

HEALTH PANEL

ONCOLOGIC

ENGLISH VERSION

\sim	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021	
FullDNA	Prescriber:	Health Insurance:		Ⅲ ⅢⅢ Ⅲ ⅠⅢ ■Ⅲ ■Ⅱ ■ 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 laboratory is not responsible for any treatment based on the results.

If necessary, this laboratory has scientific advice to discuss these results with your attending clinician.

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.

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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, developed in Israel, allows the high level of precision and reliability of our exams in the fundamental aspects for a genetic exam.





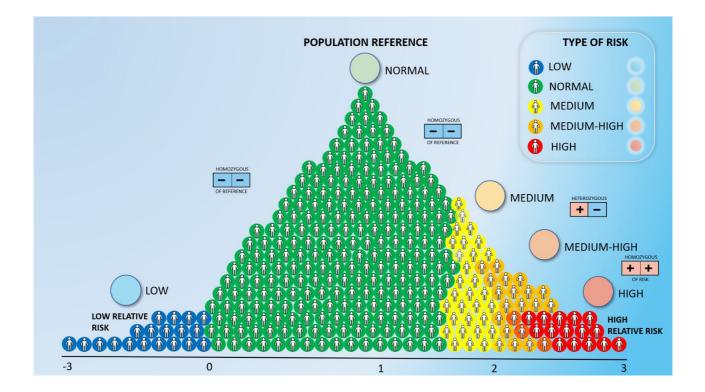
How to interpret the exam:

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 ball will appear
- If the susceptibility is AVERAGE, a YELLOW ball will appear
- If the susceptibility is NORMAL a GREEN ball will appear
- If the susceptibility is LOW, a BLUE ball will appear
-) If the condition is not identified GRAY ball

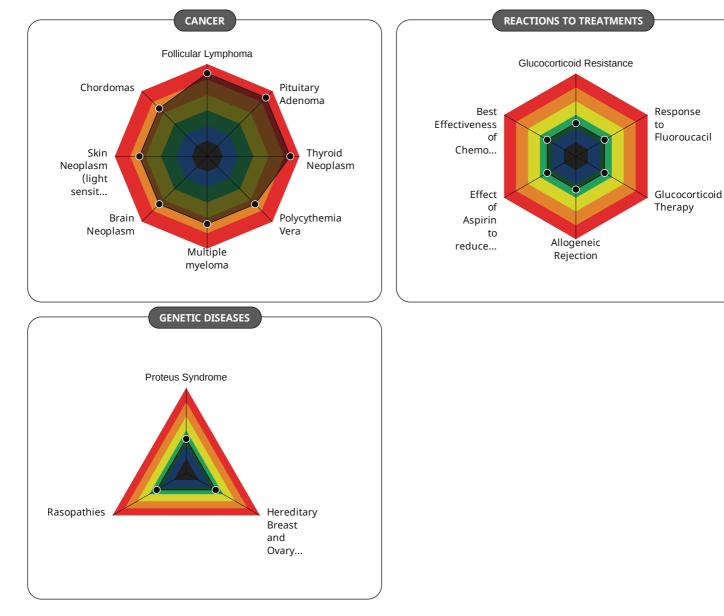


PART TWO

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



MOST RELEVANT CONDITIONS BY CATEGORY





Name: Sample			
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SUMMARY OF RESULTS

Cancer	
Follicular Lymphoma	0 1 + - 0 + + HIGH
Pituitary Adenoma	0 0 + - 1 + + HIGH
Thyroid Neoplasm	3 2 + - 1 + + HIGH
Polycythemia Vera	8 0 + - 2 + + MEDIUM- HIGH
Multiple myeloma	3 1 + - 2 + + MEDIUM- HIGH
Brain Neoplasm	0 2 + - 0 + + MEDIUM- HIGH
Skin Neoplasm (light sensitivity)	0 2 + - 0 + + MEDIUM- HIGH
Chordomas	0 0 + - 1 + + OMEDIUM- HIGH
Colorectal Neoplasm	6 3 + - 4 + + OMEDIUM- HIGH
Neuroblastoma	1 0 + - 3 + + MEDIUM- HIGH
Metastasis	1 1 + - 0 + + OHEDIUM- HIGH
Nasopharyngeal Neoplasm	2 - 2 + - 1 + + MEDIUM- HIGH
Astrocytoma	2 - 0 + - 1 + + MEDIUM- HIGH
Bladder Neoplasm	2 - 2 + - 1 + + MEDIUM- HIGH
Neoplasms (General Risk)	13 - 11 + 5 + + MEDIUM- HIGH
Neoplasm: Leukemia (Blood)	1 1 + - 1 + + OMEDIUM- HIGH
Gastrointestinal Stromal Tumor	2 3 + - 0 + + MEDIUM- HIGH
Glioma	4 2 + - 1 + + MEDIUM- HIGH
Cervical Neoplasm	0 0 + - 1 + + OMEDIUM- HIGH
Skin Neoplasm (Basic Cell Carcinoma - BCC)	2 0 + - 1 + + MEDIUM- HIGH
Stomach Neoplasm	4 2 + - 0 + + MEDIUM- HIGH
Colorectal Neoplasm (Meat Consumption)	4 4 + - 0 + + OHEDIUM- HIGH

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FullDNA

Name:	Sample
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Age: Prescriber:

Gender: F

Health Insurance:



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Lung Neoplasm	15 10 + - 1 + + 🕖 MEDIUM
Diffuse large B-cell lymphoma	0 1 + - 0 + + OMEDIUM
Liver Neoplasm	2 2 + - 0 + + MEDIUM
Meningioma	1 1 + - 1 + + MEDIUM
Acute Lymphoblastic Leukemia (ALL)	5 2 + - 0 + + MEDIUM
Breast neoplasm	49 10 + - 1 + + OMEDIUM
Ovary Neoplasm	17 - 1 + - 1 + + OMEDIUM
Breast Neoplasm (Family)	12 1 + - 0 + + 🔵 NORMAL
Hereditary Cancer Predisposition Syndrome	20 1 + - 0 + + 🔵 NORMAL
Tumor calcinosis	2 0 + - 0 + + NORMAL
Myeloproliferative Disorder	6 0 + - 0 + + 🔵 NORMAL
Tumor Necrosis Factor Alpha	1 0 + - 0 + + 🔵 NORMAL
Non-Hodgkin's Lymphoma	6 1 + - 0 + + 🔵 NORMAL
Peripheral T cell lymphoma	1 0 + - 0 + + 🔵 NORMAL
Medulloblastoma	6 0 + - 0 + + NORMAL
Oral Cavity and Laryngeal Neoplasm	1 0 + - 0 + + NORMAL
Skin Neoplasm (in redheads)	3 0 + - 0 + + NORMAL
Li-Fraumeni Syndrome	15 0 + - 0 + + NORMAL
Lynch Syndrome	3 0 + - 0 + + NORMAL
Hereditary Tyrosinemia Type 1	5 0 + - 0 + + NORMAL
Wilms' Tumor	2 0 + - 0 + + NORMAL
Hereditary Nonpolyposis Colorectal Neoplasm	1 0 + - 0 + + NORMAL
Multiple Endocrine Neoplasm	3 1 + - 0 + + NORMAL
Pancreas Neoplasm	5 0 + - 0 + + 🔵 NORMAL
Malignant Pleural Mesothelioma	1 0 + - 0 + + 🔵 NORMAL
Retinoblastoma	9 0 + - 0 + + 🔵 NORMAL
Myelodysplastic Syndrome	3 0 + - 0 + + 🔵 NORMAL
Lung adenocarcinoma	1 0 + - 0 + + 🔵 NORMAL
JAK2 V617F mutation	1 0 + - 0 + + 🔵 NORMAL
Schwannoma	1 0 + - 0 + + 🔵 NORMAL
Skin Neoplasm (Melanoma)	9 1 + - 0 + + OLOW
Endometrial Neoplasm	5 1 + - 0 + + OLOW
Osteosarcoma	1 0 + - 0 + + OLOW

\sim	Name: Sample			
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FullDNA	Prescriber:	Health Insurance:		 Sample

Digestive s	system
Bowel polyps	2 0 + - 0 + + NORMAL
Juvenile Polyposis Syndrome	6 0 + - 0 + + NORMAL
Gener	al
Hepatocellular Carcinoma (HCC)	3 1 + - 0 + + NORMAL
Gene	S
GSTT1	1 0 + - 0 + + NORMAL
Genetic dis	seases
Proteus Syndrome	1 0 + - 0 + + NORMAL
Hereditary Breast and Ovary Cancer Syndrome	10 1 + - 0 + + 🔵 NORMAL
Rasopathies	1 0 + - 0 + + NORMAL
Hematologic	c system
Idiopathic Hypereosinophilic Syndrome	1 0 + - 0 + + NORMAL
Immune s	ystem
Neurofibromatosis	10 0 + - 0 + + NORMAL
Neurodegenerat	tive diseases
Louis-Bar Syndrome (Ataxia Telangiectasia)	4 0 + - 0 + + NORMAL
Oncolo	gic
Cervical Cancer	2 0 + - 0 + + NORMAL
BRAF V600E mutation	1 0 + - 0 + + NORMAL
Reactions to T	reatments
Glucocorticoid Resistance	1 0 + - 0 + + NORMAL
Response to Fluoroucacil	1 0 + - 0 + + 🔵 NORMAL
Glucocorticoid Therapy	0 1 + - 0 + + 🔵 NORMAL
Allogeneic Rejection	1 0 + - 0 + + 🔵 NORMAL

\sim	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021	
FullDNA	Prescriber:	Health Insurance:		II III III Sample

Effect of Aspirin to reduce risk of colorectal neoplasia	1 0 + - 0 + + 🔵 NORMAL
Best Effectiveness of Chemotherapy	1 0 + - 0 + + NORMAL

\mathbf{a}	Name: Sample	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021		
DNA	Prescriber:	Health Insurance:		Sa	

Cancer

Follicular Lymphoma

Follicular lymphoma (FL) is a type of non-Hodgkin lymphoma. FL develops when B cells become abnormal (cancerous). B-cells are white blood cells that normally help fight infection. They are sometimes called B-lymphocytes.

Gene	Genotype	Minor Allele	Alteration	Result
INTERGENIC	GT+	G	+ -	•

Pituitary Adenoma

Pituitary adenoma, also known as pituitary adenoma, is a type of benign pituitary tumor, which is a gland located in the brain and is responsible for controlling the production of hormones such as cortisol, prolactin, growth hormone and hormones that stimulate functioning ovaries and testicles, for example. This type of tumor is rare and, because it is benign, does not put life at risk, however it can cause symptoms that decrease the quality of life such as infertility, decreased libido, milk production or neurological symptoms such as headache or partial loss of vision.

Gene	Genotype	Minor Allele	Alteration	Result
TP53	GG-	C,T	+ +	•

Thyroid Neoplasm

Thyroid neoplasm is a malignant tumor of the thyroid gland that is located in the neck. It is the fifth most common type of cancer in women and the seventeenth most prevalent in men. Its clinical course is indolent, it has a good evolution and satisfactory response to treatment in most cases.

Gene	Genotype	Minor Allele	Alteration	Result
CASC8	GG+	Т	+ +	•
DIRC3	CT+	G,T	+ -	\bigcirc
HABP2	GG+	А		
INTERGENIC	CT+	Т	+ -	•
INTERGENIC	GG+	G,T		
RET	Π+	С		



HIGH

HIGH

HIGH

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FullDNA	

Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Polycythemia Vera

Polycythemia vera (PV) is a disorder of the blood-producing cells of the bone marrow that results in overproduction of red blood cells, increased hematocrit. There may also be an increase in platelets and white blood cells.

Gene	Genotype	Minor Allele	Alteration	Result
FGFR3	AA+	G		
JAK2	CC+	С	+ +	•
JAK2	GG+	A,T		
MPL	GG+	А		
MPL	GG+	A,C		
MPL	CC+	Т		
MPL	TT+	А		
MPL	CC+	Т		
NOG	TT+	G		
SH2B3	TT+	A,C,G	+ +	•

Multiple myeloma



MEDIUM-

HIGH

Cancer of plasma cells.

Gene	Genotype	Minor Allele	Alteration	Result
BRAF	AA-	С	-	
DTNB	TT+	Т	+ +	•
HRAS	GG-	A,C,T		
LIG4	CT-	А	+ -	\bigcirc
SP3	TT+	С	+ +	•
TP53	GG-	A,G,T		

\sim	Name: Sample				
	Age:	Gender: F	Report Date: 15	5/11/2021	
FullDNA	Prescriber:	Health Insurance:			

Brain Neoplasm

A brain tumor is characterized by the presence and proliferation of abnormal cells in the brain or meninges, which can happen due to genetic mutations or due to metastasis of cancer from other parts of the body. Typically, brain tumors can be classified into 4 grades according to their rate of growth: Brain tumor grade 1 and 2: Slow-growing brain tumor that rarely spreads to other regions of the brain. These are usually benign brain tumors such as glioblastoma or meningioma; Grade 3 and 4 brain tumor: Usually a malignant brain tumor that grows rapidly. The brain tumor rarely metastasizes, that is, it spreads to other parts of the body. Normally, malignant cells develop and proliferate in the brain itself. Most brain tumors are benign and have well-defined limits, that is, they are curable and can be easily treated with chemotherapy, radiotherapy or surgical removal.

Gene	Genotype	Minor Allele	Alteration	Result
MTHFR	AG-	G,T	+ -	
MTHFR	CT-	А	+ -	•

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

Gene	Genotype	Minor Allele	Alteration	Result
ASIP	AG-	Т	+ -	\bigcirc
ASIP	GT+	G	+ -	•

Chordomas

Chordomas are slow-growing malignant tumors that form as remnants of the notochord. One third forms at the base of the skull, and may also form in the lumbosacral region. Histologically, they are benign, but present malignant behavior because they are invasive and due to their capacity to metastasize.

Gene	Genotype	Minor Allele	Alteration	Result
TBXT	TT+	Т	+ +	\bigcirc





MEDIUM-HIGH

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Prescriber:



Colorectal Neoplasm

MEDIUM-HIGH

Colorectal neoplasia is the development of cancer in the colon or rectum, two segments of the large intestine.

Gender: F

Health Insurance:

Gene	Genotype	Minor Allele	Alteration	Result
CASC8	GG+	Т	+ +	•
CHEK2	Π-	C,G		
COLCA1	AA+	А		
EIF3H	AC+	С	+ -	•
GATA3	GT+	G	+ -	•
INTERGENIC	CC+	С		
INTERGENIC	AC+	С	+ -	•
MGMT	AA+	G		
MSH6	Π+	C,G		
MTHFD1	CC-	А	+ +	•
SMAD7	CC+	С	+ +	•
SMAD7	Π+	A,C	+ +	•
TCF7L2	CC+	G,T		

Neuroblastoma



MEDIUM-

HIGH

Cancer that is commonly found in the adrenal (adrenal) glands, located one over each kidney.

Gene	Genotype	Minor Allele	Alteration	Result
BARD1	TT+	G		
CASC15	CC+	C,G	+ +	•
CASC15	GG+	G	+ +	•
CASC15	AA+	А	+ +	•

Metastasis

Metastasis is the formation of a new tumor lesion from another. It can be considered the fact that a tumor has spread to other cells and organs.

Gene	Genotype	Minor Allele	Alteration	Result
FGFR4	CT-	А	+ -	\bigcirc
PDK1	CC+	Т		

\sim	Name: Sample				
	Age:	Gender: F	Report Date:	15/11/2021	
FullDNA	Prescriber:	Health Insurance:			

Nasopharyngeal Neoplasm



Most patients with nasopharyngeal cancer complain of a lump or mass in the neck. This is caused by the spread of the disease to the lymph nodes in the region, which increases in size.

Gene	Genotype	Minor Allele	Alteration	Result
GABBR1	AG-	Т	+ -	
GABBR1	CT-	C,G	+ -	•
HCG9	AA+	G		
HCG9	GG-	С	+ +	•
INTERGENIC	Π+	С		

Astrocytoma

Astrocytoma is a type of cancer that can occur in the brain or spinal cord. It begins in cells called astrocytes that support nerve cells. Some astrocytomas grow very slowly and others can be aggressive cancers that grow quickly. Astrocytoma is a type of cancer that can form in the brain or spinal cord.

Gene	Genotype	Minor Allele	Alteration	Result
KDR	CC-	G	+ +	•
KDR	CC+	Т		
VEGFR2	Π+	А		

Bladder Neoplasm

Bladder neoplasia is a malignant formation that settles in the cells that cover the inner walls of the bladder, a hollow and elastic muscular organ whose function is to store urine from the kidneys and then eliminate it through the urethra.

Gene	Genotype	Minor Allele	Alteration	Result
CASC11	GG+	A,T		\bigcirc
PTGS2	CT-	G,T	+ -	
TP63	AA-	С	+ +	•
XPC	CC-	А		
XRCC5	GT+	Т	+ -	•



MEDIUM-

HIGH





Name: Sample			
Age:	Gender: F	Report Date: 15/11/2021	
Prescriber:	Health Insurance:		Sampl

Neoplasms (General Risk)



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A disease in which abnormal cells divide uncontrollably and destroy body tissue. The graph on the right indicates the genetic predisposition to developing different types of cancer.

5	5 1 1	1 3	51	
Gene	Genotype	Minor Allele	Alteration	Result
ABCA1	GG-	Т		
ALDH2	GG+	А	+ +	•
ATM	CC-	G,T	-	\bigcirc
CCHCR1	AA-	G		
CLPTM1L	CT+	Т	+ -	•
CYP1A1	Π-	G		
CYP1A1	CC-	A,T		
CYP1A1	Π-	G,T		
CYP1B1	CG-	С	+ -	
CYP2E1	AA+	Т	+ +	•
CYP3A4	AA-	Т		
CYP3A5	GG-	С	+ +	•
DIRC3	CT+	G,T	+ -	•
EPHX1	CT+	С	+ -	
EPHX1	AA+	G,T		
FASLG	CT+	Т	+ -	\bigcirc
G6PD	GG-	Т		
MIR146A	GG+	G		
MTHFR	AG-	G,T	+ -	
MTHFR	AC-	A,C,G	+ -	•
MTHFR	CT-	А	+ -	•
MTRR	AG+	G	+ -	0
PTEN	GG+	A,C,T		
RNASEL	CC-	G	+ +	•
SLC39A6	Π-	А	+ +	•
SOD2	Π-	G		\bigcirc
XPC	CC-	А		
XPC	AC-	Т	+ -	
XRCC1	CT-	А	+ -	<u> </u>



Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Neoplasm: Leukemia (Blood)



It is a cancer that occurs in the formation of blood cells, making it difficult for the body to fight infections.

Gene	Genotype	Minor Allele	Alteration	Result
IRF4	GG+	G,T	+ +	\bigcirc
JAK2	GG+	A,T		
NQO1	CT-	А	+ -	\bigcirc

Gastrointestinal Stromal Tumor



They originate from the interstitial cells of cajal and are the most common mesenchymal neoplasm of the gastrointestinal tract, 5% to 6% of all sarcomas and

Gene	Genotype	Minor Allele	Alteration	Result
MTHFR	AG-	G,T	+ -	\bigcirc
MTHFR	AC-	A,C,G	+ -	•
MTRR	CC+	Т		
MTRR	AG+	G	+ -	
SHMT1	GG+	А		\bigcirc

Glioma



A type of tumor that occurs in the brain and spinal cord.

Gene	Genotype	Minor Allele	Alteration	Result
CCDC26	Π+	G	-	
CDKN2B-AS1	GG+	А		\bigcirc
EGFR	AG+	G,T	+ -	•
PTEN	GG+	А		
SEL1L	Π+	С		
SOD3	CC+	Т	+ +	•
TERT	GT-	А	+ -	\bigcirc



Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Cervical Neoplasm



Cervical cancer is a type of cancer that occurs in the cells of the cervix - the lower part of the uterus that connects to the vagina. The graph on the right indicates the genetic predisposition to develop this condition.

Gene	Genotype	Minor Allele	Alteration	Result
IL-12B	AA-	G	+ +	•

Skin Neoplasm (Basic Cell Carcinoma - BCC)



They are cancers that form in the skin and are characterized by the abnormal growth of cells in the skin.

Gene	Genotype	Minor Allele	Alteration	Result
INTERGENIC	Π+	G	+ +	•
KRT5	GG-	Т		ightarrow
PADI6	GG+	А		ightarrow

Stomach Neoplasm



Stomach cancer is also known as gastric cancer. Stomach cancer develops slowly over many years. Before the onset of cancer itself, precancerous changes occur in the inner lining of the stomach (mucosa). Stomach cancer is also called gastric cancer. The adenocarcinoma type accounts for about 95% of stomach tumor cases. Other types of tumors, such as lymphomas and sarcomas, can also occur in the stomach. Lymphomas are diagnosed in about 3% of cases. Sarcomas are rare tumors that start in the tissues that give rise to muscles, bones and cartilage. One type that can affect the stomach is the gastrointestinal stromal tumor, better known as GIST. Stomach adenocarcinoma affects mostly men in their 60s to 70s. About 65% of patients are over 50 years old. In Brazil, stomach cancer is the third most common type among men and the fifth among women.

Gene	Genotype	Minor Allele	Alteration	Result
AURKA	GG-	С		
LTA	CC+	Т		
MTHFR	AG-	G,T	+ -	\bigcirc
MTHFR	CT-	А	+ -	•
PLCE1	AA+	G		
SERPINE1	CC+	Т		



Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Colorectal Neoplasm (Meat Consumption)



Excessive consumption of red meat can be a risk factor for colorectal cancer, especially in individuals with these polymorphisms, if indicated in orange or red.

Gene	Genotype	Minor Allele	Alteration	Result
GATA3	GT+	G	+ -	•
MLH1	AG+	C,G,T	+ -	\bigcirc
NAT2	AG+	G	+ -	
NAT2	CC+	Т		
NAT2	CT+	Т	+ -	\bigcirc
NAT2	GG+	А		
NAT2	GG+	А		
NAT2	CC+	A,T		\bigcirc

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FullDNA

Age:





Health Insurance:

Gender: F



Lung Neoplasm

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Cancer that starts in the lungs and most often occurs in smokers.

Gene	Genotype	Minor Allele	Alteration	Result
ABCB1	CC-	G		
ABCB1	Π-	G		
ATM	CC-	G,T		
CHEK2	Π-	C,G		
CHRNA3	CT-	А	+ -	•
CXCR4	CC-	А		
CYP1A1	AC-	A,T	+ -	•
CYP1A1	Π-	G		
CYP1A1	CC-	A,T		
CYP1A1	Π-	G,T		
CYP24A1	CC+	Т		
EGFR	GG-	Т		\bigcirc
ERCC2	Π+	A,G		\bigcirc
FASLG	CT+	Т	+ -	•
G6PD	GG-	Т		
НҮКК	Π+	С		
KLF6	CC+	Т		
MGMT	AA+	G		
MTHFR	AG-	G,T	+ -	
MTHFR	CT-	А	+ -	•
NQO1	CT-	А	+ -	\bigcirc
PER3	AG-	Т	+ -	
SOD2	TT-	G	+ +	•
TERT	GT-	А	+ -	0
XPC	AC-	Т	+ -	
XRCC1	CT-	А	+ -	<u> </u>

\sim	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021	
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Diffuse large B-cell lymphoma



MEDIUM

Diffuse large B-cell lymphoma (DLBCL) is a cancer of B cells, a type of lymphocyte that is responsible for producing antibodies. It is the most common form of non-Hodgkin lymphoma among adults, with an annual incidence of 7–8 cases per 100,000 people per year in the US and UK. This cancer occurs primarily in older individuals, with a median age of diagnosis at ~70 years, although it can occur in young adults and, in rare cases, children. DLBCL can arise in virtually any part of the body and, depending on various factors, is often a very aggressive malignancy. The first sign of this illness is typically the observation of a rapidly growing mass or tissue infiltration that is sometimes associated with systemic B symptoms, e.g. fever, weight loss, and night sweats.

Gene	Genotype	Minor Allele	Alteration	Result
IL-6	CG+	G	+ -	<u> </u>

Liver Neoplasm

Liver Neoplasm is a malignant tumor that originates from cells that make up the liver. Liver cancer is divided into two categories: primary liver and secondary, or metastatic (originated in another organ and affecting the liver as well). The term "primary liver" is used in tumors originating in the liver, such as hepatocarcinoma or hepatocellular carcinoma (most frequent primary malignant tumor occurring in more than 80% of cases), cholangiocarcinoma (which affects the bile ducts within the liver), angiosarcoma (blood vessel tumor) and, in children, hepatoblastoma. Despite not being among the most prevalent neoplasms, hepatobiliary cancer requires high complexity in its diagnosis and treatment proficiency.

Gene	Genotype	Minor Allele	Alteration	Result
CTLA4	AG+	G,T	+ -	•
FUT2	AA+	Т		
TERT	GT-	А	+ -	\bigcirc
TNF	GG+	А		lacksquare

Meningioma

MEDIUM

A benign tumor that usually arises in the membranes surrounding the brain and spinal cord.

Gene	Genotype	Minor Allele	Alteration	Result
MTRR	AG+	G	+ -	
PTEN	GG+	А		ightarrow
SOD3	CC+	Т	+ +	•





Acute Lymphoblastic Leukemia (ALL)



B-cell Acute Lymphoblastic Leukemia (ALL). A type of leukemia in which many B-cell lymphoblasts are found in the blood and bone marrow. It is the most common type of acute lymphoblastic leukemia. Also called precursor B lymphoblastic leukemia and B-cell acute lymphocytic leukemia.

Gene	Genotype	Minor Allele	Alteration	Result
ADAM28	CC+	G,T		
ARID5B	GT+		+ -	\bigcirc
ARID5B	AA+	G		
HLA-DQA1	GG+	А		\bullet
HLA-DQA1	AA+	G		
IKZF1	GT+		+ -	•
LTBR	Π+	A,C		\bigcirc

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FullDNA

Age:

Prescriber:





Breast neoplasm



Breast cancer is a disease caused by the multiplication of abnormal cells in the breast, which form a tumor. The graph on the right indicates the genetic predisposition to develop this condition.

ABCC4 GG+ A,T Image: Constraint of the second	Gene	Genotype	Minor Allele	Alteration	Result
AKT1GG+GMATMTT+CMATMCC+TMATMAA+TMATMCC+A,TMATMGG+CMATMGG+CMATMCC+TMATMCC+TMATMCC+TMATMCC+TMAURKAGG-CMBRCA1AG-A,CMBRCA1AG-A,CMBRCA1GG-CMBRCA1GG-G,TMBRCA1GG-TMBRCA2TT-C,GMBRCA2CC+A,TMBRCA2AA+GMBRCA2AA+GMBRCA2AA+GMBRCA2AA+TMBRCA2AA+TMBRCA2AA+TMBRCA2AA+GMBRCA2AA+TMBRCA2CC+A,GMBRCA2CC+A,GMBRCA2CC+A,GMBRCA2CC+A,GMBRCA2CC+A,GMCHEK2TT-C,GMCHEK2TT-C,GMCHEK2TT-C,GMCHEK2TT-C,GMCHEK2TT-C,GM<	ABCC4	GG+	A,T		
ATMTT+CIATMCC+TIATMAA+TIATMCC+A,TIATMGG+CIATMGG+CIATMCC+TIATMCC+TIATMCC+TIAUKKAGG-CIBRCA1AG-ACIBRCA1AG-ACIBRCA1AG-ACIBRCA1GG-GIBRCA1GG-GIBRCA1GG-GIBRCA2TT-C,GIBRCA2CC+A,TIBRCA2CC+TIBRCA2AA+GIBRCA2AA+GIBRCA2AA+GIBRCA2AA+GIBRCA2AA+TIBRCA2AA+TIBRCA2AA+TIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GIBRCA2CC+A,GICHEK2TT-C,GICHEK2TT-C,GICHEK2TT-C,G	AGER	GG-	А		
ATM CC+ T Image: Constraint of the second sec	AKT1	GG+	G		
ATM AA+ T Ist ● ATM CC+ A,T Ist ● ATM GG+ C Ist ● ATM TT+ C Ist ● ATM TT+ C Ist ● ATM CC+ T Ist ● AURKA GG- C Ist ● BRCA1 AG- A,C Ist ● BRCA1 AG- G,G Ist ● BRCA2 T- C,G Ist ● BRCA2 AA+ G Ist ● BRCA2 AA+ G Ist ●	ATM	TT+	С		
ATM CC+ A,T E ● ATM GG+ C E ● ATM TT+ C E ● ATM CC+ T ● ● AURKA GG- C E ● BRCA1 AG- A,C ● ● BRCA1 GG- G,T ● ● BRCA1 GG- G,T ● ● BRCA1 GG- T ● ● BRCA2 TT- C,G ● ● BRCA2 TT- C,G ● ● BRCA2 AA+ G ● ● BRCA2 AA+ T ● ● BRCA2	ATM	CC+	Т		
ATM GG+ C I O ATM TT+ C I O ATM CC+ T I O AURKA GG- C I O BRCA1 AG- A,C I O BRCA1 GG- G,T I O BRCA1 GG- G,T I O BRCA2 TT- C,G I O BRCA2 AA+ G I O BRCA2 AA+ T I O BRCA2	ATM	AA+	Т		
ATM TT+ C I I I ATM CC+ T I I I I AURKA GG- C I I I I I BRCA1 AG- A,C I <t< td=""><td>ATM</td><td>CC+</td><td>A,T</td><td></td><td></td></t<>	ATM	CC+	A,T		
ATM CC+ T Image: Second	ATM	GG+	С		
AURKA GG- C Image: Constraint of the second s	ATM	Π+	С		
BRCA1 AG- A,C M- O BRCA1 AA- C M- O BRCA1 AG- A,C M- O BRCA1 CC- A M- O BRCA1 GG- G,T M- O BRCA1 GG- T M- O BRCA2 TT- C,G M- O BRCA2 AA+ G M- O BRCA2 AA+ G M- O BRCA2 AA+ G M- O BRCA2 AA+ T M- O BRC	ATM	CC+	Т		
BRCA1AA-CImage: Constraint of the second se	AURKA	GG-	С		
BRCA1 AG- A,C Image: Constraint of the second	BRCA1	AG-	A,C	+ -	•
BRCA1CC-AIBRCA1AA-C,GIBRCA1GG-G,TIBRCA1GG-TIBRCA2TT-C,GIBRCA2AA+GIBRCA2CC+A,TIBRCA2AA+GIBRCA2AA+GIBRCA2AA+GIBRCA2AA+GIBRCA2AA+TIBRCA2CC+TIBRCA2AA+TIBRCA2AA+CIBRCA2AA+TIBRCA2CC+A,GIBRCA2CC+GICASC16CC-GICDKN1BCC+CICHEK2TT-C,GICHEK2TT-C,GI	BRCA1	AA-	С		
BRCA1 AA- C,G I O BRCA1 GG- G,T I O BRCA1 GG- T I O BRCA2 TT- C,G I O BRCA2 AA+ G I O BRCA2 AA+ G I O BRCA2 CC+ A,T I O BRCA2 AA+ G I O BRCA2 AA+ G I O BRCA2 AA+ G I O BRCA2 AA+ T I O BRCA2 CC+ A,G I O CASC16 CC- G I O CHEK2 TT-	BRCA1	AG-	A,C	+ -	
BRCA1 GG- G,T I I BRCA1 GG- T I I BRCA2 TT- C,G I I BRCA2 AA+ G I I BRCA2 CC+ A,T I I BRCA2 AA+ G I I BRCA2 CC+ A,T I I BRCA2 AA+ G I I BRCA2 AA+ G I I BRCA2 AA+ G I I BRCA2 AA+ T I I BRCA2 CC+ T I I BRCA2 AA+ C I I BRCA2 AA+ T I I BRCA2 AA+ T I I BRCA2 AA+ T I I BRCA2 CC+ A,G I I CASC16 CC- G I I CHEK2 TT-	BRCA1	CC-	А		
BRCA1 GG- T Image: Constraint of the stress of the s	BRCA1	AA-	C,G		
BRCA2 TT- C,G I I BRCA2 AA+ G I I BRCA2 CC+ A,T I I BRCA2 AA+ G I I BRCA2 AA+ G I I BRCA2 AA+ G I I BRCA2 AA+ T I I BRCA2 CC+ T I I BRCA2 AA+ C I I BRCA2 AA+ T I I BRCA2 AA+ C I I BRCA2 AA+ T I I BRCA2 AA+ T I I BRCA2 AA+ T I I BRCA2 CC+ A,G I I CASC16 CC- G I I CHEK2 TT- C,G I I	BRCA1	GG-	G,T		
BRCA2AA+GBRCA2CC+A,TBRCA2AA+GBRCA2AA+TBRCA2AA+TBRCA2CC+TBRCA2CC+TBRCA2AA+CBRCA2AA+CBRCA2AA+TBRCA2CC+A,GBRCA2CC+GCASC16CC-GCDKN1BCC+CCHEK2TT-C,G	BRCA1	GG-	Т		
BRCA2 CC+ A,T Image: Constraint of the second	BRCA2	TT-	C,G		
BRCA2 AA+ G Image: Constraint of the second s	BRCA2	AA+	G		
BRCA2 AA+ T Image: Constraint of the second s	BRCA2	CC+	A,T		
BRCA2 CC+ T Image: Constraint of the second s	BRCA2	AA+	G		
BRCA2 AA+ C Image: Constraint of the state of	BRCA2	AA+	Т		
BRCA2 AA+ T Image: Constraint of the second s	BRCA2	CC+	Т		
BRCA2 CC+ A,G Image: Constraint of the state of	BRCA2	AA+	С		
CASC16 CC- G ● CDKN1B CC+ C ● CHEK2 TT- C,G ●	BRCA2	AA+	Т		
CDKN1B CC+ C O CHEK2 TT- C,G O	BRCA2	CC+	A,G		
CHEK2 TT- C,G O	CASC16	CC-	G		
	CDKN1B	CC+	С		
CHEK2 GG- T	CHEK2	Π-	C,G		
	CHEK2	GG-	Т		



Name: Sample			
Age:	Gender: F	Report Date:	15/11/2021
Prescriber:	Health Insurance:		



COMT	CC+	Т		
CYP1B1	CG-	С	+ -	
FANCC	CC-	А		
FGFR2	CT-	G	+ -	
FGFR2	AG+	G,T	+ -	•
FGFR2	CT-	G	+ -	•
FGFR4	CT-	А	+ -	•
FTO	Π+	A,G		
HMMR	CC+	Т		
INTERGENIC	AA+	G		
INTERGENIC	AA+	G		
INTERGENIC	AA+	G		\bigcirc
MAP3K1	AC+	А	+ -	\bigcirc
MIR146A	GG+	G		
NCOA3	GG+	A,C		
NQO1	CT-	А	+ -	
RNF146	AA+	G		
SLCO1B3	AA+	А		
SLCO1B3	AA+	G		
TCF7L2	GG+	A,T	-	
TERT	CT+	Т	+ -	
TNF	GG+	А	-	
TP53	GG-	C,T	+ +	•
TP53	GG-	A,G,T		
TP53	CC-	A,C		
TP53	GG-	A,T		
VTCN1	AA+	А		
WRN	CC+	Т		

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	Age:	Gender: F	Report Date:	15/11/2021	
ulidna	Prescriber:	Health Insurance:			

Ovary Neoplasm

F



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	Genotype	Minor Allele	Alteration	Result
BRCA1	AG-	A,C	+ -	•
BRCA1	AA-	С		
BRCA1	CC-	А		
BRCA1	AA-	C,G		
BRCA1	GG-	G,T		
BRCA2	Π-	C,G		
BRCA2	AA+	G		
BRCA2	AA+	G		
BRCA2	TT+	С		
BRCA2	AA+	Т		
BRCA2	CC+	Т		
CYP3A4	AA-	Т		
ERCC2	TT+	A,G		\bullet
ESR1	GG+			
HNF1B	GG-	G,T	+ +	•
INTERGENIC	GG+	А		
MAGEC3	AA+	А		
TIPARP	TT+	Т		
TP53	GG-	Т		

\sim	Name: Sample		
	Age:	Gender: F	Report Date: 15/11/2021
FullDNA	Prescriber:	Health Insurance:	



Breast Neoplasm (Family)

NORMAL

Cancer that forms in the cells of the breasts.

Gene	Genotype	Minor Allele	Alteration	Result
ABCC1	GG+	А		\bullet
BRCA1	AG-	A,C	+ -	\bigcirc
BRCA1	AA-	С		\bigcirc
BRCA1	CC-	А		\bigcirc
BRCA1	GG-	G,T		\bigcirc
BRCA2	Π-	C,G		
BRCA2	AA+	G		
BRCA2	CC+	A,T		
BRCA2	AA+	G		
BRCA2	AA+	Т		
BRCA2	CC+	Т		
CHEK2	TT-	C,G		
CHEK2	GG-	Т		

Name: Sample			
Age:	Gender: F	Report Date:	15/11/2021
Prescriber:	Health Insurance:		

Hereditary Cancer Predisposition Syndrome

Describes a mutation in an inherited gene that increases the risk for one or more types of cancer.

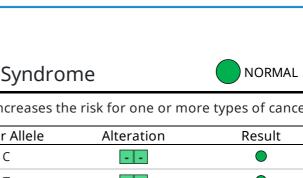
	5			5		
Gene	Genotype	Minor Allele	Alteration	Result		
ATM	Π+	С				
ATM	CC+	Т				
ATM	CC+	A,T		ightarrow		
ATM	Π+	С		\bigcirc		
BRCA1	AG-	A,C	+ -	•		
BRCA1	AA-	С				
BRCA1	CC-	А				
BRCA1	AA-	C,G				
BRCA1	GG-	G,T				
BRCA2	Π-	C,G				
BRCA2	AA+	G				
BRCA2	CC+	A,T				
BRCA2	AA+	G		ightarrow		
BRCA2	AA+	Т				
BRCA2	CC+	Т		ightarrow		
CHEK2	Π-	C,G				
CHEK2	GG-	Т				
ERCC4	AA+	G				
MSH2	GG+	A,T		•		
MSH6	Π+	C,G				
PTEN	Π+	A,G				

Tumor calcinosis

NORMAL

A rare type of extraosseous calcification characterized by large cystic and elastic masses containing calcium phosphate deposits. The condition is more prevalent in periarticular tissue and preserves osteoarticular structures.

Gene	Genotype	Minor Allele	Alteration	Result
KL	CC+	Т		
KL	TT+	A,G		







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	Name: Sample				
•	Age:	Gender: F	Report Date:	15/11/2021	
A	Prescriber:	Health Insurance:			

Myeloproliferative Disorder

FullDN



Also called myeloproliferative neoplasms, it starts in the bone marrow, when stem cells, those that give rise to red blood cells responsible for oxygenation in our body, white blood cells, which fight bacteria and infections and platelets, responsible for blood clotting, proliferate up excessively.

Gene	Genotype	Minor Allele	Alteration	Result
FGFR3	AA+	G		\bigcirc
MPL	GG+	А		
MPL	GG+	A,C		
MPL	CC+	Т		\bigcirc
MPL	TT+	А		\bigcirc
MPL	CC+	Т		

Tumor Necrosis Factor Alpha

Tumor necrosis factor is a cell signaling protein involved in systemic inflammation and is one of the cytokines that make up the acute phase reaction

Gene	Genotype	Minor Allele	Alteration	Result
TNF	GG+	А		

Non-Hodgkin's Lymphoma

Cancer that starts in the lymphatic system, which is a complex network of vessels and small structures called lymph nodes that transport lymph fluid from tissues back to the circulatory system.

Gene	Genotype	Minor Allele	Alteration	Result
CBS	AG+	А	+ -	
ITGB3	Π+	С		
LTA	CC+	Т		ightarrow
PRRC2A	CC-	G		
SELPLG	Π+	A,C	-	
TLR6	TT-	G		
TNF	GG+	А		•





NORMAL

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FullDNA

Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Peripheral T cell lymphoma

NORMAL

Peripheral T-cell lymphoma. This type of lymphoma is rare and develops from more mature forms of T cells.

Gene	Genotype	Minor Allele	Alteration	Result
VCAM1	TT+	С		

Medulloblastoma

It is a cancer of the cerebellum, which grows rapidly, is not very invasive and is more common in children. It originates in the most primitive neurological cells of the medulla of the cerebellum.

Gene	Genotype	Minor Allele	Alteration	Result
BRCA2	CC+	A,G		
NRAS	GG-	A,G,T		
TP53	GG-	A,G,T		
TP53	GG-	A,G,T		
TP53	CC-	A,C		
TP53	GG-	A,T		

Oral Cavity and Laryngeal Neoplasm

Oral cancer, oral cancer or oral cancer is a type of malignant neoplasm of the tissues of the oral cavity. It includes malignant tumors of the lips, gums, tongue, floor of the mouth, hard palate, soft palate (roof of mouth) and oropharynx.

Gene	Genotype	Minor Allele	Alteration	Result
ADH7	CC+	G	-	

Skin Neoplasm (in redheads)

NORMAL

NORMAL

Cancer that affects mainly lighter people, such as redheads.

Gene	Genotype	Minor Allele	Alteration	Result
MC1R	CC+	A,G,T		\bigcirc
MC1R	CC+	т	-	
MC1R	GG+	A,C		\bullet





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ge:	Gender: F	Report Date: 15/11/2021
rescriber:	Health Insurance:	

Li-Fraumeni Syndrome



Gene mutation conducive to cancer development. The TP53 gene mutation weakens the ability to resist cancer, leading to increased susceptibility to multiple neoplasms such as sarcomas, breast cancer, adrenocortical carcinoma, brain tumors and leukemia in the patient.

Gene	Genotype	Minor Allele	Alteration	Result
TP53	GG-	A,G,T		\bullet
TP53	CC-	Т	-	
TP53	GG-	A,G,T		
TP53	GG-	A,T		
TP53	Π-	G		
TP53	GG-	G		
TP53	CC+	Т		
TP53	CC-	A,C		
TP53	GG-	A,T		
TP53	Π-	G		
TP53	GG-	A,G,T		
TP53	GG-	A,T		
TP53	Π-	G		
TP53	AA-	A,C		
TP53	AA-	C,G		

Lynch Syndrome

NORMAL

Also called hereditary colorectal cancer (autosomal dominant transmission) non-polypoid, it is a type of hereditary cancer of the digestive tract, which affects especially the colon and rectum, representing 3% to 5% of cancer cases in these two locations. Endometrial cancer is the extracolonic neoplasm most frequently associated with Lynch II syndrome, with a cumulative risk of 42% at 80 years of age. In addition, the cumulative risk for the development of other extra-colonial tumors, such as cancers of the stomach, ovaries, and biliary and urinary tracts (19%, 9%, 18% and 10%, respectively), is greater than the expected at random. It is estimated that in families with Lynch I syndrome, 40% have mutations in MSH2 and 30% in MLH1 (2). More than 100 mutations have been described in MHS2, predominantly substitutions and small deletions

Gene	Genotype	Minor Allele	Alteration	Result
MSH2	GG+	A,T		
MSH2	GG+	С		\bigcirc
MSH6	TT+	C,G		

	Name: Sample				
-	Age:	Gender: F	Report Date:	15/11/2021	
Α	Prescriber:	Health Insurance:			

Hereditary Tyrosinemia Type 1

This is a rare disease in which the body is unable to completely degrade tyrosine, an amino acid, and as a result harmful substances form, causing serious liver problems and liver cancer.

Gene	Genotype	Minor Allele	Alteration	Result
FAH	CC+			
FAH	AA+			
FAH	GG+		-	
FAH	GG+			
FAH	GG+			

Wilms' Tumor

Malignant embryonic neoplasm arising from the metanephric blastema, which is the most common renal tumor during childhood.

Gene	e Genotype	Minor Allele	Alteration	Result
PAX	5 CC+	Т		
WT1	AA-	С		

Hereditary Nonpolyposis Colorectal Neoplasm

Colorectal cancer (CCR) is the fifth most common type of cancer in Brazil. It is estimated that hereditary CCR represents 10% to 15% of all diagnosed cases. Two hereditary colorectal cancer syndromes have been further investigated: Familial Adenomatous Polyposis (FAP) and Hereditary Non-Polyposis Colorectal Cancer Syndrome (HNPCC). While FAP is a rare syndrome, representing less than 1% of diagnosed RCC cases, the HNPCC syndrome has the highest incidence.

Gene	Genotype	Minor Allele	Alteration	Result
MSH6	TT+	C,G		ightarrow

Multiple Endocrine Neoplasm

It is an inherited syndrome characterized by tumors in the parathyroids, pancreatic islet cells and pituitary

Gene	Genotype	Minor Allele	Alteration	Result
CDKN1B	CC+	С		
CDKN1B	TT+	A,C,G		\bullet
RET	CT+	Т	+ -	\bigcirc
RET	AA+	G		•



NORMAL

NORMAL

NORMAL

NORMAL

\sim	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021	
FullDNA	Prescriber:	Health Insurance:		

Pancreas Neoplasm



The pancreas is a gland located in the upper abdomen, behind the stomach, and is one of the organs that make up the digestive system. It is composed of three parts - head, body and tail - and has two distinct functions: the endocrine function, responsible for the production of insulin (a hormone that controls the blood glucose level) and the exocrine function, responsible for the production of involved enzymes in the digestion and absorption of food. Pancreatic cancer is rare in young people under 30 years of age. The disease affects practically the same proportion of men and women, in general, over the age of 50, especially between 65 and 80 years of age. In most cases, it is not possible to determine the cause of the disease, but the most important risk factor is smoking. The others are: chronic pancreatitis, previous radiotherapy applications, type 2 diabetes mellitus, prolonged exposure to pesticides and chemicals, certain genetic syndromes, and surgeries to treat ulcers or remove the gallbladder.

Gene	Genotype	Minor Allele	Alteration	Result
ATM	CC-	G,T		
CCKBR	CC+	А		
GP2	Π+	С		
GP2	GG+	А		
MTRR	CC+	Т	-	

Malignant Pleural Mesothelioma



Mesothelioma is a type of neoplasm that develops from the cells of the mesothelium - tissue of mesodermal origin that forms the epithelium that externally lines the viscera. The area most commonly affected is the pleura, but it can also occur less frequently in the peritoneum and, more rarely, in the pericardium and tunica vaginalis. Signs and symptoms of mesothelioma can include dyspnoea, ascites, chest pain, cough, fatigue, and weight loss. These symptoms are progressive but may be delayed due to the generally slow course of the disease. Individuals with the CC alleles of the rs4880 polymorphism were at higher risk.

Gene	Genotype	Minor Allele	Alteration	Result
SOD2	Π-	G		

\sim	Name: Sample			
	Age:	Gender: F	Report Date: 15/11/2021	
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Retinoblastoma



Retinoblastoma is a rare type of eye cancer that usually develops in infancy, usually before age 5 years. This form of cancer develops in the retina, which is the specialized light-sensitive tissue at the back of the eye that detects light and color. In children with retinoblastoma, the disease usually affects only one eye. However, one in three children with retinoblastoma develops cancer in both eyes. The most common first sign of retinoblastoma is a visible whiteness of the pupil called "cat's eye reflex" or leukocoria. This unusual whiteness is particularly visible in low light or in photographs taken with a flash. Other signs and symptoms of retinoblastoma include crossed eyes or eyes not pointing in the same direction (strabismus), which can cause strabismus; a change in the color of the colored part of the eye (iris); redness, pain or swelling of the eyelids; blindness or lack of vision in the affected eye(s). Retinoblastoma is often curable when diagnosed early. However, if not treated immediately, this cancer can spread beyond the eye to other parts of the body. This advanced form of retinoblastoma can be fatal. When retinoblastoma is associated with a genetic change (mutation) that occurs in every cell in the body, it is known as hereditary (or germinal) retinoblastoma. People with this form of retinoblastoma often develop cancer in both eyes and are also at increased risk of developing many other types of cancer outside the eye.

Gene	Genotype	Minor Allele	Alteration	Result
RB1	GG-	А		
RB1	GG+	Т		
RB1	TT+	А	-	
RB1	CC+	Т		
RB1	CC+	Т		
RB1	CC+	Т		
RB1	CC+	Т		
RB1	CC+	Т		
RB1	TT+	А		

Myelodysplastic Syndrome



Myelodysplastic syndrome (MDS) refers to a group of neoplasms of bone marrow precursor blood cells, characterized by an increase in the number of immature cells, which can move to the spleen and liver, and a production of inefficient and abnormal blood cells (anemia, leukopenia or thrombocytopenia). There is a 20-30% risk of developing acute myeloid leukemia. All three bone marrow cell lines may be involved (white or granulocytic series, red or erythrocytic series, and platelet or megakaryocytic series).

Gene	Genotype	Minor Allele	Alteration	Result
HRAS	GG-	A,C,T		
TP53	GG-	A,G,T		
TP53	CC-	A,C		

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FullDNA

Name: Sample				
Age:	Gender: F	Report Date:	15/11/2021	
Prescriber:	Health Insurance:			Sample

Lung adenocarcinoma

NORMAL

NORMAL

About 40% of lung cancers are adenocarcinomas. These tumors start in the cells that line the alveoli and produce substances like mucus. This type of lung cancer occurs mainly in smokers and ex-smokers, but it is also the most common type in non-smokers.

Gene	Genotype	Minor Allele	Alteration	Result
PDCD1	CC-	Т		

JAK2 V617F mutation

The JAK2 V617F mutation is an acquired, somatic mutation present in the majority of patients with myeloproliferative cancer (myeloproliferative neoplasms) i.e. nearly 100% of patients with polycythemia vera and in about 50% of patients with essential thrombocytosis and primary myelofibrosis.

Gene	Genotype	Minor Allele	Alteration	Result
JAK2	GG+	A,T		lacksquare

Schwannoma

Schwannoma is a rare type of tumor that forms in the nervous system. Schwannoma grows from cells called Schwann cells. Schwann cells protect and support the nerve cells of the nervous system. Schwannoma tumors are often benign, which means they are not cancer. But, in rare cases, they can become cancer.

Gene	Genotype	Minor Allele	Alteration	Result
SEL1L	Π+	С		

Skin Neoplasm (Melanoma)

Melanoma is the most serious type of skin cancer.

Gene	Genotype	Minor Allele	Alteration	Result
BRCA2	AA+	G		
CDK4	CC-	A,T		
ERCC2	Π+	A,G		\bigcirc
INTERGENIC	CT+	С	+ -	
MC1R	Π+	С		
MC1R	CC+	A,G		
MC1R	CC+	Т		
MC1R	GG+	A,C		
MCR1R	AA+	G		
PIGU	GG+	A,C		ightarrow



LOW

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Endometrial Neoplasm



LOW

UNDEFINED

UNDEFINED

UNDEFINED

Endometrial cancer is a malignant disease in which cells form in the tissues of the endometrium, which is the inside of the uterus, a hollow muscular organ located in a woman's pelvis. Endometrial cancer is the extracolonic neoplasm most frequently associated with Lynch II syndrome, with a cumulative risk of 42% at 80 years of age. In addition, the cumulative risk for the development of other extra-colonial tumors, such as cancers of the stomach, ovaries, and biliary and urinary tracts (19%, 9%, 18% and 10%, respectively), is greater than the expected at random. It is estimated that in families with Lynch I syndrome, 40% have mutations in MSH2 and 30% in MLH1 (2). More than 100 mutations have been described in MHS2, predominantly substitutions and small deletions.

Gene	Genotype	Minor Allele	Alteration	Result
CDKN1B	CC+	С		
COMT	CC+	Т		
ESR1	AG+	G	+ -	\bigcirc
MSH6	Π+	C,G		
MUTYH	GG-	Т		
PTEN	GG+	A,C,T		

Osteosarcoma

A type of bone cancer that starts in the cells that form the Skeletal System (bones).

Gene	Genotype	Minor Allele	Alteration	Result
ERCC2	TT+	A,G		\bigcirc

Ductal Carcinoma (Breast)

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.

Colon Carcinoma

Cancer of the colon or rectum, located at the lower end of the digestive tract.

Tumor Carcinosis

A rare type of extraosseous calcification characterized by large cystic and elastic masses containing calcium phosphate deposits. The condition is more prevalent in periarticular tissue and preserves osteoarticular structures.

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Colorectal Neoplasm (Family)

Cancer of the colon or rectum, located at the lower end of the digestive tract. Most colorectal cancers occur in people without a family history of colorectal cancer. Still, 20% of people who develop the disease have other family members who have been affected by the disease. People with a history of colorectal cancer or adenomatous polyps in one or more first-degree relatives are at increased risk. The risk is doubled in patients with only one affected first-degree relative. This risk is even greater if this relative was diagnosed with less than 45 years of age, or if more than one first-degree relative was affected.

Gallbladder Neoplasm

Cancer that develops in the gallbladder, a small organ below the liver. The signs and symptoms of gallbladder cancer usually only appear when the disease is at an advanced stage, but in some cases they may appear at an earlier stage when treatment may be more effective.

Skin Neoplasm (Melanoma - Hereditary)

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

Tumor predisposition syndrome (BAP1)

Tumor predisposition syndrome that is related to mutations in the BAP1 gene

Gastrointestinal Tumor (GIST)

It is a type of cancer that can develop along the entire digestive tract, that is, from the esophagus, through the stomach, small intestine, large intestine, to the rectum

Biliary Pathway Neoplasm

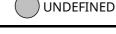
Cholangiocarcinoma, also known as bile duct cancer, is a form of cancer that is formed by mutated epithelial cells (or cells showing characteristics of epithelial differentiation) that originate in the bile ducts that drain bile from the liver into the small intestine.



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Hodgkin's lymphoma

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Lymphoma or Hodgkin's disease is a type of cancer that originates in the lymphatic system, composed of organs (lymph nodes or ganglia) and tissues that produce the cells responsible for immunity and vessels that carry these cells through the body. Hodgkin's lymphoma has the characteristic of spreading in an orderly way, from one group of lymph nodes to another group, through the lymphatic vessels. The disease arises when a lymphocyte (the body's defense cell), most often a type B cell, turns into a malignant cell, capable of multiplying uncontrollably and spreading. The malignant cell begins to produce, in the lymph nodes, identical copies, also called clones. Over time, these malignant cells can spread to nearby tissues, and, if left untreated, can reach other parts of the body. The disease originates more frequently in the neck and chest region called the mediastinum. The disease can occur in any age group; however, it is more common among adolescents and young adults (15 to 29 years), adults (30 to 39 years) and the elderly (75 years or more). Men are more likely to develop Hodgkin's lymphoma than women.

Non-Small Cell Lung Neoplasm

Lung cancer is divided into two main types, which are treated very differently: 80 to 85% of lung cancers are non-small cell lung cancer. 10 to 15% of lung cancers are of the small cell lung cancer type.

Invasive Squamous Cell Carcinoma

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.

BAP1 - Tumor Predisposition Syndrome

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).

Oligodendroglioma

Oligodendroglioma is a tumor that can occur in the brain or spinal cord. Oligodendroglioma forms from oligodendrocytes — cells in the brain and spinal cord that produce a substance that protects nerve cells. Oligodendroglioma can occur at any age, but most often affects adults.



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Craniopharyngioma

A craniopharyngioma is a rare type of brain tumor derived from pituitary gland embryonic tissue that occurs most commonly in children, but also affects adults. It may present at any age, even in the prenatal and neonatal periods, but peak incidence rates are childhood-onset at 5–14 years and adult-onset at 50-74 years.

Adrenocortical Carcinoma

Adrenocortical carcinoma is a rare disease in which malignant (cancer) cells form in the outer layer of the adrenal gland. There are two adrenal glands. The adrenal glands are small and shaped like a triangle. One adrenal gland sits on top of each kidney. Each adrenal gland has two parts.

Digestive system

Bowel polyps

A polyp is a small bulge that grows in mucosa-lined cavities. Polyps can appear in various regions of our body, such as stomach, gallbladder, uterus, nasal cavity, intestines and others. In the case of intestinal polyps, the place where they are most common is in the large intestine (colon). Intestinal polyp is a benign tumor that arises from an abnormal growth of the gut's own mucosal cells. Barely comparing, we can say that they are a kind of colon wart. These injuries are very common, being present in more than 30% of the adult population. Although they are usually benign, a small part of them have the potential to develop into colon cancer over the years. Fortunately, through colonoscopy it is possible not only to diagnose, but also to remove intestinal polyps completely and safely, preventing them from developing into colon cancer.

Gene	Genotype	Minor Allele	Alteration	Result
MUTYH	AA-	С		
MUTYH	GG-	Т		

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Juvenile Polyposis Syndrome



Juvenile Polyposis is a rare condition and belongs to the group of familial hamartomatous polyposis (FHP). It is an autosomal dominant syndrome that can be triggered by mutations in the SMAD4/DPC4 gene (which encodes an intermediate TGF-b signal). It is characterized by the appearance of 10 or more hamartomatous polyps (juvenile) in the gastrointestinal tract, predominantly in the colon. It usually manifests itself between 4 and 14 years of age. Some polyps acquire adenomatous foci despite the hamartomatous nature of the lesions and there is a chance of malignancy.

Gene	Genotype	Minor Allele	Alteration	Result
SMAD4	GG+	А		
SMAD4	GG+	А		
SMAD4	GG+	А		
SMAD4	GG+	А		
SMAD4	CC+	Т		
SMAD4	GG+	А		

Drug Reactions

Risk of Peripheral Neuropathy using Taxane

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Although it does not represent a risk to the patient's life, peripheral neuropathy depreciates the quality of life, compromising the performance of Daily Living Activities (ADLs), as well as the Instrumental Activities of Daily Living (IADLs). In addition, it represents the most common neurological complication of antineoplastic treatment and affects approximately one third of patients undergoing cytostatic drugs, among which taxanes (paclitaxel and docetaxel) and oxaliplatin, frequently used in treatments for breast, ovarian cancer, stand out. , lung and intestine.4 Some factors, such as dosage, administration of antineoplastic agents (total dose, dose per cycle, number of cycles, infusion time), age of the patient, impaired liver and kidney function, previous or concomitant use of other neurotoxic drugs and concurrent radiotherapy treatment of the central nervous system may interfere with the incidence and severity of neurotoxicity. Several studies show that almost all patients who use oxaliplatin have some degree of peripheral neurological dysfunction and, depending on the intensity at which they occur, it is recommended to reduce the dose by 25% to 50% or even to interrupt treatment, with risk progression of cancer. In addition, the fact that chronic neuropathy is observed in 29% -70% of people who use this drug is highlighted. This antineoplastic agent is mainly used in first and second line therapeutic regimens for colon and rectal cancer. Taxanes, in turn, are frequently associated with dosedependent, dose-limiting and cumulative peripheral neuropathy. Paclitaxel is an antineoplastic agent commonly used to treat lung, breast and ovarian cancer. The incidence of peripheral neurotoxicity after its use is 62%, of which 30% of patients present it in grade 2, that is, with some limitation in activities of daily living. Symptoms are characterized by tingling, numbness and pain in the hands and feet; fine motor changes; difficulty walking; loss of deep tendon reflexes; myalgia and transient arthralgia, especially in joints and limbs. Docetaxel, a drug used in the treatment of advanced or metastatic breast cancer, non-small cell lung carcinoma, among others, is also associated with peripheral neuropathy, but in lesser intensity and frequency, with approximately 13% of patients manifesting their signs. and symptoms.

General

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Hepatocellular Carcinoma (HCC)



Hepatocellular carcinoma (HCC) is the most common type of primary liver cancer in adults and is currently the most common cause of death in people with cirrhosis. HCC is the third leading cause of cancer-related death worldwide. It occurs in the setting of chronic liver inflammation, and is most closely linked to chronic viral hepatitis infection (hepatitis B or C) or exposure to toxins such as alcohol, aflatoxin, or pyrrolizidine alkaloids. Certain diseases, such as hemochromatosis and alpha 1-antitrypsin deficiency, markedly increase the risk of developing HCC. Metabolic syndrome and NASH are also increasingly recognized as risk factors for HCC.

Gene	Genotype	Minor Allele	Alteration	Result
MIR146A	GG+	G		
MIR196A2	Π+	С		\bigcirc
PNPLA3	CG-	G	+ -	•
TP53	CC-	Т		

Genes

GSTT1				NORMAL
GSTT1 gene plays species(ROS). A nul	s an important ro I variant in this gene	e in detoxification has been demonstra	and clearance of a clearance of a clear and clear and a clear and a clear a cl	of reactive oxygen er susceptibility.
Gene	Genotype	Minor Allele	Alteration	Result
GSTT1	Π-	G		

Genetic diseases

Proteus Syndrome

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	Genotype	Minor Allele	Alteration	Result
AKT1	Π+	С		



NORMAL

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Hereditary Breast and Ovary Cancer Syndrome

NORMAL

Cancer that forms in the cells of the breasts and ovaries.

Gene	Genotype	Minor Allele	Alteration	Result
BRCA1	AG-	A,C	+ -	•
BRCA1	AA-	С		
BRCA1	CC-	А		
BRCA1	AA-	C,G		
BRCA1	GG-	G,T		
BRCA2	Π-	C,G		
BRCA2	AA+	G		
BRCA2	CC+	A,T		
BRCA2	AA+	G		
BRCA2	AA+	Т		
BRCA2	CC+	Т		

Rasopathies

NORMAL

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The rasopathies that encompass Noonan Syndrome and syndromes related to it (Legius, for example), are monogenic diseases caused by mutations in genes belonging to the same signaling pathway (RAS-MAPK) important for growth, immune system, growth and differentiation of cells. Short stature is a cardinal sign and also difficulty feeding in the first months of life.

Gene	Genotype	Minor Allele	Alteration	Result
BRAF	GG-	G		

Bloom Syndrome

It is a rare inherited disease that is found more frequently in Ashkenazi Jews than in other populations. A feature of Bloom's syndrome is a high risk of cancer. Bloom's syndrome is sometimes considered a disease of premature or accelerated aging. Babies with this disease are born small and remain shorter than normal as they grow. Their skin may look red, and they have more lung and ear infections than children normally have.

Peutz-Jeghers syndrome

Peutz-Jeghers' syndrome (SPJ), also called hereditary intestinal polyposis syndrome (SPHI) is a genetic disease characterized by the development of hamartomous polyps in the digestive system and dark spots on the lips and in the mouth mucosa. It is a rare, autosomal dominant genetic disease, probably due to a mutation of the STK11 (LKB1) gene on Chromosome 19 (human), a gene that suppresses the appearance of cancer. Therefore, the risk of developing internal cancers is often greater in these patients.

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Hematologic system

Idiopathic Hypereosinophilic Syndrome



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PDGFRA-associated chronic eosinophilic leukemia is a form of blood cell cancer characterized by an elevated number of cells called eosinophils in the blood. These cells help fight infections by certain parasites and are involved in the inflammation associated with allergic reactions. However, these circumstances do not account for the increased number of eosinophils in PDGFRA-associated chronic eosinophilic leukemia.Another characteristic feature of PDGFRA-associated chronic eosinophilic leukemia is organ damage caused by the excess eosinophils. Eosinophils release substances to aid in the immune response, but the release of excessive amounts of these substances causes damage to one or more organs, most commonly the heart, skin, lungs, or nervous system. Eosinophil-associated organ damage can lead to a heart condition known as eosinophilic endomyocardial disease, skin rashes, coughing, difficulty breathing, swelling (edema) in the lower limbs, confusion, changes in behavior, or impaired movement or sensations. People with PDGFRA-associated chronic eosinophilic leukemia can also have an enlarged spleen (splenomegaly) and elevated levels of certain chemicals called vitamin B12 and tryptase in the blood.Some people with PDGFRA-associated chronic eosinophilic leukemia have an increased number of other types of white blood cells, such as neutrophils or mast cells. Occasionally, people with PDGFRA-associated chronic eosinophilic leukemia develop other blood cell cancers, such as acute myeloid leukemia or B-cell or T-cell acute lymphoblastic leukemia or lymphoblastic lymphoma.PDGFRA-associated chronic eosinophilic leukemia is often grouped with a related condition called hypereosinophilic syndrome.

Gene	Genotype	Minor Allele	Alteration	Result
PDGFRA	Π+	С		

Hormone

Adiponectin Flag

Adiponectin is a protein hormone that modulates several metabolic processes, including blood glucose regulation and fatty acid catabolism. It is exclusively secreted from adipose tissue into the bloodstream and its levels in blood plasma are inversely related to the percentage of body fat in adults, while this association is not well defined in children. This hormone plays a role in suppressing metabolic events that can cause Type 2 Diabetes, Obesity, Atherosclerosis, Non-Alcoholic Fatty Liver Disease, and Metabolic Syndrome. Adiponectin is secreted into the bloodstream, where it represents about 0.01% of all plasma proteins. There is a sexual dimorphism in their plasma concentrations, with women having higher levels than men. Adiponectin levels are reduced in diabetics compared to non-diabetics. Loss of body weight significantly increases the concentration of this hormone in plasma. This hormone exerts part of its weight loss effects on the brain. This action is similar to that of leptin, but the two hormones have complementary actions, and may have additive effects. Green result indicates low flag, orange result indicates intermediate flag and red result indicates high flag. Low adiponectin levels are associated with systemic organ failure in acute pancreatitis.

Immune system

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Neurofibromatosis

NORMAL

Neurofibromatosis (NF1 and NF2) is an autosomal dominant inherited disease.

Gene	Genotype	Minor Allele	Alteration	Result
NF1	CC+	Т		
NF1	CC+	G		
NF1	GG+	A,T		ightarrow
NF1	GG+	А		
NF2	AA+	С		lacksquare
NF2	CC+	Т		
NF2	CC+	Т		ightarrow
NF2	CC+	G,T		\bigcirc
NF2	GG+	Т		
NF2	TT+	С		ightarrow

Neurodegenerative diseases

Louis-Bar Syndrome (Ataxia Telangiectasia)

NORMAL

NORMAL

It is a rare, neurodegenerative and hereditary disease that causes severe disability.

Gene	Genotype	Minor Allele	Alteration	Result
ATM	TT+	С		
ATM	CC+	Т		
ATM	CC+	A,T		
ATM	TT+	С		

Oncologic

Cervical Cancer

Cervical cancer is a type of cancer that occurs in the cells of the cervix — the lower part of the uterus that connects to the vagina.

Gene	Genotype	Minor Allele	Alteration	Result
HRAS	GG-	A,C,T		
TP53	GG-	A,T		



BRAF V600E mutation



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NORMAL

NORMAL

NORMAL

A specific mutation (change) in the BRAF gene, which makes a protein that is involved in sending signals in cells and in cell growth. This BRAF gene mutation may be found in some types of cancer, including melanoma and colorectal cancer. It may increase the growth and spread of cancer cells. Checking for this BRAF mutation in tumor tissue may help to plan cancer treatment.

Gene	Genotype	Minor Allele	Alteration	Result
IRF4	CC+	Т		

Choroidal Melanoma

Choroidal melanoma is the cancer that most affects the eye of adults. It is also called ocular melanoma or uveal melanoma. The annual incidence of ocular melanoma is about 10 cases per million inhabitants.

Reactions to Treatments

Glucocorticoid Resistance

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	Genotype	Minor Allele	Alteration	Result
NR3C1	GG-	G,T		ightarrow

Response to Fluoroucacil

It is an antitumor agent widely used in the treatment of several types of cancers.

Gene	Genotype	Minor Allele	Alteration	Result
DPYD	GG-	G,T		

Glucocorticoid Therapy

Glucocorticoids are a class of steroid hormones characterized by their ability to bind to the cortisol receptor and trigger similar effects.

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





Sample

Allogeneic Rejection



NORMAL

NORMAL

In the allogeneic transplant technique, the patient's own stem cells are used, which are treated with high doses of radiation or chemotherapy to ensure that there are no cancer cells.

Gene	Genotype	Minor Allele	Alteration	Result
TNF	GG+	А		

Effect of Aspirin to reduce risk of colorectal neoplasia

Effect of Aspirin to reduce risk of colorectal neoplasia

Gene	Genotype	Minor Allele	Alteration	Result
CASC8	GG+	Т		lacksquare

Best Effectiveness of Chemotherapy

Although the effectiveness of chemotherapy can be affected by many factors, genetic variation such as polymorphism plays a significant role in drug response. The location or site of the polymorphism will determine the effect, as the polymorphism present within a coding sequence and leading to an amino acid change (referred to as a non-synonymous SNP or mutation) can modify the protein's activity or function. If the mutation is synonymous, translation rates or mRNA half-life may be affected. If the mutation causes a premature stop codon, it can lead to the production of a truncated protein product or the nonsense mediated decay phenotype. Therefore, due to differences or changes at the genetic level, causing changes in cell phenotype could explain part of the variability in response or toxicity.

Gene	Genotype	Minor Allele	Alteration	Result
TP53	GG-	C,T		\bigcirc



Age:





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