EasyDNA

Know Yourself DNA Test
Unlock your genetic secrets today!

SAMPLE REPORT

EasyDNA

Your Ancestry Composition

AFRICAN	2.3%	EUROPEAN	97.7%
AMERICAN	0%	OCEANIAN	0%
SOUTH ASIAN	0%	MIDDLE EAST ASIAN	0%
EAST ASIAN	0%		

IBERIAN 52.5%

The Iberians are the original inhabitants of the Iberian Peninsula, in southwestern Europe, including the countries of Andorra, Portugal and Spain. Although the term Iberian has been used to identify all the inhabitants of the peninsula, there is another more restricted ethnic sense that identifies them as the ethnic group that occupied the east and south of the Iberian Peninsula. These peoples, defined by the Greeks and Romans, would have been influenced by other Mediterranean peoples such as the Phoenicians or the Greeks themselves.It is known that the first settlers arrived in the Iberian Peninsula more than 40,000 years ago from the south of France. From that point and throughout history the Iberian Peninsula has been a crossroads for many ethnic groups from the Mediterranean Sea, Central and Eastern Europe, which is reflected in the great cultural diversity of its current populations. In addition, due to the role played by Spain and Portugal since the end of the 15th century, at the time of the discoveries and subsequent colonization of America, a large proportion of the inhabitants of today's South American countries are descendants of the original Iberian Peninsula population.

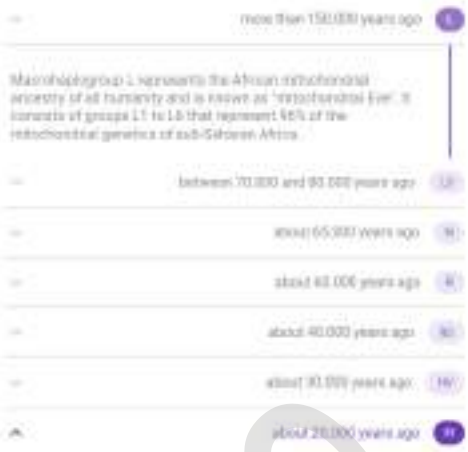
GREEK AND BALKAN 13.7%

The Balkan peninsula, delimited to the north by the Balkan Mountains, is one of the three great peninsulas of southern Europe. The area, besides being surrounded by seas, has several rivers, such as the Danube, the Var or the Struma, which facilitate communication between the different regions. The area comprises several countries such as Greece, Bulgaria, Croatia, Slovenia, Albania and part of Turkey, with a population of almost 53 million inhabitants. A multitude of different languages are spoken in the area, predominantly the Slavic language group (Bulgarian, Slovenian or Macedonian), Greek, and a group of Neo-Latin languages (such as Romanian or Moldovan). In addition, the heterogeneity of the area is also evident in the different cultures of its inhabitants. The majority ethnic group in the area are the Greeks or Hellenes, with a population of more than 15 million people. They are native to Greece, Cyprus and some other Balkan regions, although, as a result of their enormous historical expansion, they constitute a significant diaspora, with Greek communities established all over the world. In ancient times, the Greeks were organized in city-states where the concept of democracy originated. Their own language is the Greek language, spoken since the time of Ancient Greece. It is an Indo-European language that constitutes a branch in itself and is closely related to Armenian and Indo-Iranian languages. This ethnic group is credited with very notable contributions to the field of universal culture, such as the first alphabet or important foundations in the field of philosophy.

Your maternal haplogroup is

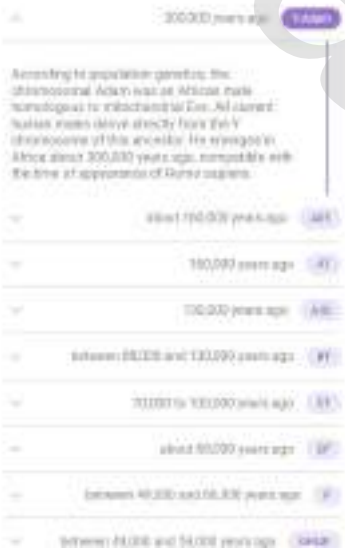
H

Migration of your maternal lineage



Your paternal haplogroup is

R1b



For Sure

1.5%

of Neanderthal DNA

We have analyzed about 9000 genetic variants from the Neanderthal, of which 333 are present in your DNA. These results mean that you have a 16.12% less Neanderthal genetic material than the average of our clients.

Origin and extinction

Neanderthals emerged as a species approximately 230,000 years ago in Europe, the Near East, the Middle East, and Central Asia. It is estimated that the Neanderthal population was constant, not exceeding 7,000 individuals across the continent, reaching its peak 100,000 years ago.

On the other hand, their extinction dates back to 28,000 years ago, and the causes are not fully known. Most studies suggest that the expansion of our species, Homo sapiens, from Africa would have been the main cause of the decline and disappearance of them, despite the interbreeding that occurred between the two.

DEMO ADVANCED
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MIDDLE EAST 0.0

WESTERN ASIAN	0.0
Bedouin	0.0
Morocco - Bedouin	0.0
Egyptian, Levantine and Arab	0.0
Egypt - Egyptian, Levantine and Arab	0.0
Turkish	0.0
Turkey - Turkish, Caucasian and Iranian	0.0

EUROPE 97.7

EUROPEAN	97.70
Greek and Balkan	13.70
Greece - Greek and Balkan	13.7
Basque	14.00
France - Basque	14.0
Italian	0.0
Italy - Italian	0.0
French	0.0
France - French	0.0
Iberian	52.50
Spain - Iberian	52.5
Eastern European	0.0
Poland - Eastern European	0.0
Finnish	0.0

Finland - Finnish	0.0
Sardinian	3.70
Sardinia (Italy) - Sardinian	3.7
Scandinavian	8.00
Norway - Scandinavian	8.0
German	0.0
Germany - German	0.0
Orcadian	0.0
Orkney Islands (United Kingdom) - Orcadian	0.0
British and Irish	0.0
United Kingdom - British and Irish	0.0
Ashkenazi Jew	5.80
Germany - Ashkenazi Jew	5.8

AMERICA 0.0

NATIVE AMERICAN	0.0
Native American(North)	0.0
United States - Native American (North)	0.0
Native American (Central and South)	0.0
Argentina - Native American (Central and South)	0.0

AFRICA 2.3

AFRICAN	2.30
West African (Nigerian and Ghanaian)	0.0
Ghana - West African (Nigerian and Ghanaian)	0.0

Maghrebi	2.30
Morocco - Maghrebi	2.3
West African (Sierra Leonean and Liberian)	0.0
Liberia - West African (Sierra Leonean and Liberian)	0.0
Pygmy and San hunter-gatherer	0.0
South Africa - Pygmy and San hunter-gatherer	0.0
West African (Gambian and Senegalese)	0.0
Gambia - West African (Gambian and Senegalese)	0.0
East African (Kenyan and Ugandan)	0.0
Kenya - East African (Kenyan and Ugandan)	0.0

EAST ASIA 0.0

EAST ASIAN	0.0
Dai Chinese and Indochinese	0.0
Cambodia - Dai Chinese and Indochinese	0.0
Korean	0.0
South Korea - Korean	0.0
Han Chinese	0.0
China - Han Chinese	0.0
Eastern Siberian Natives	0.0
Russia - Eastern Siberian Native	0.0
Siberian Eskimo	0.0
Chukchi Autonomous Okrug (Russia) - Siberian Eskimo	0.0
Japanese	0.0

Japan - Japanese	0.0
Mongolian and NorthChinese	**0.0**
China - Mongolian and North Chinese	0.0

SOUTH ASIA 0.0

CENTRAL AND SOUTH ASIAN	**0.0**
Punjabi and Northwest Indian	**0.0**
India - Punjabi	0.0
Bengali, Bangladeshi and Northeast Indian	**0.0**
Bangladesh - Bengali, Bangladeshi and Northeast Indian	0.0
Sri Lankan and Southern Indian	**0.0**
Sri Lanka - Sri Lankan and Southern Indian	0.0
Pakistani and Afghan	**0.0**
Pakistan - Pakistani and Afghan	0.0
Central Asian	**0.0**
Kazakhstan - Central Asian	0.0
Gujarati Indian	**0.0**
India - Gujarati Indian	0.0

OCEANIA 0.0

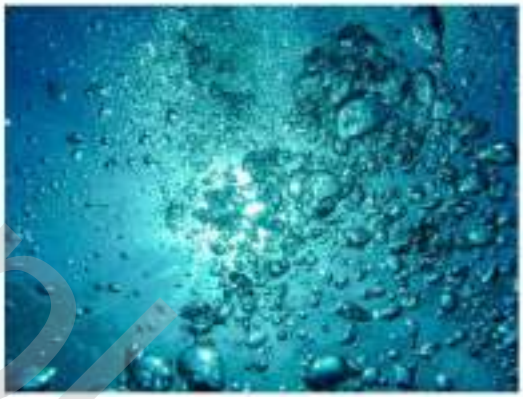
OCEANIAN	**0.0**
Melanesian	**0.0**
Solomon Islands - Melanesian	0.0
Papuan	**0.0**
Papua New Guinea - Papuan	0.0

Your Wellness

WELLNESS

NAME	RESPONSE
Ability to eliminate harmful substances	Slightly reduced
Adipose tissue formation	Normal
Antioxidant capacity	Slightly reduced
Beta-carotene levels and vitamin A metabolization	Slightly increased
Bitter Taste Perception	You may perceive the bitter taste
Blood Glucose	92 mg/dl (average)
Blood Glucose in Obese Children	Increased insulin resistance
Caffeine And Anxiety	Normal levels

Ability to eliminate harmful substances



Xenobiotics are chemical substances that are foreign to animal life and that can cause damage to the organism either directly or through the substances originated after their metabolism. This includes substances such as smoking-induced oxidative substances, drugs, pesticides, cosmetics, food additives, industrial chemicals and environmental pollutants.

Benzene is an important industrial chemical that is ubiquitous in the environment owing to vaporization from petroleum products and combustion of hydrocarbons. Numerous enzyme systems are involved in the metabolism of benzene and its metabolites that take part mainly in the liver: Cytochrome enzymes (CYP), microsomal epoxide hydrolase (EPHX), various glutathione-S transferases (GSTs), NAD(P)H, quinone oxidoreductase (NQO1) and peroxidases; all of them are considered detoxifying enzymes. Here we focus in the EPHX gene which polymorphisms have been associated with several types of cancer and other pathologies; in particular, lung and colorectal cancers show the most convincing evidence.

TECHNICAL REPORT

SNP: rs1051740
Gen or Region: EPHX1

USER	SNP USED	GENOTYPE	RESULT
demo.advancedwn@yopmail.com	rs1051740	TC	Slightly decreased activity of the EPHX1 enzyme. Probably higher risk of lung cancer after exposure to pollutants.

Epoxide hydrolases are enzymes present in all living organisms, transform epoxide-containing lipids to diols by the addition of a molecule of water. The hydrolase EPHX1 is located at in the endoplasmic reticulum of cells and detoxifies a wide range of substrates. Mutations in the EPHX1 gene have been associated with various diseases, including cancer (Václavíková et al. 2015).

The rs1051740 polymorphism (c.337T>C o p. Tyr113His) produces a tyrosine to histidine substitution in exon 3 and is the one of the most studied EPHX1 polymorphisms. This variation decreases the enzyme activity about 40%-50%.

Therefore, presenting the rs1051740-C polymorphism could be linked to a decrease in enzymatic activity and therefore, a greater probability of lung cancer after exposure to contaminating substances.

Due to EPHX1 capability to eliminate carcinogenicity of toxic compounds like epoxides EPHX1 polymorphisms have been extensively studied but results about its relation with cancer predisposition remains unclear and vary among different ethnic groups. Recently, a study performed in infants showed that rs1051740 polymorphism increases the risk of acute leukaemia possibly because the detoxifications of several pollutants, such as benzene from tobacco, are less effective (Ericson et al. 2018).

Several meta-analises conclude that there is evidence that rs1051740-C may be related to an increase of the susceptibility to develop lung cancer in smokers and other studies point out that this polymorphism increase the risk of lung cancer after benzene produced from petroleum products and combustion of hydrocarbons, especially in Asians (Etemadi et al. 2013; Kim et al. 2007; Lee et al. 2011; Liu et al. 2013; Zhang et al. 2015).

NAME	RESPONSE
Caffeine and sports performance	Without effect
Exercise-associated muscle cramps	Low risk
Exercise-induced muscle damage (initial phase)	Probable risk
Exercise-induced muscle damage (regeneration capacity)	Usual ability
Exercise-induced muscle damage (second phase)	Protective effect
Glucose Tolerance Response With Exercise	Typical
Muscle Endurance	Probably endurance athlete
Response To Exercise	No weight loss with exercise and reduced calorie intake
Tendinopathies in lower extremities (legs)	Probable risk
Tendinopathies in upper extremities (arms)	Probable risk

Caffeine and sports performance

Caffeine is the most popular drug consumed worldwide, approximately 80% of the worldwide population consumes caffeine. It is found in more than 60 plants (in the beans, leaves or fruits) and the main source of caffeine in the human diet are coffee beans and tea leaves.

Caffeine is rapidly absorbed in the intestinal tract and metabolized in the liver through enzymatic reactions mediated by cytochrome P450 that produce three metabolites (paraxanthine, theophylline, and theobromine) that together with caffeine can also influence athletic performance. The amount of caffeine increases notably in the bloodstream after 15 to 45 minutes of consumption, reaching the highest level around 60 minutes. Caffeine levels are reduced by 50 to 75% within 3 to 6 hours after consumption.

One primary caffeine's site of action is the central nervous system, due to its liposoluble it crosses the blood-brain barrier without difficulty. Furthermore, caffeine also exerts its effects on the peripheric nervous system and the skeletal muscle.

HOW CAFFEINE IMPROVE EXERCISE PERFORMANCE?

Researchers have indicated that during exercise caffeine can decrease glycogen utilization and increase dependence on free fatty acid mobilization improving significantly the performance.

Caffeine acts as an antagonist of adenosine receptors. Adenosine is a neurotransmitter that transfers the signals related to drowsiness and exertion, thus caffeine reverts the fatigue induced by exercise. Moreover, adenosine inhibits the release of dopamine, thereby reducing mental alertness and motivation; thus caffeine consumption before exercise reverts this effect enhancing dopamine availability and improving alertness and motivation during exercise.

TECHNICAL REPORT

SNP: rs762551
Gen or Region: CYP1A2

USER	SNP USED	GENOTYPE	RESULT
demo.advancedem@yopmail.com	rs762551	AC	Considered slow metabolizer of caffeine. Caffeine consumption probably does not have a beneficial effect in physical performance. For further information please read the text below.

Caffeine is metabolized by the liver through cytochrome P450 enzymatic action and results in three metabolites: paraxanthine, theophylline, and theobromine. The CYP1A2 gene encodes for an enzyme that forms part of the cytochrome P450 superfamily and is responsible for approximately 95% of all the caffeine metabolism (Goldstein et al., 2010).

The rs762551 or -163C>A polymorphism is ubicated in the CYP1A2 gene is a well-known genetic variation that has been related to the ergogenic (or enhancing physical performance) effects of caffeine.

Womack et al. published a study performed in 35 cyclists and proved the ergogenic effects of caffeine in cyclists that are classified as "fast caffeine metabolizers" (genotype AA) in comparison to cyclists with the AC and CC genotypes. After, Guest et al. study (2018) corroborated these results in 10-km male cyclist athletes using low and moderate caffeine doses, 2mg, and 4mg respectively. The authors suggested that the effects of the AA genotype on exercise performance appear to be most prominent during the exercise of longer duration or an accumulation of fatigue (aerobic or muscular endurance) (Guest et al., 2019).

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GENERAL REPORT FOR WELLNESS

DEMO ADVANCED
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NAME	RESULT
Ability to eliminate harmful substances	Slightly reduced
Adipose tissue formation	Normal
Antioxidant capacity	Slightly reduced
Beta-carotene levels and vitamin A metabolization	Slightly increased
Bitter Taste Perception	You may perceive the bitter taste
Blood Glucose	92 mg/dl (average)
Blood Glucose In Obese Children	Increased insulin resistance
Caffeine And Anxiety	Normal levels
Caffeine Consumption	Increased consume
Caffeine and sports performance	Without effect
Celiac disease predisposition	Not predisposed
Childhood Obesity Measurements	Normal
Diet Response	Worse response
Exercise-associated muscle cramps	Low risk
Exercise-induced muscle damage (initial phase)	Probable risk
Exercise-induced muscle damage (regeneration capacity)	Usual ability
Exercise-induced muscle damage (second phase)	Protective effect
Food intake control	Typical
Genetic predisposition to peanut allergy	Slightly increased
Glucose Tolerance Response With Exercise	Typical
HDL Cholesterol Levels	Slightly less
Histamine intolerance	Slight predisposition to DAO deficiency and histamine intolerance
LDL Cholesterol Levels	Slightly less
Lactose Intolerance	Problably tolerant
Long-chain omega fatty acids levels	Decreased

Muscle Endurance	Probably endurance athlete
Obesity Measurements	Increased BMI
Preference For Sweets	Increased
Response To Exercise	No weight loss with exercise and reduced calorie intake
Tendinopathies in lower extremities (legs)	Probable risk
Tendinopathies in upper extremities (arms)	Probable risk
Vitamin B12 levels	Normal
Vitamin C levels	Slightly decreased vitamin C levels
Vitamin D levels	Slightly decreased vitamin D levels
Vitamin E levels	Slightly increased vitamin E levels

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Your Traits

TRAITS

NAME	RESPONSE
Aggressive Prostate Cancer	Typical risk
Alcohol Addiction	Typical alcohol consumption
Alcohol Flush Reaction	Do not show reaction
Asparagus odor detection	More likely to be able to smell asparagus metabolites in urine
Biological Ageing	Slightly reduced
Birth Weight	Slightly increased
Blood Group ABO/Rh	Most likely to be blood type A, Rh-

Aggressive Prostate Cancer

Prostate cancer is very frequent in men in Spain (27.800 cases diagnosed in 2012 according to the Spanish Society of Medical Oncology, SEOM). Although there is a simple treatment for the majority of cases, it is the third cause of death among men, resulting in 5.461 deaths in 2012 only following colorectal and lung cancers. The reason for this elevated number is that there is a type that is 15% more aggressive (known as metastatic *castration-resistant prostate cancer*).

It is now known that there is a gene, GATA2, that is especially active in these types of tumors and that it is resistant to chemotherapy in the treatment of prostate cancer. GATA2 is a gene related to the differentiation and development of eukaryotic organisms (those which have complex cells like those of humans) and which has been linked to some lung and blood cancers. A route has been discovered, a procession of processes that begins in GATA2 (called master gene) and which extends to the phases that influence cellular proliferation.

Each step in these chain reactions involve new genes (or their proteins, which in reality are those that act). IGF2, another of the involved proteins (a growth factor) is also important in this succession of processes, the reason being that, among other causes, there are already drugs that inhibit it and are well-tolerated by persons.

To determine the most adequate treatment for prostate cancer, it is important to classify the tumor and to determine its phase of development.

The classification system most widely used is the TNM system where:

- **T** describes the size of the original (primary) tumor and whether it has invaded nearby tissue.
- **N** describes nearby (regional) lymph nodes that are involved.
- **M** describes distant metastasis (spread of cancer from one part of the body to another).

TECHNICAL REPORT

SNP: rs4054823
Gen or Region: 17p12

USER	SNP USED	GENOTYPE	RESULT
demo.advanceden@yopmail.com	rs4054823	CC	In patients with prostate cancer, typical risk of show an aggressive form of this disease.

The rs4054823 polymorphism is a SNP located on chromosome 17p12 region. This SNP is associated with the risk of aggressive prostate cancer. Most prostate cancers progress slowly and can be treated successfully, even if not detected early. But there is a subset of prostate cancers that represent an aggressive form that is more difficult to treat, especially if are not detected early.

In a study of 4,629 and 12,205 patients with prostate cancer more and less aggressive, respectively, was found that individuals who had genotype rs4054823 (T;T) had more aggressive presentation of prostate cancer compared with the less aggressive form in each of the seven different populations studied.

This SNP is not a predictor for increased prostate cancer risk; only means that patients with prostate cancer who are carriers of this genotype may have a greater predisposition to suffer an aggressive prostate cancer.

Therefore, male carriers of this polymorphism could benefit performing more frequent examinations at early stage or having a more effective prostate cancer treatment if it is detected.

GENERAL REPORT FOR TRAITS

DEMO ADVANCED
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NAME	RESULT
Aggressive Prostate Cancer	Typical risk
Alcohol Addiction	Typical alcohol consumption
Alcohol Flush Reaction	Do not show reaction
Asparagus odor detection	More likely to be able to smell asparagus metabolites in urine
Biological Ageing	Slightly reduced
Birth Weight	Slightly increased
Blood Group ABO/Rh	Most likely to be blood type A, Rh-
Blood coagulation, factor V Leiden and 20210G-A	Does not have factor V Leiden and 20210G-A mutations
C-reactive Protein Levels	1,88 mg/L (average)
CCR5Delta32 and susceptibility to HIV infection	Typical
Cocaine dependence	Increased
Deep sleep	Typical
Drug Abuse	Typical risk
Duffy Antigen (Malaria Resistant)	Probably no resistant
Earlobe type	Low probability of earlobe attachment
Earwax type/Armpit odor	Likely wet earwax. Habitual body odor.
Error Avoidance Capacity	Increased capacity
Eye Color	Probably clear eyes (blue, green)
Freckles	Typical
Gambling behaviour	Slightly increased odds of gambling

HLA-B27 antigen	Absent
Hair Color: Blond Hair	Low probability of blond hair
Hair Color: Red Hair	Probably no red hair
Hair Texture	Likely to have wavy hair
Hair thinckness	Less likely to have thick hair
Height	Tendency to be taller
Heroin Addiction	Typical risk
Intelligence Measurements	Normal
Longevity	Slightly increased
MTHFR gene	Has one copy of A1298C and one copy of C667T in MTHFR
Malaria Complications	Slight protection
Male baldness	Increased risk
Memory	Slightly reduced
Nasion prominence	Low probability of prominent nasion
Nicotine Addiction	Typical depence
Nicotine Dependence And Adolescence	Less risk
Norovirus resistance	Susceptible
PSA (Prostate-specific Antigen) Levels	Increased
Pain Sensitivity	Increased
Permanent tooth eruption	Slightly late eruption
Persistence Of Fetal Hemoglobin	Persistent
Photic Sneeze Reflex	Absent
Pigmented Rings On The Iris	Typical grade
Preterm birth	Increased probability
QT Intervals	More short

Secretor status and ABH antigens (FUT2 gene)	Secretor
Sex hormone regulation	Probably decreased SHBG levels
Skin pigmentation	High likely to have light skin
Sleep movement	Likely to move more than average
Smell, The Sense Of (Olfaction)	Perceives floral aroma
Thyroid function	Increased TSH levels
Tooth morphology	No shovel-shaped incisors

EasyDNA

COMPLEX DISEASES

ALL | Cancer | Cardio | Digestive | Eyes | Mental

HIGH RISK

NAME	RESEARCH STATUS	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Non-alcoholic fatty liver disease (NAFLD)		▲		1.71x

REDUCED RISK

NAME	RESEARCH STATUS	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Crohn's disease		0.03 %	0.2 %	0.14x
Primary biliary cirrhosis (PBC)		▼		0.63x

TYPICAL RISK

NAME	RESEARCH STATUS	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Ulcerative colitis		▲▼		0.94x
Biliary calculus or gallstones		▲▼		0.9x

Non-alcoholic fatty liver disease (NAFLD)

Fatty liver disease or nonalcoholic steatohepatitis or NASH is a metabolic illness that consists of an accumulation of fat in the hepatic cells. Alcohol is the most frequent cause of the build-up of fat in the liver, but when alcohol is not a cause in its development, another origin or cause must be stressed.

Why fat accumulates in the liver is not known with certainty but some mechanisms are very important in the development of the disease, such as insulin resistance, oxidative stress and the liberation of endotoxins, which favor the development of a series of reactions that determine the presence of inflammation and cirrhosis.

Steatohepatitis encompasses a set of clinical conditions that go from the simple presence of fat in the tissue to the development of a hepatic inflammation with the possibility of developing fibrosis, and finally, leading to chronic hepatic damage.

Approximately between 20 – 30% of adults in the general population suffer from this disease and this number increases to between 70 – 90% in those persons who are obese, have diabetes, hypercholesterolemia and hypertriglyceridemia (high levels of blood fats and cholesterol).

SNP : rs738409
Gen or Region : PNPLA3

USER	SNP USED	GENOTYPE	ODDS RATIO * ADJUSTED
demo.advancedem@yopmail.com	rs738409	CG	1.71

Studies in families have shown that the probability of developing non-alcoholic fatty liver disease (NAFLD) is higher in first-degree relatives with NAFLD, suggesting that genetics may play a role in the development of the disease.

The genome-wide association studies or GWAS carried out in recent years has identified multiple genes that could be related to the development of non-alcoholic fatty liver. Among these genes highlights the PNPLA3 gene, which was associated with the disease for the first time in the GWAS performed by Romeo et al. (2008).

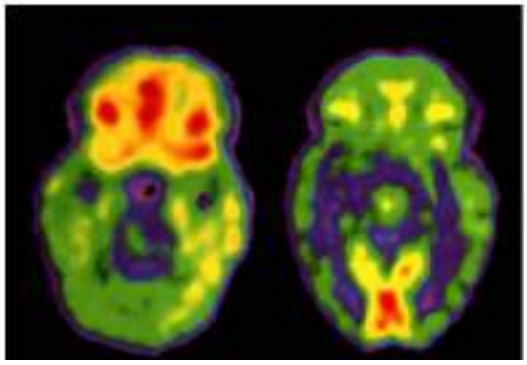
The PNPLA3 ("phospholipase domain-containing protein 3") gene is strongly associated with fat accumulation in hepatocytes and encodes for an enzyme, triacylglycerol lipase, which is found in the smooth endoplasmic reticulum of liver cells.

The rs738409 SNP produces the substitution of an isoleucine by a methionine at position 148 (I148M), which is located near the catalytic center of that lipase. This change appears to produce a reduction in enzyme activity. Romeo et al. observed that liver fat levels were more than doubled in homozygotes for I148M (genotype GG). The rs738409-G allele has also been shown to significantly increase the risk of liver cirrhosis.

HIGH RISK

NAME	RESEARCH STATUS	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Schizophrenia		11.07 %	2.5 %	4.43x
Intracranial aneurysm		2.64 %	2 %	1.32x
Ankylosing spondylitis (AS)		1.45 %	1 %	1.45x
Coronary heart disease		1.28 %	0.5 %	2.57x

Schizophrenia



Schizophrenia is a chronic mental disorder, severe and disabling, that affects close to 1% of the population. The persons who suffer from schizophrenia often hear voices that others do no hear, think that others can read their mind, control their thoughts or conspire to harm them. This can cause fear and turn those affected by the illness into withdrawn and easily irritable persons.

Schizophrenia affects men and women equally. Symptoms like hallucinations and delirium generally start between ages 16 – 30 and men tend to exhibit the symptoms earlier than women. In most cases, a person will not develop schizophrenia after age 45.

SYMPTOMS

Schizophrenia symptoms can be classified into three main categories:

- Positive symptoms: Positive symptoms are psychotic behaviors not generally seen in healthy people (unreal mental contents generated or seen by the patient as real).For some people, these symptoms come and go. For others, they stay stable over time. Sometimes they are severe, and at other times hardly noticeable. The symptoms include auditory hallucinations perceived as voices, the most common hallucination in schizophrenics.
- Negative symptoms: Negative symptoms are associated with disruptions to normal emotions and behaviors. These symptoms are harder to recognize as part of the disorder and can be mistaken for depression or other conditions. These patients need help with their daily tasks.
- Cognitive symptoms: the cognitive symptoms of schizophrenia are subtle, but for others, they are more severe and patients may notice changes in their memory or other aspects of thinking. Similar to negative symptoms, cognitive symptoms may be difficult to recognize as part of the disorder. They make it difficult to lead a normal life and can be distressing to individuals with schizophrenia.

PREVENTION

The exact causes of schizophrenia are unknown although diverse factors that favor its appearance have been discussed. The most known is a genetic predisposition; a 10% of the persons with schizophrenia have a close family member (parents, siblings) who has been diagnosed with it. Diverse genetic mutations related with the development of the disease are also known, although the genetic map of schizophrenia is still being studied.

On the other hand, it is possible that different interactions such as viral infections, psychosocial factors can increase the risk of schizophrenia. There are triggering factors that can be easily avoided, but it seems that hereditary factors play a very important role in developing schizophrenia, which makes it difficult to prevent.

- Persons whose parents have or have had schizophrenia have a genetic predisposition
- Social and psychic factors such as trauma, stress and stressful situations can also contribute to bringing on the disease. Persons with genetic predisposition should avoid stress as much as possible
- Those persons with a genetic predisposition can also avoid schizophrenic episodes by avoiding drug use, especially hallucinogenics such as LSD can trigger schizophrenic psychosis.

TECHNICAL REPORT

SNP : rs7341475
Gen or Region : RELN

USER	SNP USED	GENOTYPE	ODDS RATIO * ADJUSTED
demo.advancedeo@yopmail.com	rs7341475	GG	1.09

According to the WHO (World Health Organization), schizophrenia is relatively more common in men than in women. Furthermore, data seem to indicate that men start with the symptoms at an earlier age. Despite the data, the cause of this bias is currently unknown.

The SNP rs7341475 was identified as a risk factor associated with schizophrenia in a study based on 2274 cases of sick patients, and was found to be linked to female sex. Specifically, the estimated risk for rs7341475(G/G) homozygotes is 1.58.

This SNP is located in the RELN gene that encodes for the protein Reelin. This protein is mainly found in the brain and is crucial for the regulation of neuronal migration processes and positioning in the development of this organ. Defects in this protein have been related to the functional alteration suffered by dopamine neurotransmitters in schizophrenia.

HIGH RISK

NAME	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Schizophrenia	11.07 %	2.5 %	4.43x
Intracranial aneurysm	2.64 %	2.0 %	1.32x
Ankylosing spondylitis (AS)	1.45 %	1.0 %	1.45x
Coronary heart disease	1.28 %	0.5 %	2.57x
Heart attack	0.99 %	0.5 %	1.97x
Rheumatoid Arthritis	0.28 %	0.11 %	2.51x
Colorectal cancer	0.02 %	< 0.01 %	1.83x
Melanoma	0.02 %	< 0.01 %	1.8x
Testicular cancer	< 0.01 %	< 0.01 %	1.48x
Psoriasis	▲		1.38x
Cerebrovascular Accident	▲		1.93x
Thyroid cancer	▲		1.39x
Allergic Asthma	▲		1.55x
Sjögren syndrome	▲		3.0x
Priapism in Sickle Cell Anemia	▲		1.61x
Sudden Cardiac Death	▲		1.55x
Hypertriglyceridemia	▲		1.99x
Non-alcoholic fatty liver disease (NAFLD)	▲		1.71x
Herniated Disc	▲		1.45x
Sarcoidosis	▲		1.4x
Abdominal Aortic Aneurysm	▲		1.28x
Essential tremor	▲		1.22x

DEMO ADVANCED
TWJE-DCBA-3565
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REDUCED RISK

NAME	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
High blood pressure	13.81 %	30.7 %	0.45x
Diabetes Mellitus, Type 2	0.89 %	1.49 %	0.6x
Gout	0.61 %	1.8 %	0.34x
Crohn's disease	0.03 %	0.2 %	0.14x
Neuroblastoma	< 0.01 %	< 0.01 %	0.52x
Diabetes Mellitus, Type 1	< 0.01 %	< 0.01 %	0.54x
Macular degeneration, age-related	< 0.01 %	< 0.01 %	0.06x
Prostate cancer	< 0.01 %	< 0.01 %	0.76x
Systemic lupus erythematosus (SLE)	< 0.01 %	< 0.01 %	0.19x
Multiple sclerosis	▼		0.63x
Lung cancer	▼		0.62x
Osteoarthritis of the knee	▼		0.65x
Primary biliary cirrhosis (PBC)	▼		0.63x
Peripheral arterial hypertension disease	▼		0.65x
Periodontitis	▼		0.72x
Dyslexia	▼		0.63x
Hyperuricemia	▼		0.65x
Chronic obstructive pulmonary disease (COPD)	▼		0.44x
Grave's disease	▼		0.64x
Vitiligo	▼		0.11x
Fuchs dystrophy	▼		0.25x
Narcolepsy	▼		0.66x

Craniofacial anomalies (CFA). Cleft lip and/or cleft palate	0.43x
Exfoliative glaucoma	0.41x
Selective deficiency of IgA	0.72x
Cluster headache	0.66x
Cannabis-induced psychosis	0.78x
Idiopathic membranous nephropathy	0.54x
Osteoarthritis of the hip	0.65x
Progressive supranuclear palsy	0.2x

DEMO ADVANCED
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TYPICAL RISK

NAME	YOUR RISK	AVERAGE RISK	COMPARED TO AVERAGE
Asthma	4.14 %	4.5 %	0.92x
Bipolar disorder	1.36 %	1.2 %	1.13x
Amyotrophic lateral sclerosis	< 0.01 %	< 0.01 %	1.01x
Glioma	< 0.01 %	< 0.01 %	1.04x
Parkinson's disease	< 0.01 %	< 0.01 %	1.09x
Meningioma			0.84x
Bladder cancer			0.95x
Migraine			1.15x
Depression			0.84x
Hypothyroidism			0.83x
Atrial fibrillation			1.06x
Autism			0.89x
Ulcerative colitis			0.84x
Deep vein thrombosis			0.91x
Non-Hodgkin's lymphoma			0.93x
Pulmonary Fibrosis			0.96x
Basal cell carcinoma			0.84x
Cataracts			0.99x
Chronic lymphocytic leukemia			0.88x
Osteoporosis			0.99x
Pancreatic cancer			0.85x
Primary open-angle glaucoma			1.11x
Seasonal affective disorder			0.9x
Biliary calculus or gallstones			0.9x
Chronic kidney disease			0.9x

DEMO ADVANCED
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Eosinophilic esophagitis (EoE)		0.98x
Myasthenia gravis		1.18x
Multiple myeloma		0.84x
Atopic dermatitis or eczema		0.87x
Esophageal Cancer		0.98x
Glioblastoma		1.05x
Oral and throat cancer		1.06x

Your PHARMACOLOGY

EasyDNA

ALL | EFFICACY | INTERACTION | ADVERSE REACTIONS | DOSAGE

PHARMACOLOGY

NAME	RESEARCH STATUS	PHARMACOLOGICAL ACTION	RESPONSE
Atazanavir, Ritonavir (Interaction)		Antivirals	Normal or extensive metabolizer for voriconazole. Usual interactions.
Pseudocholinesterase (Interaction)		General anesthetics	Read technical report

Atazanavir, Ritonavir (Interaction)

It is very frequent for HIV-infected patients to develop fungal infections. In this regard, it is quite common for patients with HIV to be co-prescribed atazanavir/ritonavir (ATZ/RIT) together with the antifungal voriconazole (it is a triazole antifungal agent) or other antifungals.

However, there may be interactions between the antiretroviral agent and the antifungals used that can lead to serious adverse reactions, such as liver damage.

For this reason, it is very important to monitor the adverse effects that occur when these two drugs are prescribed concomitantly and, if necessary, eliminate the antifungal agent, reduce the dose, or change it to another agent that does not cause these interactions with atazanavir.

The EMA ("European Medicines Agency") states that the co-administration of atazanavir/ritonavir together with voriconazole is not recommended unless a benefit/risk study is established that justifies the use of both drugs combined. For this reason, it is highly important to be able to determine certain genetic variants that are involved in the metabolism of voriconazole and other triazole antifungal agents. If it is not possible to carry out a genetic study of these variants, strict monitoring of the safety and efficacy of this combined treatment must be carried out.

TECHNICAL REPORT

SNP : rs4244285
Gen or Region : CYP2C19

USER	SNP USED	GENOTYPE	RESULT
demo.advanceden@yopmail.com	rs4244285	GG	Does not present the CYP2C19*2 allele. In the absence of some other non-functional variant of CYP2C19 is extensive or normal metabolizer for voriconazol and interactions between atazanavir and voriconazole will have the usual impact.

PHARMACOLOGY

NAME	RESEARCH STATUS	PHARMACOLOGICAL ACTION	RESPONSE
Amitriptyline (Dosage)		Cholinergics	Intermediate metabolizer
Amitriptyline (Dosage)		Antidepressants	CYP2D6 Intermediate metabolizer/CYP2C19 Normal metabolizer
Arformoterol (Efficacy)		Respiratory System	CYP2D6 Intermediate metabolizer/UGT1A1 Intermediate metabolizer (Poor Metabolizer)
Aripiprazole (Dosage)		Antipsychotics	Intermediate metabolizer
Atomoxetine (Dosage)		Central-acting psychostimulants	Intermediate metabolizer. May present side effects
Brexpiprazole (Dosage)		Antipsychotics	Intermediate metabolizer
Carbamazepine (Dosage)		Antiepileptics	Ultrarapid metabolizer

Click on the link to learn more about this term in the technical report

Arformoterol (Efficacy)

Arformoterol is a bronchodilator that relaxes the muscles in the respiratory passages to improve breathing. Inhalation of arformoterol is used to prevent bronchoconstriction in people with chronic obstructive pulmonary disease (COPD), including chronic bronchitis and emphysema. Arformoterol will not treat an attack of bronchospasm that has already begun.

Arformoterol is not a rescue medicine. This medicine will not work fast enough to treat an attack of bronchospasm.

Arformoterol may increase the risk of death or hospitalization in people with asthma, but the risk is unknown in people with COPD.

Arformoterol is in the group of drugs called LABA, long-acting beta-2 agonist bronchodilators (such as formoterol, salmeterol, and indacaterol).

ACTION MECHANISM

Arformoterol is a long-acting β2-adrenergic agonist indicated for the treatment of chronic obstructive pulmonary disease (COPD). It is sold as a solution that will be administered twice a day (morning and afternoon) by nebulization.

CAUTIONS

Inform your doctor if you are using any other LABA, such as formoterol, salmeterol or indacaterol. You should not use these medications together with arformoterol. Your doctor will tell you which medications you should use and which ones you should stop taking.

Caution (tell your doctor) if you are taking concomitantly: amlexophylline, amiodarone, antidepressants, beta-blockers, diet pills; disopyramide, diuretics, erythromycin, asthma or cold medications, oral steroids such as dexamethasone, methylprednisolone, and prednisone; pimozide, procainamide, quinidine, thioridazine. Your doctor may need to change the dosage of your medications or monitor you closely for side effects.

TECHNICAL REPORT

SNP : rs5030862
Gen in Region : CYP2D6

USER	SNP USED	GENOTYPE	RESULT
demo.advancedeu@yopmail.com	rs5030862	CC	Does not present the CYP2D6*12 loss of function allele. In the absence of other loss of function alleles will be extensive metabolizer (EM). Standard dose. See also UGT1A1 genotype.

SNP : rs4148323
Gen in Region : UGT1A1

USER	SNP USED	GENOTYPE	RESULT
demo.advancedeu@yopmail.com	rs4148323	GG	Does not present the UGT1A1*6 loss of function isoform. In the absence of other alleles of loss of function will be extensive metabolizer (EM). Standard dose. See also CYP2D6 genotype.

NAME	RESULT
Amifampridina (Dosage)	Intermediate metabolizer
Amitriptyline (Dosage)	CYP2D6 Intermediate metabolizer/CYP2C19 Normal metabolizer
Arformoterol (Efficacy)	CYP2D6 Intermediate metabolizer/UGT1A1 Intermediate metabolizer (Poor Metabolizer)
Aripiprazole (Dosage)	Intermediate metabolizer
Atomoxetine (Dosage)	Intermediate metabolizer. May present side effects
Brexpiprazole (Dosage)	Intermediate metabolizer
Carbamazepine (Dosage)	Ultrarapid metabolizer
Clomipramine (Dosage)	CYP2D6 Intermediate metabolizer/CYP2C19 Normal metabolizer
Clozapine (Dosage)	Intermediate metabolizer
Codeine (Dosage)	Intermediate metabolizer
Cyclophosphamide, Fluorouracil, Methotrexate (Efficacy)	Probably diminished response
Cyclosporine (Dosage)	Poor metabolizer
Desipramine (Dosage)	Intermediate metabolizer
Disulfiram (Efficacy)	Sensitive to disulfiram
Eliglustat (Dosage)	Intermediate metabolizer. Standard dose: 2 doses of 84 mg per day
Fesoterodine (Dosage)	Intermediate metabolizer
Flecainide (Dosage)	Intermediate metabolizer. Dose reduction by 25%
Fluoxetine (Efficacy)	Intermediate metabolizer
Fluvoxamine (Dosage)	Intermediate metabolizer

Haloperidol (Dosage)	Intermediate metabolizer
Iloperidone (Dosage)	Intermediate metabolizer
Methotrexate in Rheumatoid Arthritis (Adverse effects)	Slightly increased risk of adverse effects
Methotrexate in rheumatoid arthritis (Efficacy)	Inefficient response
Metoprolol (Dosage)	Intermediate metabolizer
Nortriptyline (Dosage)	Intermediate metabolizer
Paroxetin (Dosage)	Intermediate metabolizer
Perphenazine (Dosage)	Intermediate metabolizer
Pimozide (Dosage)	Intermediate metabolizer
Pravastatin (Efficacy)	Probably does not respond adequately to pravastatin
Propranolol (Efficacy)	Intermediate metabolizer
Risperidone (Dosage)	Intermediate metabolizer
Tramadol (Dosage)	Intermediate metabolizer
Valproic Acid (Adverse effects)	Increased risk of liver toxicity
Venlafaxine (Dosage)	Intermediate metabolizer
Zuclopenthixol (Dosage)	Intermediate metabolizer
Abacavir (Adverse effects)	No expected variations
Acenocoumarol, Phenprocoumon (Dosage)	Normal or extensive metabolizer
Acetaldehyde (Adverse effects)	No expected variations
Atazanavir, Ritornavir (Interaction)	Normal or extensive metabolizer for voriconazole. Usual interactions.
Brivaracetam (Dosage)	Normal or extensive metabolizer
Celecoxib (Dosage)	Normal or extensive metabolizer

Citalopram (Dosage)	Normal or extensive metabolizer
Clopidogrel (Dosage)	Normal or extensive metabolizer
Diazepam (Efficacy)	Normal or extensive metabolizer
Erythromycin/Sulfisoxazole (Adverse effects)	No risk
Escitalopram (Dosage)	Normal or extensive metabolizer
Esomeprazole (Efficacy)	Normal or extensive metabolizer
Fluorouracil (Adverse effects)	No expected variations
Fluorouracil, Capecitabine, Tegafur (Adverse effects)	Normal or extensive metabolizer
Glipizide (Dosage)	Normal or extensive metabolizer
Irinotecan (Adverse effects)	Read technical report
Ivacaftor (Efficacy)	Does not present the disease
Lansoprazole (Dosage)	Normal or extensive metabolizer
Lorazepam (Efficacy)	Intermediate metabolizer
Mercaptopurine (Dosage)	Normal or extensive metabolizer
Mipomersen (Efficacy)	No symptoms
Omeprazole (Efficacy)	Normal or extensive metabolizer
Oral antidiabetic drugs sulfonylureas (Dosage)	No expected variations
Pantoprazole (Efficacy)	Normal or extensive metabolizer
Pegylated interferon alfa + Ribavirin / Bocepevir (Efficacy)	Read technical report
Phenytoin (Dosage)	Normal or extensive metabolizer

Phenytoin (Efficacy)	No expected variations
Prasugrel (Dosage)	Normal or extensive metabolizer
Prasugrel, Ticagrelor, Clopidogrel (Efficacy)	Normal or extensive metabolizer
Pseudocholinesterase (Interaction)	Read technical report
Rabeprazole (Efficacy)	Normal or extensive metabolizer
Sertraline (Dosage)	Normal or extensive metabolizer
Statins (Adverse effects)	No expected variations
Tacrolimus (Dosage)	Poor metabolizer
Thioguanine, Azathioprine, Mercaptopurine (Dosage)	No expected variations
Ticagrelor (Dosage)	Normal or extensive metabolizer
Voriconazole (Dosage)	Normal or extensive metabolizer
Warfarin (Dosage)	Normal or extensive metabolizer
Acetylsalicylic Acid (ASA) (Adverse effects)	Increased risk
Acetylsalicylic Acid (ASA) in cardiovascular events prevention (Efficacy)	No expected variations
Acetylsalicylic acid (ASA) in colorectal cancer prevention (Efficacy)	Chemoprophylaxis with Aspirin may be beneficial
Amitriptyline (Adverse effects)	CYP2D6 Intermediate Metabolizer (Decrease Dose)/ CYP2C19 Normal Metabolizer
Antidepressants (Efficacy)	Very high input level of antidepressant to the brain (very high response to antidepressants, caution in the dose)
Atazanavir (Adverse effects)	PM for UGT1A1, high probability of developing

	hyperbilirubinemia
Cabozantinib (Efficacy)	Does not respond to treatment
Caffeine (Adverse effects)	Poor metabolizer
Calcium channel blockers and diuretics (Efficacy)	It is better to treat with calcium channel blockers drugs
Chlorthalidone (Efficacy)	Better response
Cisplatin (Efficacy)	Read technical report
Donepezil, Galantamine (Dosage)	Read technical report
Fentanyl (Efficacy)	Poor metabolizer
Gemcitabine in pancreatic cancer treatment (Efficacy)	Good response
General anesthetics (Adverse effects)	Increased risk of postoperative nausea and vomiting
Glimepiride, Glyburide, Gliclazide (Efficacy)	Normal or extensive metabolizer
Inhaled corticosteroids (Efficacy)	Read technical report
Metformin (Efficacy)	Inefficient response
Metoprolol, Carvedilol, Propranolol (Efficacy)	Increased response
Olaparib (Efficacy)	Does not respond to olaparib
Oral antidiabetic drugs sulfonylureas (Efficacy)	Usual response
Paclitaxel (Dosage)	Intermediate metabolizer (significantly increased toxicity)
Paclitaxel in solid tumour treatment (Adverse effects)	Usual neuropathy risk
Palonosetron (Efficacy)	Intermediate metabolizer
Risperidone (Efficacy)	Responds better than usual to risperidone
Tamoxifen (Efficacy)	Intermediate metabolizer
Tolbutamide (Efficacy)	Normal or extensive

	metabolizer
Ziprasidone (Efficacy)	Low response to ziprasidone
Acenocoumarol, Phenprocoumon (Efficacy)	Significantly increased response. Dose adjustment is recommended
Atenolol (Efficacy)	Decreased response
Bevacizumab (Efficacy)	Increased response
Bleomycin (Efficacy)	No expected variations
Bupropion in smoking cessation (Efficacy)	Bupropion is more efficient
Carbamazepine (Adverse effects)	No expected variations
Carisoprodol (Adverse effects)	Normal or extensive metabolizer
Clobazam (Efficacy)	Normal or extensive metabolizer
Cyclophosphamide, Fluorouracil, Methotrexate (Adverse effects)	Side effects more numerous than the usual
Duloxetina (Efficacy)	Slightly better than normal response to duloxetine
Floxacillin (Adverse effects)	No expected variations
Fluoxetine, Citalopram, Escitalopram (Efficacy)	Increased response
Furosemide, Torasemide (Efficacy)	Usual response
Gabapentin (Efficacy)	Probably increased efficacy, decrease dose to avoid adverse effects
Gefitinib (Adverse effects)	No expected variations
Gonadotrophins and Ovulation Stimulants (Efficacy)	Probably ineffective response
Hydrochlorothiazide (Efficacy)	Better response
Iloperidone (Adverse effects)	Probable risk of QT interval prolongation
Imatinib (Efficacy)	Respond to treatment

Lithium (Efficacy)	Effective response
Lumacaftor + Ivacaftor (Efficacy)	It does not present the deletion. Treatment not recommended
Methotrexate in chemotherapy (Adverse effects)	Slightly increased risk of adverse effects
Methotrexate in transplant rejection (Efficacy)	Increased risk of rejection
Methylphenidate (Efficacy)	Probably poor response
Mirtazapine (Efficacy)	Increased response
Morphine, Oxycodone, Fentanyl (Dosage)	Increased response
Naltrexone (Efficacy)	No expected variations
Nelfinavir (Efficacy)	Normal or extensive metabolizer
Olanzapine (Adverse effects)	Much more likely
Paclitaxel (Efficacy)	Usual response
Paracetamol (Efficacy)	Likely adequate or acceptable response
Risperidone (Adverse effects)	Much more likely
Rosiglitazone (Efficacy)	Increased response to Rosiglitazone
Simvastatin (Dosage)	Normal or extensive metabolizer
Trastuzumab (Efficacy)	No expected variations
Triptans (Efficacy)	Likely good efficacy
Venlafaxine (Efficay)	Elevated response to venlafaxine
Vincristine (Efficacy)	Decreased relapse risk
Warfarin (Efficacy)	Highly increased sensitivity to warfarin
Acetylcholinesterase Inhibitors Galantamine, Donepezil, Rivastigmine (Efficacy)	Decreased response
Acetylsalicylic Acid (ASA) (Efficacy)	No expected variations

Alfentanil (Efficacy)	Efficient response
Amisulpride, Aripiprazole, Clozapine, Olanzapine, Haloperidol, Quetiapine, Risperidone, Ziprasidone, Palperidone (Adverse effects)	Slight increased risk of weight-gain
Bisoprolol (Efficacy)	Efficient response
Bupropion (Adverse effects)	Common side effects
Carvedilol (Dosage)	Intermediate metabolizer
Corticosteroids + B2 Agonists (Efficacy)	Better response
Docetaxel (Adverse effects)	Less risk
Gemcitabine (Adverse effects)	Probability of toxicity
Gemcitabine in breast cancer treatment (Efficacy)	Good response
Gemcitabine in non-small cell lung cancer treatment (Efficacy)	Ineffective response
Gemcitabine in the treatment of malignant mesiotheloma (Efficacy)	Good response
Iloperidone (Efficacy)	Decreased response
Methadone (Dosage)	Higher dose of Methadone
Montelukast (Dosage)	Fast metabolizer
Montelukast (Efficacy)	Decreased response
Morphine (Adverse effects)	Usual risk of side effects
Nelfinavir on lymphocytes count (Efficacy)	Usual response
Olanzapine (Efficacy)	No expected variations
Paclitaxel in ovarian cancer treatment (Adverse effects)	Hematotoxicity risk slightly increased
Quetiapine (Dosage)	Normal or extensive metabolizer
Selective Serotonin Reuptake Inhibitors (SSRIs) (Adverse effects)	No expected variations
Sildenafil (Viagra) Response	Usual response
Vaccination (Adverse effects)	Some possibility of adverse reaction
Vincristine (Adverse effects)	Protective effect against side effects
Vincristine (Dosage)	Poor metabolizer

Voriconazole (Adverse effects)	Increased plasma levels

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MONOGENIC DISEASES

MONOGENIC DISEASES

NAME	RESEARCH STATUS	RESPONSE
Hereditary hemochromatosis associated with HFE		Variant present
ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay)		Variant absent
Acute intermittent porphyria		Variant absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)		Variant absent
Alpha-1 Antitrypsin Deficiency		Variant absent
Alpha-mannosidosis		Variant absent
Autosomal recessive polycystic kidney disease		Variant absent
Beta Thalassemia		Variant absent
Biotinidase deficiency		Variant absent
Birt-Hogg-Dube syndrome		Variant absent

Hereditary hemochromatosis associated with HFE

Hemochromatosis is an illness in which the organism accumulates too much iron, an indispensable element for health. When there is an excess of iron, it is not possible to control its functions, converting it into a harmful substance capable of generating free radicals.

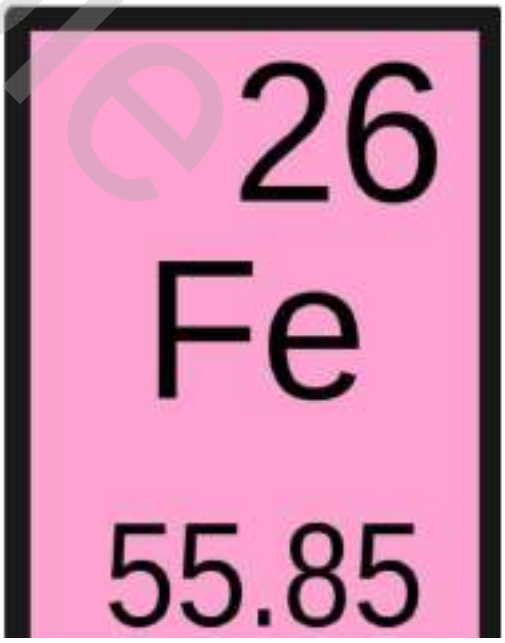
Free radicals are highly reactive chemical substances that irreversibly damage the molecules they react with. This molecular damage then becomes tissue and organ damage. The liver, pancreas, heart, hormonal systems and joints are the organisms most affected.

There are various types of hemochromatosis:

- Primary or hereditary hemochromatosis (HH): this type is brought on by inherited genes that cause the stomach and intestines to absorb too much iron.
- Secondary hemochromatosis: this type is caused by treatments or illnesses that cause an overload of iron in the body. This includes an excess of iron in nutrition, juvenile hemochromatosis, anemia, thalassemia major, sickle-cell disease, chronic anemia) chronic hepatic illness, etc.

If the hemochromatosis is detected early, it is easy to treat. Nonetheless, if it is not treated, the accumulated iron can cause serious damage to the liver, pancreas, heart, hormonal systems, pituitary glands and joints leading to liver cirrhosis, liver cancer, diabetes, heart and joint disease.

Hemochromatosis is caused by an autosomal recessive genetic defect, which means that the child will probably develop hemochromatosis if the two mutations of the HFE gene, one from each parent, are inherited. However, not all persons with two copies of the gene will develop the signs and symptoms of HH.



SYMPTOMS

In the early stages of HH, it is asymptomatic but if the total amount of iron continues increasing, the following symptoms will become apparent:

- Joint and bone pain (the most common)
- Fatigue
- Abdominal pain
- Decreased libido
- Cardiac problems
- Damage to the adrenal gland, leading to adrenal insufficiency

If the illness is not treated in its early stages, the iron can accumulate in the body's tissues causing more serious problems such as:

- Arthritis of the hands (especially the second and third MCP joints), but also the knee and shoulder joints
- Liver cirrhosis, hepatomegaly, cirrhosis , cancer and renal insufficiency
- Pancreatic problems causing insulin resistance and diabetes
- Congestive heart failure, abnormal heart rhythms or pericarditis
- Erectile dysfunction and hypogonadism
- Early menopause
- Darkening of the skin
- Thyroid deficiency
- Damage to the adrenal gland, leading to adrenal insufficiency

PREVENTION

Hemochromatosis can be prevented only by restrictions. Changes in life style and measures to reduce iron intake prevents possible triggering effects and therefore, help to protect the liver:

- Avoid excessive consumption of red meats and uncooked shellfish
- Avoid vitamin C supplements and iron supplements
- Avoid alcohol consumption

Nevertheless, there are no measures that can be taken to prevent hereditary hemochromatosis. It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected by the HFH mutation, are carriers, or are at risk of being carriers or affected. If the genetic mutation is found in only one parent or neither, a genetic mutation of hemochromatosis in offspring can excluded.

TECHNICAL REPORT

SNP : rs1799945
Gen or Region : HFE

USER	SNP USED	GENOTYPE	RESULT
demo.advanceden@yopmail.com	rs1799945	CG	Has one copy of the c.187C>G mutation in the HFE gene. A person with one copy of this mutation typically does not have hemochromatosis but could pass the mutation to the offspring.

SNP : rs1800562
Gen or Region : HFE

USER	SNP USED	GENOTYPE	RESULT
demo.advanceden@yopmail.com	rs1800562	GG	Does not have the mutation c.845G>A in the HFE gene associated with hemochromatosis. May have other mutations (not reported here).

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GENERAL REPORT FOR MONOGENIC DISEASES

DEMO ADVANCED
TWJE-DCBA-3565
06:41:46 - 18/03/2022

NAME	RESULT
Hereditary hemochromatosis associated with HFE	Variant present
ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay)	Variant absent
Acute intermittent porphyria	Variant absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	Variant absent
Alpha-1 Antitrypsin Deficiency	Variant absent
Alpha-mannosidosis	Variant absent
Autosomal recessive polycystic kidney disease	Variant absent
Beta Thalassemia	Variant absent
Biotinidase deficiency	Variant absent
Birt-Hogg-Dube syndrome	Variant absent
Bloom syndrome	Variant absent
Brugada Syndrome	Variant absent
Canavan Disease	Variant absent
Classical homocystinuria due to CBS deficiency	Variant absent
Complete achromatopsia (type 2) and Incomplete achromatopsia	Variant absent
Congenital disorder of glycosylation type 1a (PMM2-CDG)	Variant absent
Congenital muscular -dystroglycanopathy and Walker-Warburg syndrome	Variant absent
Congenital myasthenic syndrome	Variant absent
Congenital stationary night blindness 1C	Variant absent
Cystic fibrosis	Variant absent
Cystinosis	Variant absent
D-Bifunctional Protein Deficiency	Variant absent
Diastrophic dysplasia	Variant absent
Dihydrolipoamide Dehydrogenase Deficiency	Variant absent
Dilated Cardiomyopathy 1A	Variant absent
Dubin-Johnson syndrome	Variant absent
Ehlers-Danlos Syndrome (EDS)	Variant absent

Familial Hypercholesterolemia	Variant absent
Familial Hypertrophic Cardiomyopathy (HCM)	Variant absent
Familial Mediterranean fever	Variant absent
Familial Transthyretin Amyloidosis	Variant absent
Familial adenomatous polyposis	Variant absent
Familial advanced sleep phase disorder (FASPS)	Variant absent
Familial dysautonomia (Riley-Day syndrome)	Variant absent
Familiar hyperinsulinism (ABCC8-related)	Variant absent
Fanconi Anemia (FANCC-related)	Variant absent
GRACILE syndrome	Variant absent
Gaucher disease	Variant absent
Glucose-6-phosphate dehydrogenase deficiency(G6PD deficiency)	Variant absent
Glutaric Acidemia type 1	Variant absent
Glutaric Acidemia type 2	Variant absent
Glycogen storage disease type 1A (Von Gierke Disease)	Variant absent
Glycogen storage disease type 1B	Variant absent
Glycogen storage disease type 2 or Pompe Disease 1 & 2	Variant absent
Glycogen storage disease type 3	Variant absent
Glycogen storage disease type 5	Variant absent
Hemophilia A	Variant absent
Hereditary fructose intolerance	Variant absent
Homocystinuria due to MTHFR deficiency	Variant absent
Hypokalemic Periodic Paralysis	Variant absent
Hypophosphatasia	Variant absent
Junctional Epidermolysis Bullosa	Variant absent
Leigh Syndrome, French-Canadian type (LSFC)	Variant absent
Leukoencephalopathy with vanishing white matter	Variant absent
Li-Fraumeni Syndrome	Variant absent

Limb-girdle muscular dystrophy	Variant absent
Malignant Hyperthermia	Variant absent
Maple syrup urine disease type 1B	Variant absent
Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)	Variant absent
Metachromatic leukodystrophy	Variant absent
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	Variant absent
Mucolipidosis IV	Variant absent
Mucolipidosis type II	Variant absent
Multiple endocrine neoplasia 2B	Variant absent
Neuronal Ceroid-Lipofuscinoses type 1 (associated to PPT1)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 3 (associated to CLN3)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 5 (associated to CLN5)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 6 (associated to CLN6)	Variant absent
Neuronal Ceroid-Lipofuscinoses type 7 (associated to MFSD8)	Variant absent
Niemann-Pick disease type A	Variant absent
Non-syndromic mitochondrial hearing loss	Variant absent
Nonsyndromic Hearing Loss and Deafness, DFNB1	Variant absent
Pendred syndrome	Variant absent
Peters plus syndrome	Variant absent
Phenylketonuria	Variant absent
Pontocerebellar hypoplasia	Variant absent
Primary hyperoxaluria type 1 (PH1)	Variant absent
Primary hyperoxaluria type 2 (PH2)	Variant absent
Pyridoxine-dependent epilepsy	Variant absent
Refsum disease	Variant absent
Retinitis pigmentosa	Variant absent
Rhizomelic Chondrodysplasia Punctata Type 1	Variant absent
Salla Disease	Variant absent

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Short chain acyl-CoA dehydrogenase deficiency (SCADD)	Variant absent
Sjögren-Larsson syndrome	Variant absent
Spinal muscular atrophy	Variant absent
Tay-Sachs disease	Variant absent
Type 1 Oculocutaneous albinism (tyrosinase negative)	Variant absent
Type 2 oculocutaneous albinism (tyrosinase positive)	Variant absent
Tyrosinemia type I	Variant absent
Usher syndrome	Variant absent
Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)	Variant absent
Wilson disease	Variant absent
Zellweger syndrome	Variant absent
cblA Type Methylmalonic aciduria	Variant absent
cblB Type Methylmalonic aciduria	Variant absent