











Genetics for people

# »» Genetic Testing Healthy Ageing

A photograph of a family (mother, father, and child) running on a beach at sunset. The child is in the foreground, wearing a striped shirt and a cap. The father is in the middle, wearing a white tank top and shorts. The mother is in the background, wearing a white t-shirt and dark pants. They are all running towards the right, with the ocean and sunset in the background.

# My Ageing

## SUMMARY OF RESULTS

Process	Risk	
METABOLIC SYNDROME	LOW RISK	
VISCERAL FAT	MODERATE RISK	
HEPATIC DETOXIFICATION	LOW RISK	
OXIDATIVE STRESS	MODERATE RISK	
DOPAMINE	LOW RISK	
SEROTONINE	LOW RISK	
GABA	MODERATE RISK	
INFLAMMATION	LOW RISK	
TELOMERIC SHORTENING	LOW RISK	
NEOPLASMS	LOW RISK	

## SUMMARY OF RECOMMENDATIONS

Based on your genetic results, you are predisposed to develop the disorder. The most recommended supplements, vitamins and drugs, as well as their dosage, are listed below. Remember, this report indicates the genetic predisposition and does not necessarily mean that you will develop it. Before starting a therapeutic regimen, we recommend that you consult your specialist doctor to confirm the diagnosis.

GABA		
Supplementation	Drugs	Otherss
Theanine (50-150 mg) Phenibut (50-150 mg) Passionflower (80-125 mg)	Anxiolytics Anticonvulsants	–

OXIDATIVE STRESS		
Supplementation	Drugs	Otherss
Vitamin E (50-100 iu) Vitamin C (250-500 mg) Chelated Selenium (25-50 mcg) Chelated Zinc (8-16 mg) Copper (1-3 mg) Manganese (0.5-1 mg) Pycnogenol (25-50 mg) Alpha-lipoic Acid (50-100 mg) Vitamin B12 (50-150 mg) Cysteine (200-600 mg)	–	–

Based on the genes you have with slow or intermediate enzyme activity, the following groups of drugs are indicated that are metabolised by these genes and may present a problem in the metablylation of these drugs. Remember, this report indicates genetic predisposition and does not necessarily mean that you will develop it. Before starting a treatment regimen, we recommend that you consult your specialist doctor before making any changes to your treatment.

Antidepressant	
Gene	Drug(s)
CYP2D6	Amitriptyline, Citalopram, Clomipramine, Desipramine, Fluoxetine, Fluvoxamine, Imipramine, Nefazodone, Nortriptyline, Paroxetine, Protriptyline, Timipramine, Venlafaxine, Vortioxetine

Antiarrhythmic	
Gene	Drug(s)
CYP2D6	Propafenone, Quinine
NAT2	Isosorbide

Antibiotic	
Gene	Drug(s)
NAT2	Isoniazid, Pyrazinamide, Rifampicin

Antineoplastic	
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Gene	Drug(s)
UGT1A1	Irinotecan, Nilotinib, Pazopanib
CYP2D6	Cabazitaxel, Sunitinib, Sirolimus, Ruxolitinib
MTHFR	Ethinyl Estradiol
Attention Deficit	
Gene	Drug(s)
CYP2D6	Atomoxetine
Analgesic	
Gene	Drug(s)
CYP2D6	Codeine, Tramadol
CYP2C9	Celecoxib
Anticoagulant	
Gene	Drug(s)
CYP2C9	Prasugrel, Warfarin
CYP3A5	Prasugrel
NAT2	Clopidogrel
Anxiolytic	
Gene	Drug(s)
CYP2D6	Doxepin
Antimalarial	
Gene	Drug(s)
CYP2D6	Quinine sulphate
Contraceptive	
Gene	Drug(s)
MTHFR	Norelgestromin
Narcolepsy Treatment	
Gene	Drug(s)
CYP2D6	Modafinil
Gaucher Disease	
Gene	Drug(s)
CYP2D6	Eliglustat
Antihypertensive	
Gene	Drug(s)
CYP2D6	Cavedilol, Metoprolol, Propranolol
Antiinflammatory	
Gene	Drug(s)

CYP2C9	Flurbiprofen
<b>Antipsychotic</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Aripiprazole, Iloperidone, Clozapine, Perphenazine, Pimozide, Risperidone, Thioridazine
<b>Muscle Relaxant</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Tolteridone
NAT2	Hydralazine
<b>Antitussive</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Dextromethorphan
<b>Bronchodilator</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Aformoterol
UGT1A1	Aformoterol, Indacaterol
<b>Korea Treatment</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Tetrabenazine
<b>Dry Mouth Treatment</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Cevimeline
<b>Amino Acid Derivatives</b>	
<b>Gene</b>	<b>Drug(s)</b>
CYP2D6	Cevimeline

Based on the genes with slow or intermediate enzyme activity, actions are indicated that can promote their correct functioning.

- **NOS3:** Assess RCV.
- **CYP3A5:** Avoid black and green tea. Use rooibos as an alternative.
- **CYP4F2:** Include coffee in your diet, as well as supplements rich in caffeic acid.
- **GSTP1:** Avoid garlic and quercetin. Include cruciferous vegetables, green tea, soya, citrus fruits, turmeric, berries and rooibos tea in your diet, as well as foods rich in resveratrol, such as red grapes and red grape products.
- **GSTM1:** Avoid garlic and quercetin. Include cruciferous vegetables, green tea, soya, citrus fruits, turmeric, berries and rooibos tea in your diet, as well as foods rich in resveratrol, such as red grapes and red grape products.
- **GSTT1:** Avoid garlic and quercetin. Include cruciferous vegetables, green tea, soya, citrus fruits, turmeric, berries and rooibos tea in your diet, as well as foods rich in resveratrol, such as red grapes and red grape products.
- **MTHFR:** Discuss homocysteine analysis with a healthcare professional and include methylfolate in your diet.
- **NAT2:** Avoid caffeine, cocoa, tea and foods containing high levels of vitamin A.
- **UGT1A1:** Include cruciferous vegetables, citrus fruits, dandelion, rooibos tea, hoeny-bush tea, rosemary, soya and turmeric in your diet. Foods rich in resveratrol (red grapes and derivatives), antaxanthin (salmon, shrimp and other marine organisms), ellagic acid (red fruits such as blueberries, cherries and pomegranates) and ferulic acid (found in vegetables, legumes, cereals, fruits and seeds) are recommended.

# DETAILED RESULTS

## 1. METABOLIC SYNDROM

LOW RISK



### INFORMATION ABOUT METABOLIC SYNDROM

Metabolic syndrome is a group of conditions that increase the risk of heart disease, stroke and type 2 diabetes. These conditions include high blood pressure, high blood sugar levels, excess body fat around the waist, and abnormal cholesterol or triglyceride levels. Risk factors for metabolic syndrome include a combination of genetics, sedentary lifestyle and an unhealthy diet. Early identification and management of metabolic syndrome is crucial to prevent serious health complications.

### GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
ADBR2	rs1042713	GG	GG
ADBR2	rs1042714	GG	GG
ADBR3	rs4994	AA	AA
APOB	rs693	GG	GA
APOB	rs1367117	GG	GG
APOB	rs5742904	CC	CC
FTO	rs9939609	TT	TA
FTO	rs8050136	CC	CA
FTO	rs1421085	TT	TC
FTO	rs3751812	GG	GT
LEPR	rs1137100	AA	AG
MC4R	rs17782313	TT	TT
MC4R	rs12970134	GG	GG
MC4R	rs2229616	TT	CC
MC4R	rs10871777	AA	AA
NOS3	rs1799983	TT	GG
NOS3	rs2070744	CC	TT

## 2. VISCERAL FAT

MODERATE RISK



### INFORMATION ABOUT VISCERAL FAT

Visceral fat refers to the accumulation of fat around the internal organs of the abdomen. This type of fat is particularly detrimental to health, as it is associated with an increased risk of cardiovascular disease, type 2 diabetes and other metabolic complications. The accumulation of visceral fat can be influenced by genetic factors, poor eating habits, lack of exercise and other lifestyle factors. Reducing visceral fat is important to improve overall health and prevent chronic diseases.

### GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
FTO	rs9939609	TT	TA
FTO	rs8050136	CC	CA
FTO	rs1421085	TT	TC
FTO	rs3751812	GG	GT



### 3. HEPATIC DETOXIFICATION

LOW RISK



#### INFORMATION ABOUT HEPATIC DETOXIFICATION

Liver detoxification refers to the process by which the liver breaks down and removes toxins from the body. The liver is central to detoxification, as it metabolises harmful substances and converts them into less harmful compounds that can be excreted. Factors such as diet, alcohol consumption, exposure to environmental pollutants and genetics can affect the liver's ability to perform this process effectively.

#### GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
EHPX1	rs2234922	AA	AA
EHPX1	rs1051740	TT	TT
CBS	rs234706	AA	GG
CBS	rs5742905	AA	AA

Gene	Reference Haplotype	Patient Haplotype	Metaboliser
CFTR	*1/*1	*1/*1	NORMAL
CYP2A6	*1/*1	*1/*1	NORMAL
CYP2B6	*1/*1	*1/*1	NORMAL
CYP2C19	*38/*38	*1/*1	NORMAL
CYP2C8	*1/*1	*1/*3	INTERMEDIATE
CYP2C9	*1/*1	*1/*2	INTERMEDIATE
CYP2D6	*1/*1	*1/*10	INTERMEDIATE
CYP3A4	*1/*1	*1/*1	NORMAL
CYP3A5	*1/*1	*3/*3	SLOW
CYP4F2	*1/*1	*4/*6	INTERMEDIATE
DPYD	*1/*1	*1/*1	NORMAL
GSTP1	*1/*1	*1A/*1B	INTERMEDIATE
MTHFR	*1/*1	*1/*2	INTERMEDIATE
NAT2	*4/*4	*5B/*5B	SLOW
NQO1	*1/*1	Activo/Activo	NORMAL
SLCO1B1	*1/*1	*1/*1	NORMAL
TPMT	*1/*1	*1/*1	NORMAL
UGT1A1	*1/*1	*1/*28	INTERMEDIATE

## 4. OXIDATIVE STRESS

MODERATE RISK



### INFORMATION ABOUT OXIDATIVE STRESS

Oxidative stress occurs when there is an imbalance between the free radicals in the body and the antioxidants available to combat them, known as **antioxidant capacity**. Antioxidants are substances that can prevent or delay some types of cell damage. This process triggers an inflammatory response and can lead to associated health problems, such as cardiovascular problems, premature ageing, among others, and a high antioxidant capacity is essential to combat them. This antioxidant capacity and oxidative stress can be modulated by a wide variety of environmental agents such as diet, physical exercise, unhealthy habits such as smoking and alcohol, and also has an important genetic component.

### GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
AOC1	rs2268999	AA	AT
AOC1	rs10156191	CC	CT
AOC1	rs1049742	CC	CC
APOA2	rs5082	GG	AA
CAT	rs1001179	CC	CT
CAT	rs524154	AA	AG
FABP2	rs1799883	TT	TC
PLIN1	rs2289487	CC	CT
PLIN1	rs1052700	AA	AT
PPARg	rs1801282	CC	CC
SOD1	rs121912442	CC	CC
SOD1	rs121912443	AA	AA
SOD2	rs4880	AA	AA
EHPX1	rs2234922	AA	AA
EHPX1	rs1051740	TT	TT
CBS	rs234706	AA	GG
CBS	rs5742905	AA	AA
IL-6	rs1800795	CC	GG
TNF-ALFA	rs1800629	GG	GG
TNF-ALFA	rs361525	GG	GG
HFE	rs1799945	CC	CG
HFE	rs1800562	GG	GG

Gene	Reference Haplotype	Patient Haplotype	Metaboliser
DPYD	*1/*1	*1/*1	NORMAL
GSTM1	*1/*1	*1/*D	INTERMEDIATE
GSTP1	*1A/*1A	*1A/*1B	INTERMEDIATE
GSTT1	*1/*1	*D/*D	SLOW
MTHFR	*1/*1	*1/*2	INTERMEDIATE
NQO1	*1/*1	Activo/Activo	NORMAL

## 5. DOPAMINE

LOW RISK



### INFORMATION ABOUT DOPAMINE

Dopamine is a neurotransmitter, a chemical found in the brain and central nervous system that plays a key role in communication between nerve cells or neurons. Dopamine plays several roles in the brain and body, being involved in a wide variety of processes, including movement control, mood regulation, motivation, pleasure and decision-making, so alterations in its production can affect mood.

### GENETIC RESULTS

Process	Gene	SNP	Reference Genotype	Patient Genotype
Conversion	DBH	rs1108580	GG	AG
Conversion	DBH	rs1611115	CC	CC
Degradation	COMT	rs4680	GG	GA
Degradation	COMT	rs4633	CC	CT
Degradation	COMT	rs769224	GG	GG
Degradation	MAOB	rs1799836	TT	TC
Synthesis	MIR4686	rs10770141	GG	GG
Synthesis	DDC	rs921451	TT	TC
Synthesis	DDC	rs12718541	AA	AG
Receptors	DRD1	rs4532	CC	TT
Receptors	DRD1	rs5326	CC	CC
Receptors	DRD2	rs1800497	GG	GA
Receptors	DRD2	rs6275	AA	GG
Receptors	DRD3	rs167771	GG	GA
Receptors	DRD3	rs6280	TT	CT
Receptors	DRD4	rs3758653	TT	TC
Receptors	DRD4	rs1800955	TT	TT
Transporters	SLC6A3	rs393795	GG	GG
Liberation	OPRM1	rs1799971	AA	AA

## 6. SEROTONINE

LOW RISK



### INFORMATION ABOUT SEROTONINE

Serotonin is a neurotransmitter that generally acts as an inhibitor in the central nervous system. The precursor of its synthesis is tryptophan, which is transported by the blood to the brain where it is taken up by nerve terminals and converted into 5-hydroxytryptophan (5-HTP) by the enzyme tryptophan hydroxylase. Serotonin is involved in learning, memory, happiness and sleep. Low levels of serotonin have been linked to depression, anxiety, sleep problems and even gastrointestinal conditions.

### GENETIC RESULTS

Process	Gene	SNP	Reference Genotype	Patient Genotype
Synthesis	TPH2	rs120074175	GG	
Synthesis	TPH2	rs120074176	CC	
Synthesis	TPH2	rs17110563	CC	
Synthesis	DDC	rs137853211	GG	
Synthesis	DDC	rs137853212	CC	
Synthesis	DDC	rs921451	TT	TC
Synthesis	DDC	rs12718541	AA	AG
Degradation	MAOA	rs72554632	AA	
Degradation	MAOA	rs587777457	AA	
Degradation	MAOA	rs12718541	AA	AG
Receptors	HTR1A	rs6295	GG	CG
Receptors	HTR1E	rs1406946	CC	CT
Receptors	HTR3A	rs1062613	TT	TC
Receptors	HRT7	rs7905446	TT	TT
Transporters	SLC6A4	rs1042173	AA	AC
Transporters	SLC6A4	rs25531	TT	

## 7. GABA

MODERATE RISK



### INFORMATION ABOUT GABA

Gamma-aminobutyric acid (GABA) is an inhibitory neurotransmitter of the central nervous system. It is synthesised from glutamic acid, another excitatory neurotransmitter in the brain. The enzyme glutamate decarboxylase (GAD) converts glutamate to GABA, which will be detected by GABA-A and GABA-B receptors on nerve cells, thereby reducing the excitability of the cell. This pathway is involved in the regulation of muscle tone, inhibition of neuronal excitability and regulation of mood and anxiety, so that stimulation of the GABAergic system causes a state of sedation, amnesia or even ataxia, while inhibition leads to sleep problems or anxiety.

### GENETIC RESULTS

Process	Gene	SNP	Reference Genotype	Patient Genotype
Synthesis	GAD1	rs769407	GG	GG
Synthesis	GAD1	rs12185692	CC	AA
Receptors	GABRA2	rs279858	TT	CC
Degradation	ABAT	rs121434578	CC	
Degradation	ABAT	rs724159992	GG	
Degradation	ABAT	rs724159991	CC	
Degradation	ALDH5A1	rs118203982	GG	
Degradation	ALDH5A1	rs118203984	CC	

## 8. INFLAMMATION

LOW RISK



### INFORMATION ABOUT INFLAMMATION

Inflammation is the immune system's response to infection, injury or irritants. While acute inflammation is a necessary part of the healing process, chronic inflammation can contribute to a number of diseases, including heart disease, arthritis and certain types of cancer. Factors such as diet, stress, recurrent infections and genetics can influence the propensity for chronic inflammation. Proper management of inflammation is crucial to maintaining long-term health.

### GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
AOC1	rs2268999	AA	AT
AOC1	rs10156191	CC	CT
AOC1	rs1049742	CC	CC
AOC1	rs2052129	GG	TT
HNMT	rs11558538	CC	CT
CAT	rs1001179	CC	CT
IL1A	rs17561	CC	CC
IL1A	rs1800587	GG	GG
IL1B	rs16944	AA	AG
IL1B	rs1143634	GG	GG
IL1RN	rs419598	CC	TT
IL-6	rs1800795	CC	GG
IL-10	rs1800896	CC	TC
CD33	rs3865444	CC	CA
MS4A	rs744373	AA	AA
TREM2	rs143332484	CC	CC
CRP	rs1205	TT	CT
NOS3	rs1799983	TT	GG
NQO1	rs1131341	GG	GG
NQO1	rs1800566	GG	GG
TNF-ALPHA 1	rs1800629	GG	GG
TNF-ALPHA 2	rs361525	GG	GG

# 9. TELOMERIC SHORTENING

LOW RISK



## INFORMATION ABOUT TELOMERIC SHORTENING

Telomeric shortening is a biological process that occurs as cells divide and age. Telomeres are protective structures at the ends of chromosomes that shorten with each cell division. When telomeres become too short, cells lose the ability to divide and function properly, contributing to ageing and the development of age-related diseases. Genetic factors, lifestyle and environment can influence the rate of telomere shortening.

## GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
BHMT	rs651852	CC	CT
BHMT	rs617219	AA	AC
BHMT	rs567754	CC	CT
BHMT	rs585800	TT	TA
MTR	rs1805087	AA	AA
MTRR	rs1802059	GG	GG
MTRR	rs162036	AA	AA
MTRR	rs10380	CC	CC
MTRR	rs1801394	AA	AG
TERT	rs2736100	AA	CC
ZBTB46	rs755017	AA	AA
MIR4454	rs7675998	GG	GG

# 10. NEOPLASMS

LOW RISK



## INFORMATION ABOUT NEOPLASMS

Neoplasms, or tumours, are abnormal growths of tissue that can be benign or malignant. Malignant tumours, or cancers, are particularly dangerous because of their ability to invade nearby tissues and spread to other parts of the body. The development of neoplasms is influenced by a combination of genetic factors, exposure to carcinogens, viral infections and other environmental factors. Early detection and appropriate treatment of neoplasms are critical to improving health outcomes.

## GENETIC RESULTS

Gene	SNP	Reference Genotype	Patient Genotype
ATM	rs3092856	CC	CC
BCMO1	rs12934922	AA	TT
CYP1A1	rs1048943	TT	TT
CYP3A4	rs2740574	TT	TT
EHPX1	rs2234922	AA	AA
EHPX1	rs1051740	TT	TT
GSTP1	rs1695	AA	AG
MTHFR	rs1801133	GG	GA
MTHFR	rs1801131	TT	TT
SOD2	rs4880	AA	AA
TERT	rs2736100	CC	CC



## TECHNOLOGY

**DNA Microarray** technology consists of a solid surface with microscopic reactions (microreactions) or DNA chip, on which molecular probes are attached to detect the presence of target DNA molecules. Probe-target hybridization is usually detected and quantified by measuring the intensity of a given fluorescence provided by the molecular probe in samples. This type of technology allows the detection of thousands of specific DNA fragments present in a DNA sample. On the other hand, the specificity in terms of DNA sequence recognition is very high since single nucleotide exchange (single-base resolution) can be detected using short oligonucleotide probes (20-25 nucleotides). As a result, DNA Microarray technology has also evolved to be applied as a DNA sequencing technique to genotype several hundred thousand single nucleotide variants (SNVs) in target genes located throughout the genome (Whole Genome DNA Microarray).

*Bead Chip Infinium Global Screening Array Orion (GSA Orion)* is a line of DNA chips developed by Illumina for its DNA Microarray iScan platform, widely used in population genetic studies and precision medicine, providing optimized content with 100 % reliable and reproducible high-quality genotyping results. The construction of the GSA Chip was carried out in collaboration with a consortium of experts, and for the selection of SNVs, information from prestigious scientific databases such as gnomAD, NHGRI-EBI-GWAS Catalog, ClinVar, MHC-HLA-KIR and PharmGKB has been used. The GSA allows the analysis of approximately 700,000 SNVs that cover variants of interest (hot spots) throughout the entire genome, impacting a wide range of genetic traits with physiological and pathophysiological implications. In addition, it allows the customization by users to incorporate Ad Hoc 50,000-100,000 variants of interest.

## QUALITY

The analysis laboratory has standard and effective procedures to protect against technical and operational problems. However, results can be altered due to problems with sample collection (contamination) and labelling (identification), delay in receiving the sample in the laboratory (integrity), among other problems. This could lead to invalidation of the test results. In such cases, the patient would be asked to repeat the entire procedure to perform the test.

As with all clinical analytical tests, there is a small chance that the laboratory may report inaccurate information. If there is a suspicion of an error in the genotype detected, further verification testing may be requested.

