakakibara, Michiko Sekiya, Naoki Fujisaki, Xiuming Quan, Koichi M. IijimaPLoS Genet. 2018 Jan; 14(1): e1007196. Published online 2018 Jan 22. doi: 10.1371/journal.pgen.1007196PMCID: PMC5794194 Triin Tekko, Triin Lakspere, Anni Allikalt, Jaanus End, Karl Rene Kõlvart, Toomas Jagomäe, Anton Terasmaa, Mari-Anne Philips, Tanel Visnapuu, Fred Väärtnõu, Scott F. Gilbert, Ago Rinken, Eero Vasar, Kersti LilleväliPLoS One. 2017; 12(3): e0172825. Published online 2017 Mar 7.

WNT10A: Ren-Jun Hsu, Jar-Yi Ho, Tai-Lung Cha, Dah-Shyong Yu, Chieh-Lin Wu, Wei-Ping Huang, Pauling Chu, Ying-Hsin Chen, Jiann-Torng Chen, Cheng-Ping YuPLoS One. 2012; 7(10): e47649. Published online 2012 Oct 19. doi: 10.1371/journal.pone.0047649PMCID: PMC3477117Qiuping Yuan, Min Zhao, Bhavna Tandon, Lorena Maili, Xiaoming Liu, Anqi Zhang, Evan H. Baugh, Tam Tran, Renato M. Silva, Jacqueline T. Hecht, Eric C. Swindell, Daniel S. Wagner, Ariadne LetraMol Genet Genomic Med. 2017 Nov; 5(6): 730–741. Pu

WSCD2: Lo M-T, Hinds DA, Tung JY, et al. Genome-wide analyses for personality traits identify six genomic loci and show correlations with psychiatric disorders. Nature genetics. 2017;49(1):152-156. doi:10.1038/ng.3736.

WWC1: Ting Lin, Li Zhang, Shuangxin Liu, Yuanhan Chen, Hong Zhang, Xingchen Zhao, Ruizhao Li, Qianmei Zhang, Ruyi Liao, Zongshun Huang, Bin Zhang, Wenjian Wang, Xinling Liang, Wei ShiMol Med Rep. 2017 Dec; 16(6): 8685–8690. Published online 2017 Oct 4. doi: 10.3892/mmr.2017.7708PMCID: PMC5779946Upal Basu-Roy, N. Sumru Bayin, Kirk Rattanakorn, Eugenia Han, Dimitris G. Placantonakis, Alka Mansukhani, Claudio BasilicoNat Commun. 2015; 6: 6411. Published online 2015 Apr 2. doi: 10.1038/ncomms7411PMCI

XBP1: McLaughlin T, Falkowski M, Park JW, et al. Loss of XBP1 accelerates age-related decline in retinal function and neurodegeneration. Molecular Neurodegeneration. 2018;13:16. doi:10.1186/s13024-018-0250-z.

XPC: Mihaela Robu, Rashmi G. Shah, Nupur K. Purohit, Pengbo Zhou, Hanspeter Naegeli, Girish M. ShahProc Natl Acad Sci U S A. 2017 Aug 15; 114(33): E6847–E6856. Published online 2017 Jul 31. doi: 10.1073/pnas.1706981114PMCID: PMC5565455B. Bidon, I. Iltis, M. Semer, Z. Nagy, A. Larnicol, A. Cribier, M. Benkirane, F. Coin, J-M. Egly, N. Le MayNat Commun. 2018; 9: 2610. Published online 2018 Jul 4. doi: 10.1038/s41467-018-05010-0PMCID: PMC6031651 Jing Yan Krzeszinski, Vitnary Choe, Jia Shao, Xin Bao

XRCC1: Cannan WJ, Rashid I, Tomkinson AE, Wallace SS, Pederson DS. The Human Ligase III?-XRCC1 Protein Complex Performs DNA Nick Repair after Transient Unwrapping of Nucleosomal DNA. The Journal of Biological Chemistry. 2017;292(13):5227-5238. doi:10.1074/jbc.M1

ZBTB10: Liu X, Jutooru I, Lei P, et al. Betulinic Acid Targets YY1 and ErbB2 through Cannabinoid Receptor-Dependent Disruption of MicroRNA-27a:ZBTB10 in Breast Cancer. Molecular cancer therapeutics. 2012;11(7):1421-1431. doi:10.1158/1535-7163.MCT-12-0026.

ZBTB46: Yu Wang, He-Ying Sun, Sandeep Kumar, Maria del Mar Puerta, Hanjoong Jo, Amir RezvanLab Invest. 2019 Mar; 99(3): 305–318. Published online 2018 Jun 8. doi: 10.1038/s41374-018-0060-5PMCID: PMC6286701Miche Rombouts, Nathalie Cools, Mandy O. J. Grootaert, Flore de Bakker, Ilse Van Brussel, An Wouters, Guido R. Y. De Meyer, Benedicte Y. De Winter, Dorien M. SchrijversPLoS One. 2017; 12(1): e0169608. Published online 2017 Jan 6. doi: 10.1371/journal.pone.0169608PMCID: PMC5218565Ansuman T. Satpath

ZFHX3: Dan Zhao, Gui Ma, Xiaolin Zhang, Yuan He, Mei Li, Xueying Han, Liya Fu, Xue-Yuan Dong, Tamas Nagy, Qiang Zhao, Li Fu, Jin-Tang DongJ Biol Chem. 2016 Jun 10; 291(24): 12809–12820. Published online 2016 Apr 20. doi: 10.1074/jbc.M116.719377PMCID: PMC4933452 Yufeng Huang, Chuchu Wang, Yufeng Yao, Xiaoyu Zuo, Shanshan Chen, Chengqi Xu, Hongfu Zhang, Qiulun Lu, Le Chang, Fan Wang, Pengxia Wang, Rongfeng Zhang, Zhenkun Hu, Qixue Song, Xiaowei Yang, Cong Li, Sisi Li, Yuanyuan Zhao, Qin Yang.

ZNF276: The protein composition of mitotic chromosomes determined using multiclassifier combinatorial proteomics. (PMID: 20813266) Ohta S . Rappsilber J Cell 2010 2 3 4 56 Complete sequencing and characterization of 21,243 full-length human cDNAs. (PMID: 14702039) Ota T . Sugano S Nature genetics 2004 3 4 56 The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). (PMID: 15489334) Gerhard DS . MGC Project Team Genome research 2004 3 4 56 Cloning a

ZNF365: Zhang Y, Shin SJ, Liu D, et al. ZNF365 promotes stability of fragile sites and telomeres. Cancer discovery. 2013;3(7):798-811. doi:10.1158/2159-8290.CD-12-0536.

ZNF536: Zhen Qin, Fangli Ren, Xialian Xu, Yongming Ren, Hongge Li, Yinyin Wang, Yonggong Zhai, Zhijie ChangMol Cell Biol. 2009 Jul; 29(13): 3633–3643. Published online 2009 Apr 27. doi: 10.1128/MCB.00362-09 PMCID: PMC2698762Eugene Lin, Po-Hsiu Kuo, Yu-Li Liu, Younger W.-Y. Yu, Albert C. Yang, Shih-Jen TsaiFront Psychiatry. 2018; 9: 290. Published online 2018 Jul 6. doi: 10.3389/fpsyt.2018.00290 PMCID: PMC6043864 Stacey J. Winham, Alfredo B. Cuellar-Barboza, Susan L. McElroy, Alfredo Oliveros.

ZNF648: Biological, clinical and population relevance of 95 loci for blood lipids. (PMID: 20686565) Teslovich TM ... Kathiresan S Nature 2010 3 39 The DNA sequence and biological annotation of human chromosome 1. (PMID: 16710414) Gregory SG ... Prigmore E Nature 2006 3 4 Characterization and evolutionary origin of novel C2H2 zinc finger protein (ZNF648) required for both erythroid and megakaryocyte differentiation in humans. (PMID: 33054117) Ferguson DCJ ... Frayne J Haematologica 2021 3 A reference map of the human binary protein interactome. (PMID: 32296183) Luck K ... Calderwood MA Nature 2020 3 UFMylation maintains tumour suppressor p53 stability by antagonizing its ubiquitination. (PMID: 32807901) Liu J ... Cong YS Nature cell biology 2020 Characterization and evolutionary origin of novel C2H2 zinc finger protein (ZNF648) required for both erythroid and megakaryocyte differentiation in humans. (PMID: 33054117) Ferguson DCJ ... Frayne J Haematologica 2021 The SNP rs10911021 is associated with oxidative stress in coronary heart disease patients from Pakistan Saleem Ullah Shahid, Shabana & Steve Humphries Lipids in Health and Disease volume 17, Article number: 6 (2018)

ZNF767P: Binghai Chen, Chengyue Wang, Jin Zhang, Yang Zhou, Wei Hu, Tao GuoCancer Cell Int. 2018; 18: 157. Published online 2018 Oct 11. doi: 10.1186/s12935-018-0652-6 PMCID: PMC6180637Pierre-Benoit Ancey, Barbara Testoni, Marion Gruffaz, Marie-Pierre Cros, Geoffroy Durand, Florence Le Calvez-Kelm, David Durantel, Zdenko Herceg, Hector Hernandez-VargasOncotarget. 2015 Dec 29; 6(42): 44877–44891. Published online 2015 Nov 2. doi: 10.18632/oncotarget.6270PMCID: PMC4792598Yue Teng, Yuzhuo Wang.

ZNF827: Liu Z, Li F, Zhang B, Li S, Wu J, Shi Y. Structural Basis of Plant Homeodomain Finger 6 (PHF6) Recognition by the Retinoblastoma Binding Protein 4 (RBBP4) Component of the Nucleosome Remodeling and Deacetylase (NuRD) Complex. The Journal of Biological Che

ZPR1: Genabai NK, Kannan A, Ahmad S, Jiang X, Bhatia K, Gangwani L. Deregulation of ZPR1 causes respiratory failure in spinal muscular atrophy. Scientific Reports. 2017;7:8295. doi:10.1038/s41598-017-07603-z.