PRECISION HEALTH

PRECISYA

PANEL FERTILITY

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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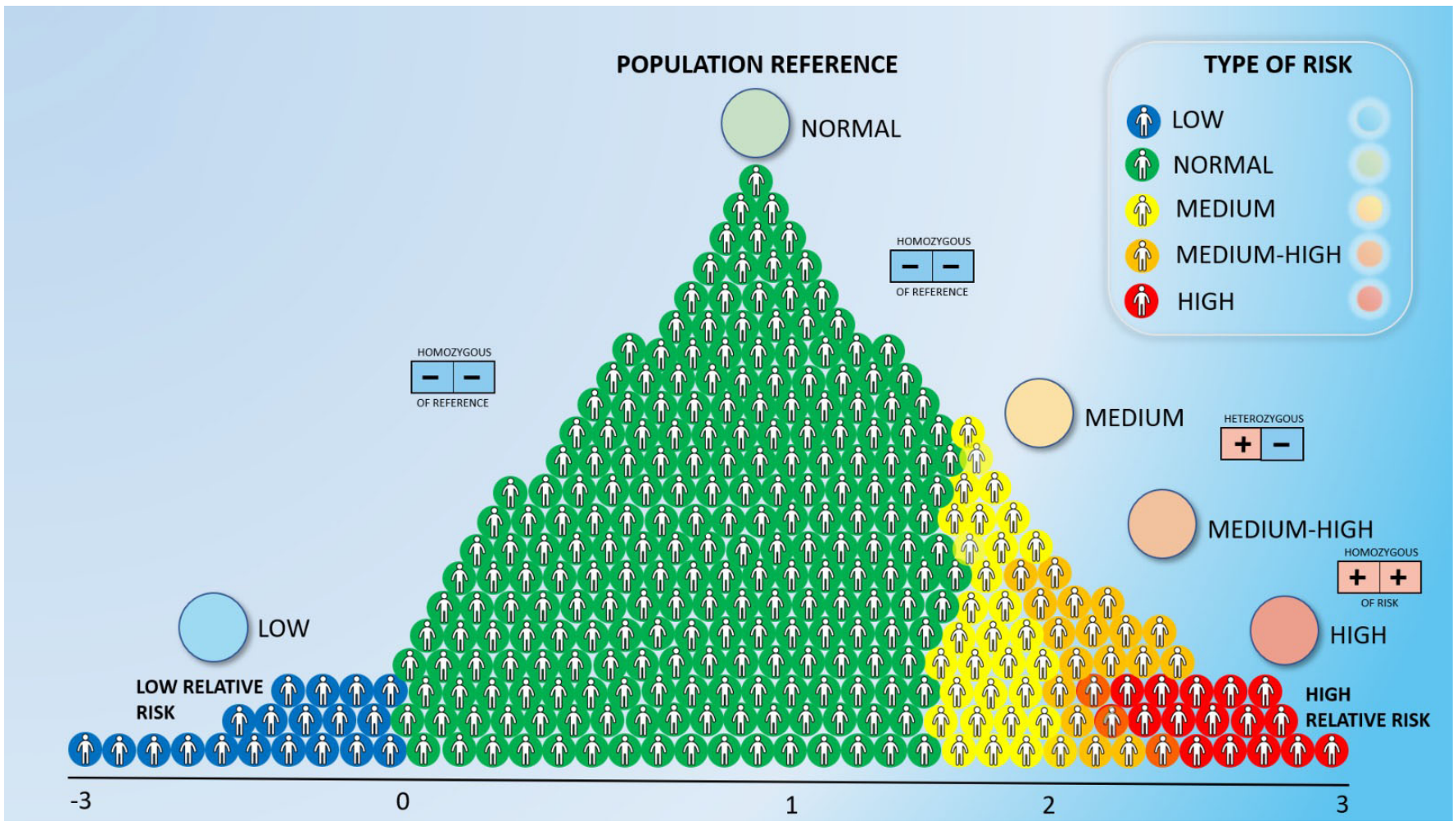
How to interpret the exam:

FIRST PART

The analyzed genetic CONDITIONS are grouped into CATEGORIES.

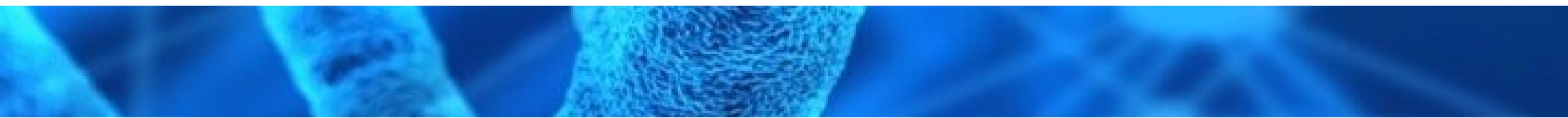
Each CONDITION is presented according to its MAGNITUDE. That is, what is the genetic susceptibility (intensity or possibility) of the analyzed condition to express itself (happen).

- 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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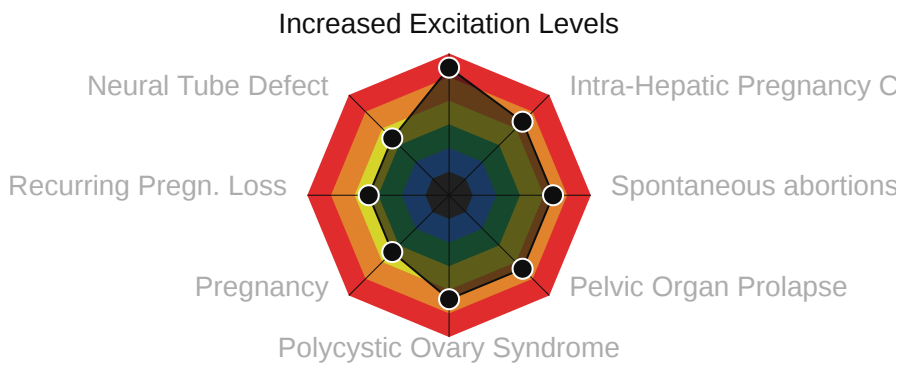
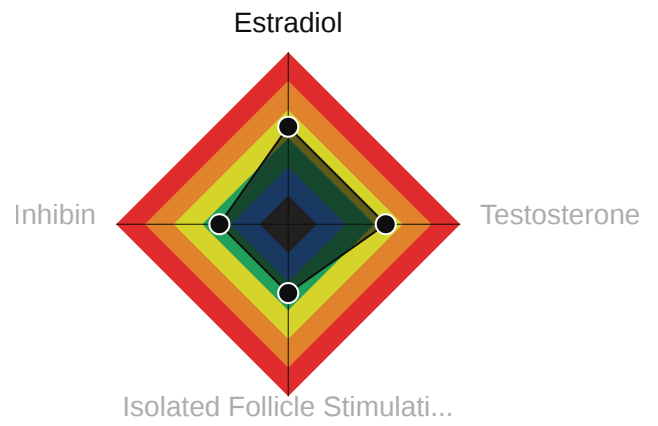
MOST RELEVANT CONDITIONS BY CATEGORY

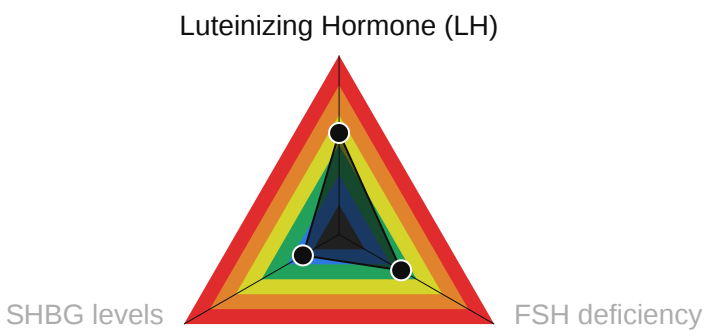
REPRODUCTIVE SYSTEM



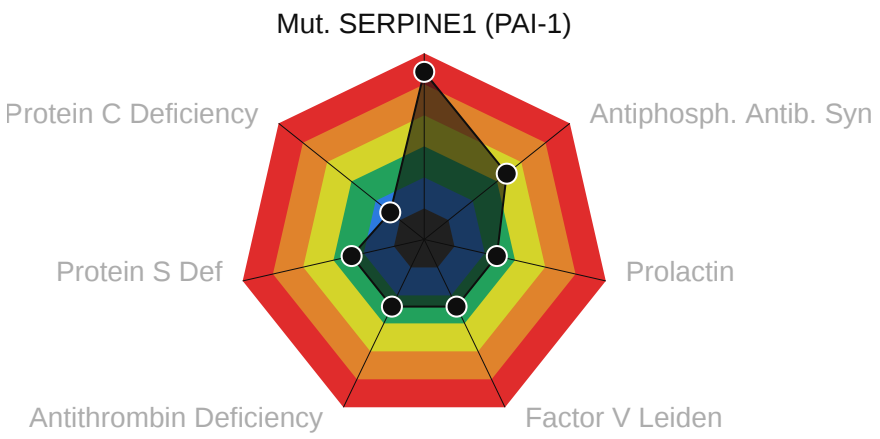
HORMONES



HORMONE



IMMUNE SYSTEM



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SUMMARY OF RESULTS

Breast cancer

Ductal Carcinoma (Breast)	1	-	-	1	+	-	0	+	+	NORMAL

Cancer

Ovary Neoplasm	40	-	-	4	+	-	0	+	+	MEDIUM

Cardiovascular

Prothrombin (G20210A Mutation)	1	-	-	0	+	-	0	+	+	NORMAL

Digestive system

Familial Intrahepatic Cholestasis	23	-	-	0	+	-	1	+	+	MEDIUM-HIGH
Constipation	3	-	-	1	+	-	0	+	+	MEDIUM

Drug Reactions

Reactions with the use of antidepressants (SSRI)	0	-	-	1	+	-	0	+	+	NORMAL

Endocrine system

Thyrotoxicosis	6	-	-	1	+	-	1	+	+	MEDIUM
Hyperthyroidism	4	-	-	0	+	-	0	+	+	LOW

General

Longer Menstrual Cycle Duration	3	-	-	0	+	-	1	+	+	MEDIUM-HIGH
Menstrual Migraine	2	-	-	0	+	-	0	+	+	NORMAL

Hematologic system

Congenital afibrinogenemia	19	-	-	0	+	-	0	+	+	NORMAL

Hereditary diseases

Fragile X Syndrome	16	-	-	0	+	-	0	+	+	NORMAL

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Hormone

Luteinizing Hormone (LH)	3 - - 0 + - 1 + +	MEDIUM
FSH deficiency	3 - - 1 + - 0 + +	NORMAL
SHBG levels	1 - - 1 + - 0 + +	LOW

Hormones

Estradiol	1 - - 0 + - 1 + +	MEDIUM
Testosterone	6 - - 3 + - 1 + +	MEDIUM
Isolated Follicle Stimulating Hormone Deficiency (FSH)	3 - - 1 + - 0 + +	NORMAL
Inhibin	1 - - 0 + - 0 + +	NORMAL

Immune system

Mutation of the SERPINE1 Gene (PAI-1)	2 - - 0 + - 1 + +	HIGH
Antiphospholipid Antibody Syndrome	19 - - 5 + - 1 + +	MEDIUM
Prolactin Promoter Polymorphism	0 - - 1 + - 0 + +	NORMAL
Factor V Leiden Mutation	2 - - 1 + - 0 + +	NORMAL
Antithrombin Deficiency	4 - - 0 + - 0 + +	NORMAL
Protein S Deficiency	12 - - 0 + - 0 + +	NORMAL
Protein C Deficiency	25 - - 1 + - 0 + +	LOW

Metabolic

Ceruloplasmin	2 - - 0 + - 0 + +	NORMAL

Methylation

MTHFR 1298 mutation (rs1801131)	0 - - 1 + - 0 + +	MEDIUM-HIGH
MTHFR 677 mutation (rs1801133)	1 - - 0 + - 0 + +	NORMAL

Psychiatric

Postpartum depression	0 - - 0 + - 1 + +	MEDIUM-HIGH

Reproductive system

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Increased Excitation Levels	0	-	-	1	+	-	1	+	+	HIGH
Intra-Hepatic Pregnancy Cholestasis	22	-	-	1	+	-	2	+	+	MEDIUM-HIGH
Spontaneous abortions	0	-	-	0	+	-	1	+	+	MEDIUM-HIGH
Pelvic Organ Prolapse	1	-	-	1	+	-	0	+	+	MEDIUM-HIGH
Polycystic Ovary Syndrome	12	-	-	1	+	-	1	+	+	MEDIUM-HIGH
Pregnancy	12	-	-	2	+	-	0	+	+	MEDIUM
Risk of Recurring Pregnancy Loss	3	-	-	2	+	-	0	+	+	MEDIUM
Neural Tube Defect	7	-	-	1	+	-	0	+	+	MEDIUM
Endometriosis	18	-	-	4	+	-	0	+	+	NORMAL
Uterine Fibroids	2	-	-	2	+	-	0	+	+	NORMAL
Uterine Fibromyoma	2	-	-	2	+	-	0	+	+	NORMAL
Gestational diabetes	4	-	-	0	+	-	0	+	+	NORMAL
Ovarian Hyperstimulation Syndrome	2	-	-	0	+	-	0	+	+	NORMAL
In vitro fertilization	3	-	-	0	+	-	0	+	+	NORMAL
Premature Ovarian Insufficiency	0	-	-	1	+	-	0	+	+	NORMAL
Sexual Motivation (Female)	3	-	-	0	+	-	0	+	+	NORMAL
Female Infertility	2	-	-	1	+	-	0	+	+	NORMAL

Skeletal system (bones)

Development Defects	1	-	-	0	+	-	0	+	+	NORMAL

Vitamins need

Riboflavin Deficiency	2	-	-	0	+	-	0	+	+	NORMAL

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Breast cancer

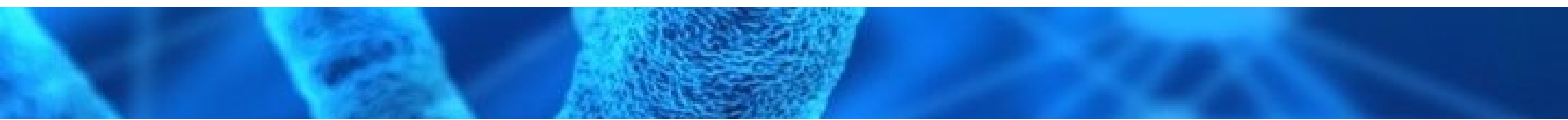
Ductal Carcinoma (Breast)

NORMAL

Premalignant or non-invasive cancerous lesion of the breast. Ductal carcinoma of the breast is a tumor derived from the cells lining the breast ducts and accounts for 80 to 90% of breast cancers. Ductal carcinoma can be divided into: In situ or intraductal: when there is proliferation of malignant cells within a duct, not exceeding the limits of the basement membrane, not invading deep structures. Invasive: when malignant cells invade structures beyond the basement membrane.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BRCA2	rs144848	GT-	C	+ -	
BRCA2	rs169547	GG-	C	- -	

Cancer



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Ovary Neoplasm

MEDIUM

Ovarian cancer is the most lethal gynecological neoplasm and the overall survival is less than 40% in five years. This is mainly because most patients have advanced stages at the time of diagnosis. In these cases, the therapeutic options - cytoreduction and chemotherapy - are only partially effective. When diagnosed early, on the other hand, the five-year survival is greater than 90% and surgery is usually the only treatment needed. However, due to the low prevalence of ovarian cancer in the population, even very specific tests produce high rates of false-positive results and increased surgical interventions to address asymptomatic adnexal masses. Based on these facts, it is essential to search for methods and strategies to detect these tumors in their early stages and, at the same time, avoid unnecessary interventions.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BRCA1	rs16942	AG-	A,C	+ -	
BRCA1	rs1799950	AA-	C	- -	
BRCA1	rs1800709	CC-	A	- -	
BRCA1	rs2227945	AA-	C,G	- -	
BRCA1	rs4986852	GG-	G,T	- -	
BRCA2	rs144848	GT-	C	+ -	
BRCA2	rs169547	GG-	C	- -	
BRCA2	rs766173	TT-	C,G	- -	
BRCA2	rs1799944	AA+	G	- -	
BRCA2	rs1799954	CC+	A,T	- -	
BRCA2	rs1801426	AA+	G	- -	
BRCA2	rs4942486	CT+	C	+ -	
BRCA2	rs4987047	AA+	T	- -	
BRCA2	rs4987117	CC+	T	- -	
BRCA2	rs28897743	GG+	A,C,T	- -	
BRCA2	rs80358807	CC+	T	- -	
BRIP1	rs4988345	CC-	A	- -	
CHMP4C	rs11782652	AA+	G	- -	
CYP24A1	rs2296241	GG+	A	- -	
CYP3A4	rs2740574	AA-	T	- -	
ERCC2	rs13181	TT+	A,G	- -	
ESR1	rs2295190	GG+		- -	
HNF1B	rs757210	AG-	G,T	- -	
INTERGENIC	rs10088218	GG+	A	- -	
PGR	rs1042838	GG-	A,G	- -	
PON1	rs662	AG-	C	+ -	

TIPARP	rs2665390	TT+	T	- -	●
TP53	rs2287498	AG-	T	- -	●
XRCC2	rs3218536	GG-	G,T	- -	●

Cardiovascular

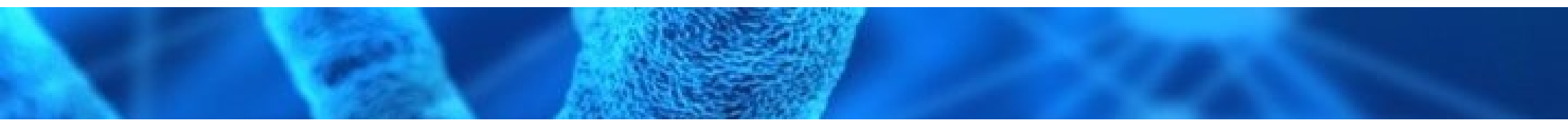
Prothrombin (G20210A Mutation)

● NORMAL

The G20210A mutation in the prothrombin gene results in increased thrombin formation and consequent exacerbated coagulation, with an increased risk for venous thrombosis, approximately 3-fold compared to the general population. This polymorphism also predisposes to pulmonary embolism and cerebral venous thrombosis, and some authors also suggest a risk of arterial thrombosis, showing that women with two of these mutations and who use oral contraceptives are up to 149 times more likely to suffer a stroke (cerebrovascular accident). However, in 99% of cases, the individual does not know that he/she has this genetic predisposition. The evaluation of these genes is also indicated for couples with repeated pregnancy losses, as they are involved in events of placental abruption, pre-eclampsia, intrauterine growth restriction and postpartum thrombosis. Therefore, the isolated or combined presence of these polymorphisms should be seen as a predisposing factor to thrombophilia and should guide the individual with appropriate prevention measures and clinical treatment. It is also recommended for this patient and their families a genetic-clinical counseling to discuss the direct and indirect implications of this result.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
F2	rs1799963	GG+	A	- -	●

Digestive system



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Familial Intrahepatic Cholestasis

MEDIUM-HIGH

Progressive familial intrahepatic cholestasis (PFIC) is a disorder that causes progressive liver disease, which typically leads to liver failure. In people with PFIC, liver cells are less able to secrete bile, and the buildup of bile in liver cells causes liver disease. PFIC is estimated to affect 1 in 50,000 to 100,000 people worldwide.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCB11	rs2287616	TT-	G	- -	
ABCB11	rs2287622	GT-	C,G,T	- -	
ABCB11	rs11568372	AA-	C	- -	
ABCB11	rs147649016	CC+	T	- -	
ABCB4	rs1202283	AA+	A	- -	
ABCB4	rs45575636	GG-	T	- -	
ABCB4	rs72552778	CC-	A	- -	
ABCB4	rs794727183	GG-	T	- -	
ABCB4	rs863225298	CC-	C	- -	
ABCC2	rs8187710	GG+	A	- -	
ABCC2	rs17222723	TT+	A	- -	
ATP8B1	rs121909100	TT-	G	- -	
NR1H4	rs113090017	CC+	A,G,T	- -	
OPRM1	rs1799971	GG+	G	+ +	
SLC25A13	rs80338722	GG-	T	- -	

Constipation

MEDIUM

The trapped intestine or constipation, consists of difficulties to evacuate, incomplete evacuation or with petrified feces, which in most cases is caused by poor diet. People who suffer from this condition can go for days and even weeks without going to the bathroom.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COMT	rs2020917	CT+	T	+ -	
RET	rs74799832	TT+	C	- -	

Drug Reactions

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Reactions with the use of antidepressants (SSRI)

NORMAL

Selective Serotonin Reuptake Inhibitors (SSRI or SSRI) are a class of drugs used to treat depressive syndromes, anxiety disorders and some types of personality disorders. Some examples of commonly used SSRIs include: Fluoxetine (prozac), Sertraline, Fluvoxamine, Paroxetine, Citalopram, Escitalopram. The rs6311 polymorphism may indicate an increased risk of sexual dysfunction with the use of SSRI antidepressants.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
HTR2A	rs6311	CT+	C	+ -	

Congenital Heart Defect due to Maternal Periconception - SSRIs

UNDEFINED

Selective Serotonin Reuptake Inhibitors (SSRI or SSRI) are a class of drugs used to treat depressive syndromes, anxiety disorders and some types of personality disorders. SSRIs increase the extracellular concentration of the neurotransmitter serotonin in the body and brain, the most important effect on the brain. The degree of selectivity for other monoamine transporters (such as dopamine or noradrenaline) is variable, although the affinity is generally negligible; therefore, they do not increase the concentration of other neurotransmitters directly. Research indicates an association between maternal periconceptional use of selective serotonin reuptake inhibitors (SSRIs) and increased risk of congenital heart defects in infants. Women who reported taking SSRIs periconceptionally, the GG and AG genotypes (BHMT rs492842) were associated with twice the risk of congenital heart defects. The GG and AG genotypes (SHMT1 rs9909104) presented an increased risk and also for the CC and AC genotypes (MGST1 rs2075237).

Endocrine system

Thyrotoxicosis

MEDIUM

Thyrotoxicosis means an excess of thyroid hormone in the body. Having this condition also means that you have a low level of thyroid-stimulating hormone, TSH, in your bloodstream, because the pituitary feels that you have “enough” thyroid hormone. If you are thyrotoxic, you may feel nervous or irritable because all your body functions are speeding up. Hyperthyroidism, also known as an overactive thyroid, is the most common cause of thyrotoxicosis and occurs when the thyroid gland overproduces thyroid hormone.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CTLA4	rs231775	AG+	G	+ -	
MTNR1B	rs1387153	CC+	C,T	+ +	

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Hyperthyroidism

LOW

Hyperthyroidism (overactive thyroid) occurs when your thyroid gland produces too much of the hormone thyroxine. Hyperthyroidism can accelerate your body's metabolism, causing unintentional weight loss and a rapid or irregular heartbeat.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MICOS10	rs12138950	AA+	A	- -	
TNF	rs1800629	GG+	A	- -	

Transient Neonatal Diabetes

UNDEFINED

Neonatal diabetes (ND) is a rare condition characterized by hyperglycemia.

General

Longer Menstrual Cycle Duration

MEDIUM-HIGH

The menstrual cycle lasts an average of 28 days and corresponds to the time interval between the first day of menstruation and the last day before the next menstruation. Genetics influence the variability of this cycle. Results in orange or red indicate a trend towards longer menstrual cycle duration.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABO	rs657152	GT-	T	- -	
INTERGENIC	rs495828	GG+	G	- -	
INTERGENIC	rs6427782	AA+	A	+ +	

Menstrual Migraine

NORMAL

Menstrual migraine is a headache that occurs during the menstrual period, usually between 2 days before and 3 days after the start of menstruation. nausea, vomiting and sensitivity to light. The main factor that contributes to its emergence is the reduction of estrogen levels during the menstrual period, which is also responsible for PMS symptoms, such as irritability and cramps.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
SYNE1	rs9371601	GG+	T	- -	
TNF	rs3093664	AA+	G	- -	

Hematologic system

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Congenital afibrinogenemia

NORMAL

Congenital afibrinogenemia is a bleeding disorder caused by impairment of the blood clotting process. Blood clots normally protect the body after an injury, sealing off damaged blood vessels and preventing further blood loss. However, bleeding is not controlled in people with congenital afibrinogenemia. Newborns with this condition often experience prolonged bleeding from the umbilical cord stump after birth. Nosebleeds (epistaxis) and bleeding from the gums or tongue are common and can occur after minor trauma or in the absence of injury (spontaneous bleeding). Some affected individuals experience bleeding from the spaces between joints (haemarthrosis) or muscles (haematoma). Rarely, bleeding occurs in the brain or other internal organs, which can be fatal. Women with congenital afibrinogenemia may experience unusually heavy menstrual bleeding (menorrhagia). Without proper treatment, women with this disorder can have difficulty getting pregnant at term, resulting in repeated miscarriages.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FGA	rs6050	AA-	T	- -	
FGA	rs121909607	GG-	A	- -	

Hereditary diseases

Fragile X Syndrome

NORMAL

Fragile X Syndrome, Escalante Syndrome or Martin & Bell Syndrome is the 2nd most common inherited cause of mental retardation, and it is also the most common known cause of autism. It is estimated to affect 1 in 4000 men and 1 in 6000 women, with 1 in 150 women carrying the FMR1 gene.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
AFF2	rs1265404	GG+	A	- -	
FMR1	rs25704	CC+	T	- -	
FMR1	rs1805421	AA+	G	- -	

Hormone

Luteinizing Hormone (LH)

MEDIUM

Luteinizing hormone (LH, also known as lutropin and sometimes lutrophin is a hormone produced by gonadotropic cells in the anterior pituitary gland. The production of LH is regulated by gonadotropin-releasing hormone (GnRH) from the hypothalamus. In females, an acute rise of LH ("LH surge") triggers ovulation and development of the corpus luteum. In males, where LH had also been called interstitial cell–stimulating hormone (ICSH), it stimulates Leydig cell production of testosterone. It acts synergistically with follicle-stimulating hormone (FSH).

Gene	RSID	Genotype	Minor Allele	Alteration	Result
LHCGR	rs2293275	GG-	C	- -	
VDR	rs2228570	CC-	C,T	+ +	

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FSH deficiency

NORMAL

FSH, known as follicle-stimulating hormone, is produced by the pituitary and has the function of regulating sperm production and egg maturation during childbearing years. Thus, FSH is a hormone linked to fertility and its concentration in the blood helps to identify whether the testes and ovaries are working properly. The reference values of the FSH test vary according to the person's age and gender and, in the case of women, according to the phase of the menstrual cycle, and can also be useful for confirming menopause. Orange or red result indicates lower FSH.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FSHB	rs10835638	GG+	T	- -	
FSHR	rs6166	AG-	T	+ -	

SHBG levels

LOW

Sex hormone-binding globulin (SHBG) helps control the amount of sex hormones available to the body — this helps keep sex hormone-related processes in balance. Higher levels of SHBG are related to lower levels of available testosterone, which can cause fatigue, less gain in lean mass, cellulite, etc. SHBG may be related to insulin function, affecting conditions such as metabolic syndrome and polycystic ovary syndrome (PCOS). SHBG is important even after menopause — those with high levels of SHBG are at lower risk of developing postmenopausal breast cancer.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
SHBG	rs727428	AG-	T	+ -	
SHBG	rs1799941	GG+	A	- -	

Anti-Müllerian hormone (AMH)

UNDEFINED

Anti-Müllerian hormone (AMH) is a marker of ovarian reserve used in assisted reproduction techniques to predict inadequate response to controlled ovarian stimulation. It can also be useful in predicting hyperresponses in patients with polycystic ovary syndrome and collaborate to individualize stimulation protocols that are most suitable for the profile of each patient and for the final success of the treatment, which is often expensive. In addition to AMH, there are other markers of ovarian reserve, such as antral follicle count (AFC) and follicle stimulating hormone (FSH), along with Estradiol (E2) and Inhibin B.

Hormones

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Estradiol

MEDIUM

Estradiol is the most important estrogen for a woman. It is a hormone produced by the ovaries that acts on reproductive function, skin, blood vessels, bones and brain. According to studies, estradiol plays more than 300 functions in the female body. In reproduction, estradiol stimulates the release of eggs from the ovarian follicles. It also acts on the fallopian tubes, stimulating muscle contractions that take the fertilized egg to the uterus. Also in the reproductive function, estradiol promotes the reaction of the uterus to the hormone progesterone, whose function is to prepare the organ for the arrival of the fertilized egg, producing a thicker endometrium. Another important function of estradiol is to drive the development of secondary sexual characteristics, such as breast growth and body changes, affecting bones, joints and fat distribution. Estradiol is also responsible for maintaining skin elasticity, blood vessel dilation and bone health. In the brain, estradiol plays a significant role in protecting brain functions such as memory, mood and mental well-being. Estradiol levels change during a woman's menstrual cycle. It starts to increase in the middle of the follicular phase (when some ovarian follicles are stimulated) and peaks in the middle of the cycle. Until it starts to fall, reaching a second peak in the luteal phase (a phase in which the corpus luteum, the structure that remains in the ovary after the egg is released, produces progesterone). Orange or red result indicates higher estradiol. Results in red indicates higher production of estradiol- and could be beneficial.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ESR2	rs1256031	CC-	A,T	+ +	
SHBG	rs1799941	GG+	A	- -	

Testosterone

MEDIUM

Testosterone is the main male sex hormone and an anabolic steroid. In humans and male animals, testosterone plays a key role in the development of male reproductive tissues such as the testes and prostate, as well as the promotion of secondary sexual characteristics such as increased muscle mass, bone growth and maturation, and growth of body hair. In addition, testosterone is involved in health, well-being and the prevention of osteoporosis. Insufficient testosterone levels in men can lead to abnormalities, including frailty and bone loss. Its decrease can lead to fatigue, memory loss, hair loss, muscular dystrophy, irritability, depression and obesity, in addition to increasing the susceptibility to diabetes, osteoporosis and cardiovascular disease.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CYP17A1	rs6162	GG+	A	+ +	
CYP19A1	rs700518	AA-	C	- -	
FAM9B	rs5934505	CT+	G	+ -	
FSHR	rs6166	AG-	T	+ -	
HSD17B3	rs9409407	GG+	T	- -	
SHBG	rs6258	CC+	T	- -	
SHBG	rs727428	AG-	T	+ -	
SHBG	rs12150660	GG+	T	- -	

Name	Sample				
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Sample

Isolated Follicle Stimulating Hormone Deficiency (FSH) NORMAL

FSH, known as follicle-stimulating hormone, is produced by the pituitary gland and has the function of regulating sperm production and egg maturation during childbearing age. Thus, FSH is a hormone linked to fertility and its concentration in the blood helps to identify whether the testicles and ovaries are functioning properly. The reference values of the FSH test vary according to the person's age and gender and, in the case of women, with the phase of the menstrual cycle, and can also be useful to confirm menopause.

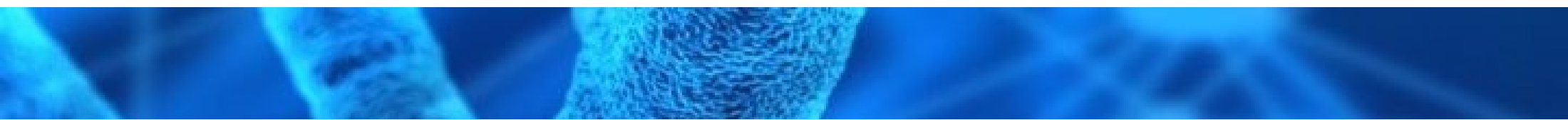
Gene	RSID	Genotype	Minor Allele	Alteration	Result
FSHB	rs10835638	GG+	T	- -	
FSHR	rs6166	AG-	T	+ -	

Inhibin NORMAL

Inhibin is a protein (hormone) produced by the testes in men and by the ovarian follicles in women, whose main function is to inhibit the production of Follicle Stimulating Hormone (FSH) by the pituitary gland. It is an antagonist (has the opposite effect) of activin. There are two types: Inhibin A and Inhibin B. Because excess testosterone and estrogen increase the risk of tumors, inhibin is important for preventing tumors. A lack of inhibin serves as a diagnostic sign of ovarian cancer.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
INHA	rs2059693	CC+	T	- -	

Immune system



Name Sample

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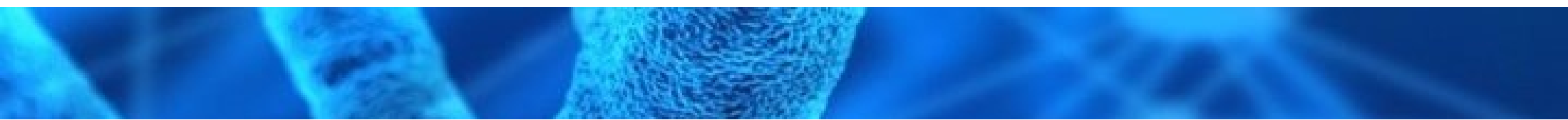
Sample

Mutation of the SERPINE1 Gene (PAI-1)

HIGH

Defects in the SERPINE1 gene are the cause of plasminogen activator inhibitor-1 deficiency (PAI-1 deficiency). PAI -1 (Plasminogen Activator Inhibitor Type 1) is a serpin protease that contributes to the control of blood clotting. This substance is secreted in different tissues such as: vascular endothelium, liver and in large quantities by adipose tissue, especially visceral adipose tissue. A high activity and concentration of PAI - 1 reduces fibrinolytic activity, which is associated with an increased risk of cardiovascular disease. Also considered a pro-inflammatory cytokine, including association with TNF-alpha and IL-6, PAI-1 is associated with clinical and functional variables, for example, showing an inverse correlation with VO2max (maximum capacity that the body has in capture oxygen from the air, transport and use it in muscles), and positive correlation with insulin resistance (pre-diabetes), cholesterol and triglyceride levels. Most importantly, a balance between t-PA (Tissue Plasminogen Activator) and PAI-1 is necessary for the blood clotting process in order to achieve adequate clotting levels. An imbalance between these or other hemostatic factors facilitates the development of atherosclerosis and thrombus formation. Some medications such as statins (control of cholesterol) and metformin (control of blood glucose) seem to contribute to the adjustment of PAI-1 concentrations. Pregnancy is another state of interest for the complex metabolic and enzymatic interaction present in the coagulation process. In this scenario, PAI-1 is associated with repeat abortions. This occurs especially when related to individuals with 4G/4G allele polymorphism, which seems to respond in up to 47% increase in PAI-1 when compared to other alleles (4G/5G and 5G/5G). Briefly, by reducing fibrinolysis, the action of PAI-1 can cause thrombosis and induce placental insufficiency, preventing fetal development

Gene	RSID	Genotype	Minor Allele	Alteration	Result
SERPINE1	rs1799889	AA+	G	- -	
SERPINE1	rs2227631	AA+	A	+ +	



Name Sample

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Sample

Antiphospholipid Antibody Syndrome

MEDIUM

Antiphospholipid antibody syndrome or anti-phospholipid antibody syndrome (APS) or Hughes syndrome is a chronic disease in which the body starts to produce antibodies that affect blood clotting, leading to the formation of clots that end up obstructing the passage of blood. blood in the veins and arteries. It is an important cause of acquired thrombophilia and repeated miscarriages; acquired because, despite not knowing the cause, there is no known genetic marker, different from congenital thrombophilias.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ATXN2	rs10774625	GG+	A	- -	
BANK1	rs10516487	CC-	A	- -	
EVA1A	rs17011455	TT+	C	- -	
INTERGENIC	rs6889746	AA+	G	- -	
INTERGENIC	rs12204683	CT+	T	+ -	
INTERGENIC	rs13403289	AC+	T	+ -	
MACROD2	rs6080100	CC+	C	+ +	
MICAL3	rs1978968	CC+	T	- -	
STAT4	rs3821236	AG+	A	+ -	
STAT4	rs10181656	CG+	C	+ -	
TLR7	rs3853839	CG+	G	+ -	

Prolactin Promoter Polymorphism

NORMAL

Prolactin (PRL) is a protein hormone that is primarily synthesized by the anterior pituitary gland. However, PRL can also be synthesized and secreted by extrapituitary tissues, particularly immune cells. A biallelic polymorphism (-1149G/T) in the prolactin promoter proved to be functionally important, as the modulation of prolactin expression was associated with lupus in some populations. Systemic lupus erythematosus (SLE) was associated with high levels of prolactin, low levels of dehydroepiandrosterone (DHEA) and induction of inflammatory cytokines in the serum of patients with the disease. One study suggests that the TT genotype may be a risk factor for lupus and may predict who could benefit from DHEA therapy.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
PRL	rs1341239	GT-	A	+ -	

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Sample

Factor V Leiden Mutation

NORMAL

Factor V Leiden (rs6025) is a variant (mutated form) of human factor V (one of several substances that help blood clotting), which causes an increase in blood clotting (hypercoagulability). Due to this mutation, protein C , an anticoagulant protein that normally inhibits the pro-coagulation activity of factor V, is not able to normally bind to factor V, leading to a hypercoagulable state, that is, an increased tendency for the patient form abnormalities and potentially harmful blood clots.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
F5	rs6020	AG-	T	+ -	
F5	rs6025	GG-	T	- -	

Antithrombin Deficiency

NORMAL

Heterozygous antithrombin deficiency has a prevalence of about 0.2 to 0.4%; approximately half of compromised people have venous thrombosis. Homozygous deficiency is likely to be lethal to the fetus in utero. Acquired deficiencies occur in patients with disseminated intravascular coagulation (DIC), liver disease or nephrotic syndrome, or during heparin therapy. Heparin exerts its anticoagulant effect through the activation of antithrombin.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
SERPINC1	rs28929469	CC-	A	- -	
SERPINC1	rs121909567	CC-	A	- -	
SERPINC1	rs201381904	CC+		- -	

Protein S Deficiency

NORMAL

It is a lack of S protein in the liquid portion of the blood. Proteins are natural substances that help prevent blood clots. Congenital protein C or S deficiency is an inherited disorder. This means that it is transmitted from parents to children. Congenital means present at birth. The disorder causes abnormal blood clotting. One in 300 people has a normal gene and a defective gene for protein C deficiency. Protein S deficiency is much less common and occurs in about 1 in 20,000 people.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
PROS1	rs6122	CC-	A	- -	
PROS1	rs121918474	AA-	C	- -	
PROS1	rs138925964	CC+		- -	
PROS1	rs387906674	CC-	A	- -	
PROS1	rs863224838	TT-	C	- -	

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Sample

Protein C Deficiency

LOW

It is a lack of C protein in the liquid portion of the blood. Proteins are natural substances that help prevent blood clots. Congenital protein C or S deficiency is an inherited disorder. This means that it is transmitted from parents to children. Congenital means present at birth. The disorder causes abnormal blood clotting. One in 300 people have a normal gene and a defective gene for protein C deficiency.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
IL-1B	rs16944	AG+	G	+ -	
PROC	rs121918143	CC+	T	- -	
PROC	rs121918150	GG+	A	- -	
PROC	rs121918153	GG+	A	- -	
PROC	rs121918154	CC+	T	- -	
PROC	rs121918160	CC+	T	- -	
PROC	rs757583846	CC+	T	- -	

Anti-Beta-2-Glycoprotein Antibody

UNDEFINED

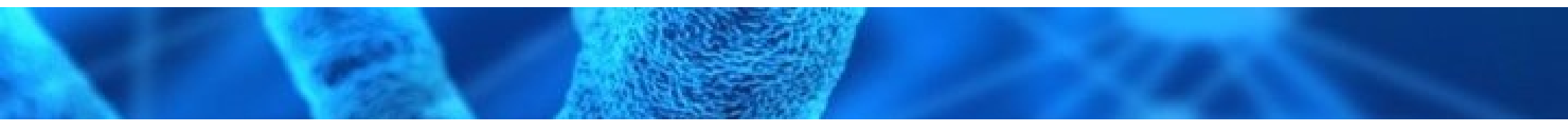
They are autoantibodies associated with an excessive tendency to coagulate, IgG, IgA or IgM classes. The B2GP1 antibodies are considered the main exponents of the antiphospholipid antibody class, capable of recognizing as lipids the lipoproteins (phospholipids) belonging to the organism to which they belong, present in the cell membrane of all cells and platelets. Anti-phospholipid antibodies interfere with blood clotting with mechanisms that are not fully understood. However, its presence is an important risk factor for the development of hypercoagulation disorders, with the consequent formation of venous and arterial thrombi. These antibodies are usually present in people affected by the autoimmune disease known as antiphospholipid syndrome (APS), associated with frequent thrombotic episodes, thrombocytopenia (low platelet count) or recurrent miscarriages and pre-eclampsia, particularly during the second and third trimesters of pregnancy .

Defect in Thyroid Hormonogenesis

UNDEFINED

Thyroid disorders are conditions that affect the thyroid, a butterfly-shaped gland located in the lower part of the neck. The thyroid plays an important role in regulating numerous metabolic processes throughout the body.

Metabolic



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Sample

Ceruloplasmin

NORMAL

Ceruloplasmin is a copper-containing glycoprotein with enzymatic activities. Produced in the liver, it contains about 90% of the total serum copper, showing late acute phase protein behavior. The main application of the use of ceruloplasmin is the diagnosis of Wilson's disease (hepatolenticular degeneration), an autosomal recessive disease, where plasma ceruloplasmin concentrations are typically reduced, and unbound and urinary copper are increased. Although the exact cause of Wilson's disease is not known, it is speculated that there is no enzyme or carrier protein capable of incorporating copper into proteins. Copper is then deposited in the kidneys, liver and brain. Unless chelation treatment is instituted, the disease is progressive and fatal. Increased values: inflammatory and neoplastic diseases (ceruloplasmin is a slow-phase protein), pregnancy, use of estrogens and copper poisoning.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CP	rs13072552	GG+	T	- -	

Methylation

MTHFR 1298 mutation (rs1801131)

MEDIUM-HIGH

Another known point mutation for the gene encoding the MTHFR enzyme is the nitrogenous base substitution at nucleotide 1298 (A1298C polymorphism). This mutation, like the C677T polymorphism, results in elevated levels of homocysteine. The homozygous genotype for the A1298C polymorphism is also considered pathogenic, as it considerably increases the risks for thrombotic events and recurrent miscarriages. Individuals heterozygous for the two polymorphisms (C677T and A1298C) are also at high risk for vascular events.

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

MTHFR 677 mutation (rs1801133)

NORMAL

MTHFR is a key enzyme for certain biological processes, including the conversion of homocysteine into methionine. Mutations in genes that encode this enzyme cause a significant reduction in its activity, causing hyperhomocysteinemia. This condition is related to increased risk for cardiovascular disease and poor pregnancy outcomes. The homozygous genotype for the C677T polymorphism is associated with a 25% increase in the plasma concentration of homocysteine, which can generate genetic alterations in the fetus of pregnant women and thromboembolism.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MTHFR	rs1801133	CC-	A	- -	

Psychiatric

Name Sample

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Sample

Postpartum depression

MEDIUM-HIGH

Postpartum depression encompasses depressive symptoms that last more than 2 weeks after delivery and interfere with activities of daily living. Postpartum depression is much more than just passing sadness. It is a common disease - it affects 20 to 35% of women - but a serious one, which can and must be treated.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
NR3C2	rs2070951	GG-	G	+ +	

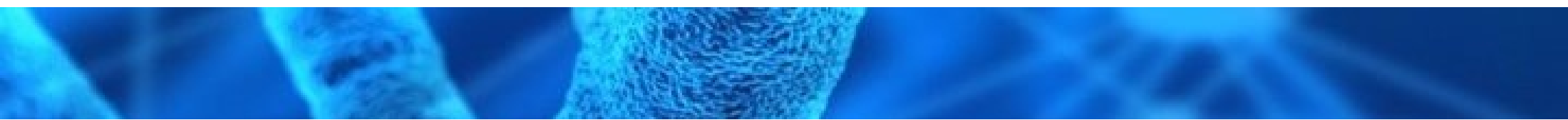
Reproductive system

Increased Excitation Levels

HIGH

The allele A of the rs53576 polymorphism (OXTR) was shown to be correlated with increased levels of excitation. The simultaneous presence of polymorphisms of the rs2234693 allele T and rs53576 allele A (OXTR) (group T + A) was correlated with increased levels of excitation. There was a statistical trend of increased desire levels in the T + A group.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ESR1	rs2234693	CT+	A,T	+ -	
OXTR	rs53576	AA+	A	+ +	



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Sample

Intra-Hepatic Pregnancy Cholestasis

MEDIUM-HIGH

It is a complication that can appear in the third trimester of pregnancy and affects about 1% of pregnant women.

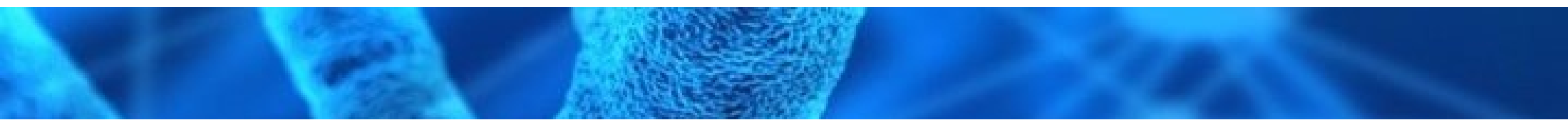
Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCB11	rs2287616	TT-	G	- -	
ABCB11	rs2287622	GT-	C,G,T	- -	
ABCB11	rs11568372	AA-	C	- -	
ABCB11	rs147649016	CC+	T	- -	
ABCB4	rs1202283	AA+	A	+ +	
ABCB4	rs45575636	GG-	T	- -	
ABCB4	rs72552778	CC-	A	- -	
ABCB4	rs794727183	GG-	T	- -	
ABCB4	rs863225298	CC-	C	- -	
ABCC2	rs3740066	AG-	G,T	+ -	
ABCC2	rs8187710	GG+	A	- -	
ABCC2	rs17222723	TT+	A	- -	
ATP8B1	rs121909100	TT-	G	- -	
NR1H4	rs113090017	CC+	A,G,T	- -	
OPRM1	rs1799971	GG+	G	+ +	
SLC25A13	rs80338722	GG-	T	- -	

Spontaneous abortions

MEDIUM-HIGH

The C allele of the CYP19A1 rs10046 polymorphism is associated with greater susceptibility to spontaneous abortions.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CYP19A1	rs10046	CC-	A	+ +	



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Sample

Pelvic Organ Prolapse

MEDIUM-HIGH

Pelvic organ prolapse occurs when the pelvic muscles and ligaments are weakened and unable to hold the pelvic organs in place. Thus, the organs can move to the pelvic region. Prolapsed women often have a feeling of heaviness over the vagina or pelvis. They may complain of a feeling of being "sitting in a ball" or notice a lump or mass coming out of the vaginal area while bathing. Sometimes this feeling of heaviness or bulging is felt more towards the end of the day, after work or standing all day. Other symptoms include pain during intercourse, urinary incontinence and bowel problems.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COL3A1	rs1800255	AG+	A	+ -	

Polycystic Ovary Syndrome

MEDIUM-HIGH

Hormonal disorder that causes an increase in the size of the ovaries, with small cysts on the outside of the ovaries.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
DENND1A	rs2479106	GG+	G	+ +	
INSR	rs1799817	CT-	A	+ -	
KISS1	rs12998	GG-	T	- -	
LEPR	rs1137101	AA+	G	- -	
LHCGR	rs2293275	GG-	C	- -	
LHCGR	rs13405728	AA+	G	- -	
MTNR1B	rs10830963	CC+	G	- -	
SUOX	rs705702	TT-	G	- -	
THADA	rs13429458	AA+	C	- -	
TNF	rs361525	GG+	A	- -	

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Sample

Pregnancy

MEDIUM

Ease or not to get pregnant.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ABCB11	rs2287622	GT-	C,G,T	- -	
ABCC2	rs2273697	GG+	A	- -	
ABCC2	rs8187710	GG+	A	- -	
ABCC2	rs17222723	TT+	A	- -	
AGT	rs699	CT-	G	+ -	
AGT	rs4762	CC-	A	- -	
AGTR1	rs5186	AA+	C	- -	
CDCA3	rs5443	CT+	T	+ -	
TCF7L2	rs12255372	GG+	T	- -	
TLR4	rs4986791	CC+	T	- -	

Risk of Recurring Pregnancy Loss

MEDIUM

The occurrence of a spontaneous pregnancy loss before the 20th week is relatively common. However, what happens over and over again is not an affliction that is easily seen among couples trying to get pregnant. Women who suffer from it often spend a long time in search of a better understanding of what may be happening and what are the possible treatments. In 2008, the American Society for Reproductive Medicine defined recurrent miscarriage as the occurrence of 2 or more miscarriages of less than 20 weeks' gestation. What many don't know is that two or more consecutive miscarriages before 20 weeks' gestation occur in 2% to 4% of couples of childbearing age. In Brazil, in 2014, there were around 3 million births, with an estimated around 90,000 new couples with recurrent pregnancy loss. Recurrent pregnancy loss is an infrequent and very complex pathology. There are numerous factors possibly responsible for the losses. Some of these factors have strong evidence, with a consensus. Other causes are widely discussed, deserving further studies. The main causes described are: Genetic, Endocrine (Hormonal), Anatomical, Infectious, Hematological (Thrombophilias), Immunological, Environmental, Nutritional status, Unknown. Orange or red result indicates "increased" risk of recurrent pregnancy loss.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ALDH2	rs671	GG+	A	- -	
IL-10	rs1800871	CT-	G	+ -	
IL-10	rs1800872	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	
MTR	rs1805087	GG+	G	- -	

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Sample

Neural Tube Defect

MEDIUM

Neural tube defects are a specific type of congenital defect of the brain, spine, and/or spinal cord.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CBS	rs4920037	GG+	A	- -	
MTHFD1	rs2236225	CC-	A	- -	
MTHFR	rs1801131	AC-	G	+ -	
MTHFR	rs1801133	CC-	A	- -	
MTHFR	rs2274976	GG-	T	- -	
MTHFR	rs3737967	CC-	A	- -	
MTRR	rs1801394	AA+	G	- -	

Endometriosis

NORMAL

A disorder in which the tissue that normally lines the uterus grows outside the uterus. This disorder can cause female infertility and have severe menstrual cramping symptoms. It affects women of different ages and even children.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
CDKN2B-AS1	rs10965235	CC+	A	- -	
ESR1	rs9340799	AG+	G	+ -	
GRCH37.1	rs12700667	AG+	A	- -	
GREB1	rs7576826	CT+	C	- -	
GREB1	rs13394619	AG+	G	- -	
IL-6	rs1800795	CG+	G	+ -	
INTERGENIC	rs10508881	AG+	A	+ -	
KSR2	rs10431397	CC+	T	- -	
MUC4	rs2246901	TT-	A,T	- -	
TCN2	rs1801198	GG+	A,C	- -	
VEZT	rs10859871	AC+	C	+ -	

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Sample

Uterine Fibroids

NORMAL

Uterine fibroids are benign uterine tumors of a smooth muscle nature. Fibroids often cause abnormal uterine bleeding, pelvic pain and pressure, urinary and bowel symptoms, as well as pregnancy complications. Diagnosis is made by clinical pelvic examination, ultrasound, or imaging tests.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BET1L	rs2280543	CC+	T	- -	
ESR1	rs9340799	AG+	G	+ -	
INTERGENIC	rs7913069	CC+	T	- -	
TNRC6B	rs12484776	AG+	G	+ -	

Uterine Fibromyoma

NORMAL

Benign tumors in the uterus that can develop during a woman's childbearing age.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
BET1L	rs2280543	CC+	T	- -	
ESR1	rs9340799	AG+	G	+ -	
INTERGENIC	rs7913069	CC+	T	- -	
TNRC6B	rs12484776	AG+	G	+ -	

Gestational diabetes

NORMAL

High blood sugar levels that affect pregnant women.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
INTERGENIC	rs7923837	AA+	A,T	- -	
SRR	rs391300	GG-	T	- -	
TCF7L2	rs7903146	CC+	G,T	- -	

Ovarian Hyperstimulation Syndrome

NORMAL

Ovarian Hyperstimulation Syndrome (OHS) is a complication resulting from ovarian stimulation with ovulation inducers, which can lead to severe conditions and, in extreme cases, even death. It is usually due to an exacerbated response of the ovaries to the hormonal stimulus and results in a large number of oocytes.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FSHR	rs28928870	CC-	A,T	- -	

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Sample

In vitro fertilization

NORMAL

In vitro fertilization is a medically assisted reproduction technique that consists of placing, in a laboratory environment, a significant number of sperm. Indication in orange and red indicate greater chances of the technique being successful.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
ESR2	rs1256049	GG-	T	- -	

Premature Ovarian Insufficiency

NORMAL

In primary ovarian failure, normal functioning of the ovaries is disrupted in women < 40 years of age. This disorder used to be called premature ovarian failure or premature menopause; however, these terms are misleading because women with primary ovarian failure do not always stop menstruating and their ovaries do not always stop working completely. Thus, a diagnosis of primary ovarian failure does not always mean that pregnancy is impossible. Furthermore, this disorder does not imply that a woman is aging prematurely; it just means that your ovaries are no longer working normally. A study found a positive association of allele A of the COMT rs4680 polymorphism with POI, reflecting on the genotype. The allele acts in a dominant mode. The study suppose that it may produce increased damage to ovarian cells, leading to POI.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
COMT	rs4680	AG+	A	+ -	

Sexual Motivation (Female)

NORMAL

Research shows that female sexual desire and arousal problems have a hereditary component. Previous molecular genetic studies of sexual desire have focused primarily on genes associated with neurotransmitters such as dopamine and serotonin. However, there are reasons to believe that hormones with more specific functions related to sexuality can have an impact on desire. We found nominally significant main effects on sexual desire of three ESR2-linked SNPs when controlled for anxiety, suggesting that individuals homozygous for the G allele of SN12 rs1271572 and the A allele of SNPs rs4986938 and rs928554 had lower levels of sexual desire. SNP rs4986938 also had a nominally significant effect on lubrication.

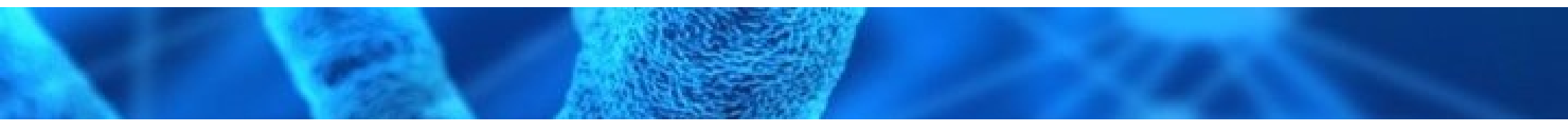
Gene	RSID	Genotype	Minor Allele	Alteration	Result
ESR2	rs4986938	GG-	T	- -	

Female Infertility

NORMAL

Causes of female infertility include ovulatory disorders, chromosomal abnormalities, endometriosis, pelvic adhesions, tubal blockage and other tubal abnormalities, and hyperprolactinemia. There is growing evidence that genetic abnormalities are present in infertile females and males.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
FSHR	rs6166	AG-	T	+ -	



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Sample

Ovarian Response to Hormonal Stimulation

UNDEFINED

Red or orange results indicate a worse ovarian response to hormonal stimulation.

Infertility in Endometriosis

UNDEFINED

While it is possible to get pregnant with endometriosis, the disease, without treatment, is still a major cause of female infertility. About 15% to 45% of patients with endometriosis actually have difficulty getting pregnant.

Skeletal system (bones)

Development Defects

NORMAL

It is the occurrence of intrauterine developmental delay or failure.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MTHFD1	rs2236225	CC-	A	- -	

Skin

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).

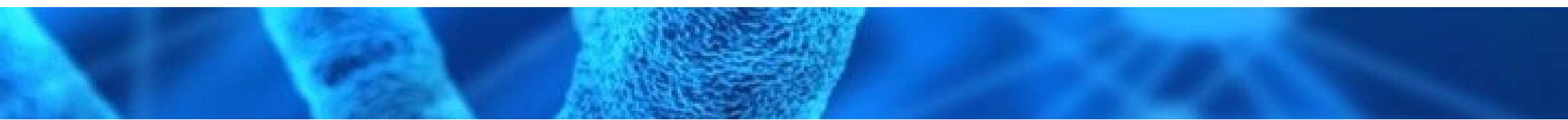
Vitamins need

Riboflavin Deficiency

NORMAL

Vitamin B2, also known as riboflavin, plays important roles in the body, such as increasing blood production, maintaining proper metabolism, promoting growth, and protecting vision and the nervous system.

Gene	RSID	Genotype	Minor Allele	Alteration	Result
MTHFR	rs1801133	CC-	A	- -	



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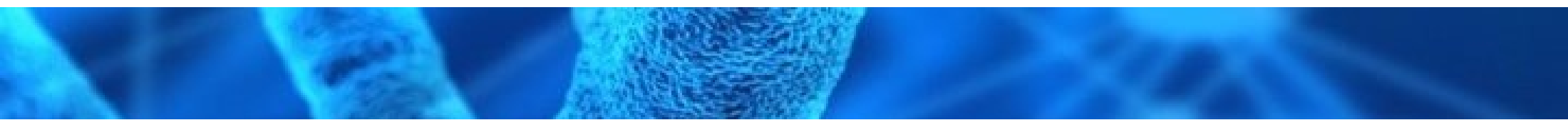

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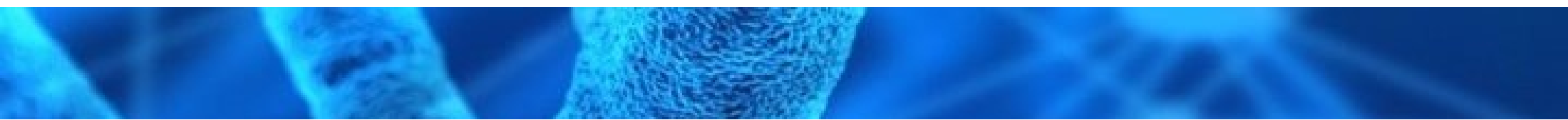

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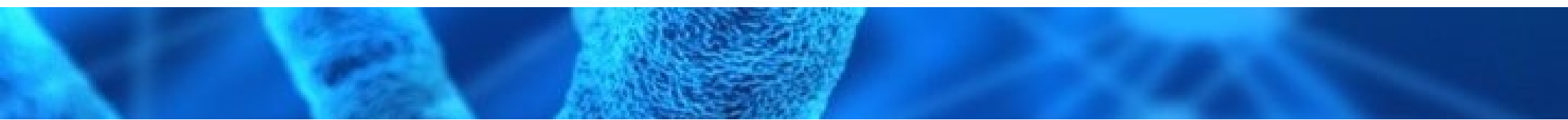

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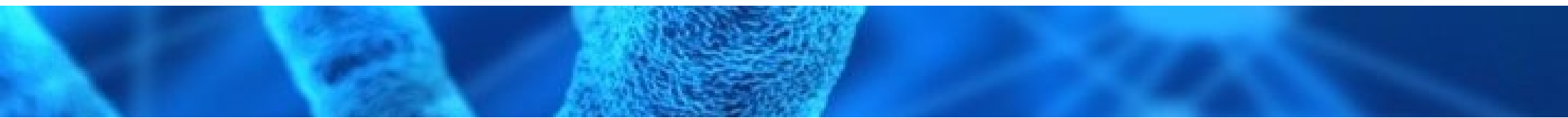

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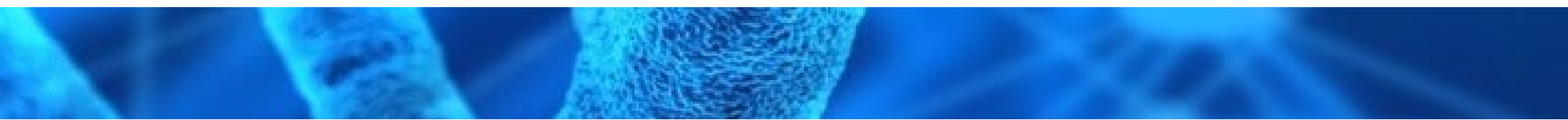

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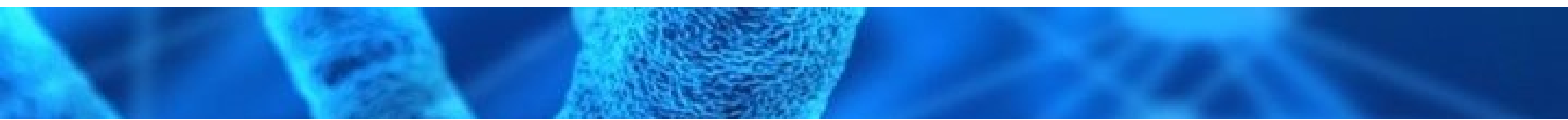

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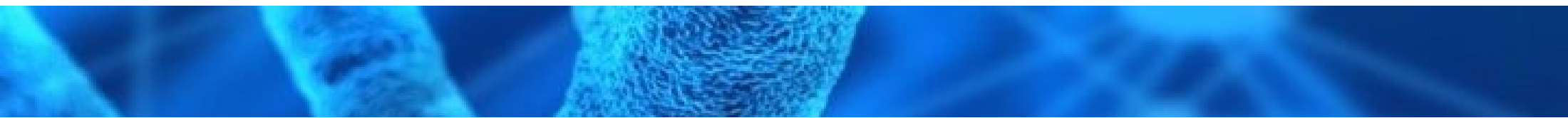

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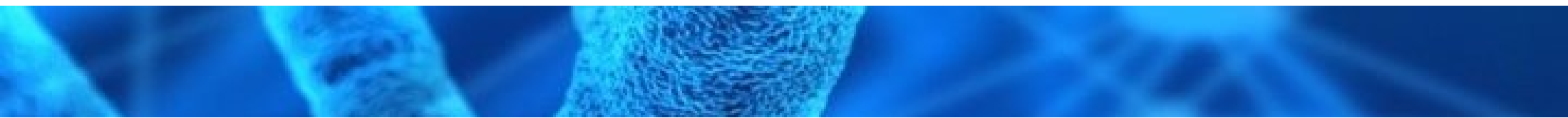

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