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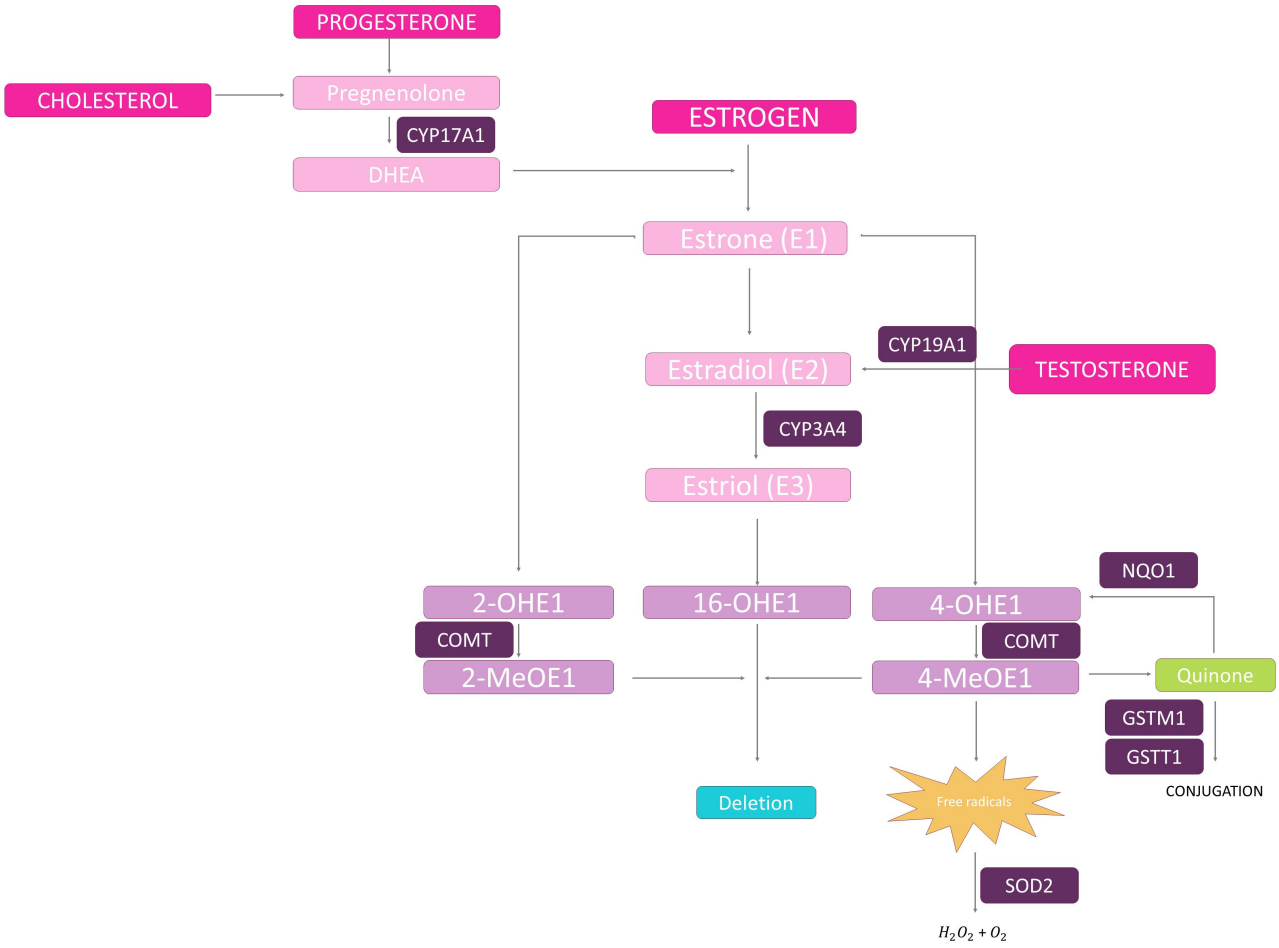
Genetics for people

Genetic Testing
Estrogens

My *Estrogens*

The MyEstrogen genetic test analyses the genes primarily involved in the metabolism of oestrogen, as well as other sex hormones such as testosterone and progesterone, and how variations in these genes influence the body's ability to metabolise and eliminate these hormones. In addition, the ability to process and neutralise toxic compounds from these processes is assessed. This analysis is of great interest for the prevention of diseases and conditions related to hormonal imbalance, so knowing the underlying genetics of these biological processes, , medical care can be adopted and personalised more effectively and provide a more accurate picture of the patient's risk of developing these diseases.



SUMMARY OF RESULTS

The following table shows the variants detected in the patient. In the case of haplotypes, the type of metabolisation based on the enzymatic activity of the haplotype is indicated:

Gene	Reference haplotype	Patient haplotype	Metaboliser
CYP17A1	TT	TT	–
CYP19A1	*1/*1	*1/*1	NORMAL
CYP1A1	*1/*1	*2/*2	NORMAL
CYP1B1	*1/*1	*2/*2	SLOW
CYP3A4	*1/*1	*1/*1	NORMAL
COMT	*1/*1	*1/*2	INTERMEDIATE
NQO1	Activo/Activo	Inactive/Inacti-ve	SLOW
SULT1A1	*1/*1	*2/*2	SLOW
GSTM1	*I/*I	*I/*I	NORMAL
GSTT1	*I/*I	*D/*D	SLOW
SOD2	*1/*1	*1/*1	NORMAL

RECOMMENDATIONS:

Gene	Food or bioactive compounds	
	Inhibitors	Inductors
GSTM1 GSTT1	Cruciferous and allium vegetables Citrus, Ghee, Sweet potato, Roseamary, Garlic, Curcumin, Oil fish, Resveratrol Soy bean, Ellagic acid, Green tea, Rooibos, honeybush	Apiaceous vegetables Quercetin Genistein
CYP3A4	Curcumin	Resveratrol, Myricetin, Soya bean, Kale, Grapefruit, Garden cress
SULT1A1		**Avoid** caffeine, cocoa, tea, high levels of vitamin A
COMT		**Avoid** green tea Resveratrol, Vitamin B6 Stress management

ANNEX I: DETAILS OF GENES ANALYSED

CYP17A1 rs2486758	
This gene encodes a member of the cytochrome P450 enzyme family that catalyses the final steps of oestrogen biosynthesis. Mutations in this gene may result in increased or decreased activity of aromatase, the enzyme responsible for catalysing the conversion of androgens to oestrogens. The TT genotype is associated with **normal enzyme activity**.	Patient genotype: **TT**
CYP19A1	
This gene encodes a member of the cytochrome P450 enzyme family that catalyses the final steps of oestrogen biosynthesis. Mutations in this gene may result in increased or decreased activity of aromatase, the enzyme responsible for catalysing the conversion of androgens to oestrogens. According to his haplotype the patient is a CYP19A1 **normal metaboliser**.	Patient haplotype: ***1/*1**
CYP1A1	
This gene encodes a member of the cytochrome P450 enzyme family and its expression is induced by polycyclic aromatic hydrocarbons, which are present in toxic substances such as tobacco smoke and which have a procarcinogenic effect on the body. It is also involved in the oxidative metabolism of oestrogens, playing an important role in the development of breast and prostate cancer. According to his haplotype the patient is a CYP1A1 **normal metaboliser**.	Patient haplotype: ***2/*2**
CYP1B1	
This gene encodes a member of the cytochrome P450 enzyme family that metabolises procarcinogens such as polycyclic aromatic hydrocarbons. It is also involved in the oxidative metabolism of oestrogens, playing an important role in the development of breast and prostate cancer. According to his haplotype the patient is a CYP1B1 **slow metaboliser**.	Patient haplotype: ***2/*2**

CYP3A4	
This gene encodes a member of the cytochrome P450 enzyme family and its expression is induced by glucocorticoids and certain drugs. It is involved in the metabolism of commonly used drugs such as paracetamol, codeine or diazepam, as well as steroids and carcinogens. According to his haplotype the patient is a CYP3A4 **normal metaboliser**.	Patient haplotype: ***1/*1**
COMT	
Catechol-O-methyltransferase is an enzyme encoded by the COMT gene that catalyses the transfer of a methyl group from S-adenosylmethionine to catecholamines, allowing the control of certain hormone levels. Accumulation of metabolites from the oestrogen pathway increases the risk of developing breast cancer. According to his haplotype the patient is a COMT **intermediate metaboliser**.	Patient haplotype: ***1/*2**
NQO1	
This gene belongs to the NAD(P)H dehydrogenase (quinone) family and encodes a cytoplasmic reductase. This FAD-binding protein forms homodimers and reduces quinones to hydroquinones. The enzymatic activity of this protein prevents the one-electron reduction of quinones that leads to the production of radical species, thus acting in the detoxification of highly mutagenic and cacinogenic quinones from tobacco, diet and oestrogen metabolism. According to his haplotype the patient is a NQO1 **slow metaboliser**.	Patient haplotype: **Inactive/Inactive**
GSTM1	
This gene encodes a glutathione S-transferase involved in the detoxification of electrophilic compounds, including carcinogens, therapeutic drugs, environmental toxins and products of oxidative stress, by conjugation with glutathione. These genetic variations may modify an individual's susceptibility to carcinogens and toxins, as well as affect the toxicity and efficacy of certain drugs. According to his haplotype the patient is a GSTM1 **normal metaboliser**.	Patient haplotype: ***I/*I**

GSTT1	
The protein encoded by this gene, glutathione S-transferase theta 1 (GSTT1), is a member of a superfamily of proteins that catalyse the conjugation of reduced glutathione to various electrophilic and hydrophobic compounds. According to his haplotype the patient is a GSTT1 **slow metaboliser**.	Patient haplotype: ***D/*D**
SOD2	
This gene encodes a protein that binds to superoxide by-products of oxidative phosphorylation and converts them into hydrogen peroxide and diatomic oxygen, scavenging free radicals from different biological processes. According to his haplotype the patient is a SOD2 **normal metaboliser**.	Patient haplotype: ***1/*1**

GLOSSARY

• **DNA:** Abbreviation for deoxyribonucleic acid. A molecule present in our cells that contains the genetic information necessary for the development and proper functioning of living organisms.

• **Allele:** each of the alternative forms of a gene, which may differ in sequence.

• **Androgens:** steroid hormones that stimulate the development and maintenance of especially male characteristics in the human body, such as facial hair and muscle development.

• **Cell:** basic structural and functional unit of life.

• **Strogens:** a group of sex hormones responsible for developing and maintaining female secondary sexual characteristics, as well as regulating the menstrual cycle.

• **Gene:** segment of DNA that represents the unit of hereditary information.

• **Haplotype:** a set of alleles at a specific set of loci on a chromosome that tend to be inherited together.

• **Heterozygous:** when the two alleles of the same gene are different.

• **Homozygous:** when the two alleles of the same gene are the same.

• **Hormone:** a chemical messenger in the body that regulates various physiological functions, such as growth, development, metabolism and reproduction.

• **Progesterone:** female sex hormone that plays a crucial role in the menstrual cycle and the maintenance of pregnancy.

• **Testosterone:** the main male sex hormone, responsible for the development of primary and secondary sexual characteristics in males.

TECHNOLOGY

DNA Microarray technology consists of a solid surface with microscopic reactions (micro-reactions) or DNA chips, on which molecular probes are attached to detect the presence of target DNA molecules. Probe-target hybridisation is usually detected and quantified by measuring the intensity of a specific fluorescence provided by the molecular probe in the 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, as single nucleotide exchange (single base resolution) can be detected using short oligonucleotide probes (20-25 nucleotides). As a consequence, 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).

QUALITY

The analytical laboratory has standard and effective procedures in place to protect against technical and operational problems. 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 testing process.
As with all screening 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, a verification test may be requested.