ProMedicDNA

Iron Sensor

Jane Doe
DEMO_DS

ProGenom
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COVER LETTER

Dear Ms. Doe,

Your sample for the analysis arrived on in the laboratory and was evaluated according to the highest laboratory quality standards. The results were evaluated and released by two independent geneticists and molecular biologists. After obtaining the results, your personal report was compiled. We hereby convey the results to you in the format of your choice.

We would like to thank you for your trust and hope that you are satisfied with our service. We are always open to questions and suggestions. Please do not hesitate to contact us. We value your feedback. This is the only way we can continuously improve our services.

We hope the analysis meets your expectations.

Kind regards,

Dr. Daniel Wallerstorfer BSc.
Laboratory Director

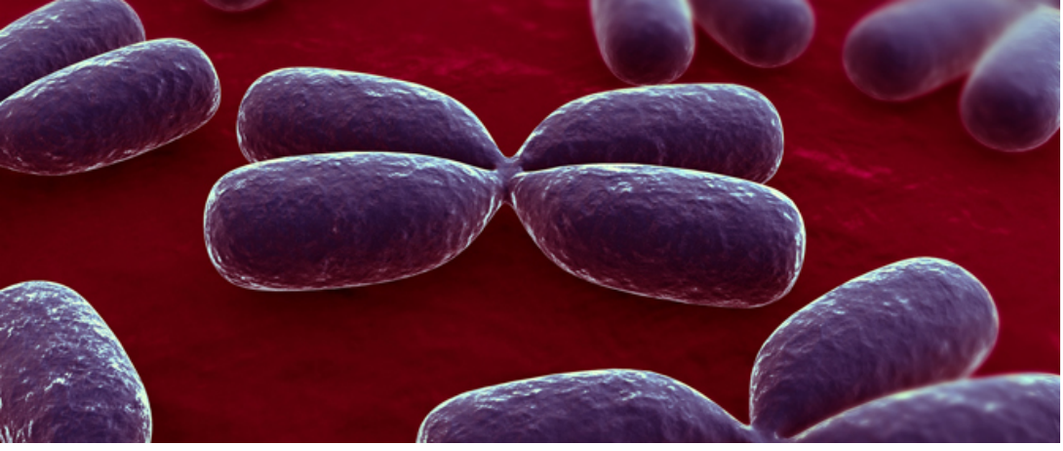
Florian Schneebauer, MSc.
Laboratory Manager

Iron Sensor

Personal analysis results for:
Jane Doe | Date of birth: 01/01/1990

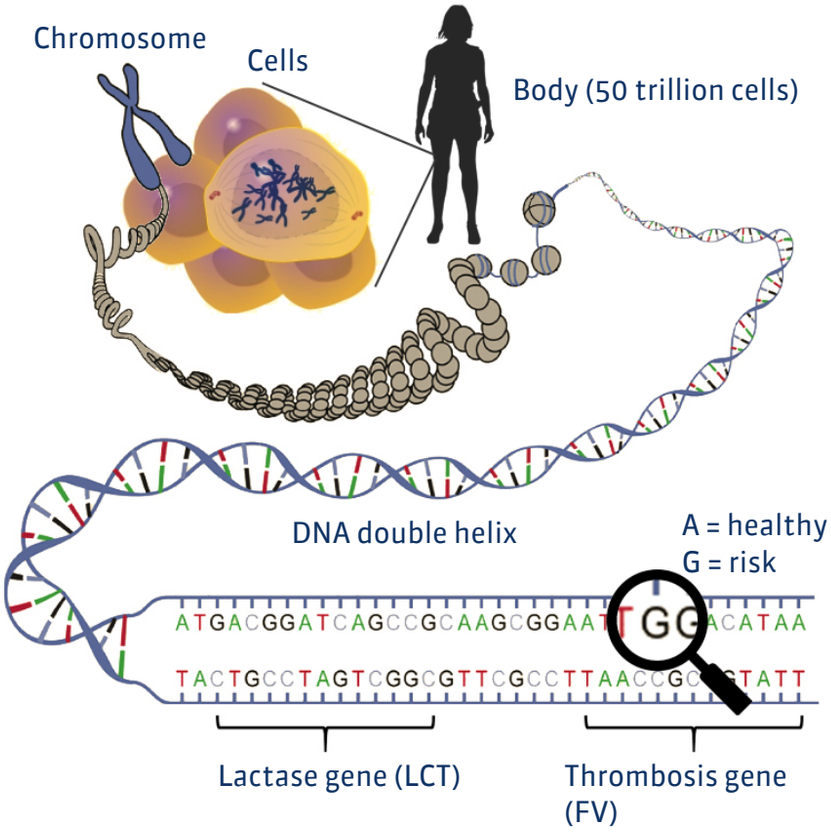
Order number:
DEMO_DS

This report contains personal medical information that is highly confidential. Data protection must be ensured.



How genes influence our health

The human body consists of about 50 trillion individual cells. Most of these cells have a nucleus, which contains 46 chromosomes. A chromosome consists of a very closely wound thread, the DNA "double helix."



DNA, the genetic code, is the blueprint of the human body. This genetic code consists of approximately 3.1 billion molecules, which are each represented by a letter. About 1% of this code makes up the genes. Each gene is an instruction for the body, usually with a single function. For example, some genes tell the body how to colour the iris and differences in these genes produce different eye colors. Every function of the body is controlled by one or more genes, including the way we break down food or medication.

Our genes are not completely error-free. The genes of each person are altered slightly by environmental effects. Most of these changes have no effect but a small number have a harmful effect. An even tinier number can produce a beneficial effect. Parents pass these changes, including defects, to their children. Thus most of our genetic defects are inherited from our parents.

In addition, our genes evolved to help us live in a completely different world, and some of our genetic traits can interact with our modern environment to create negative effects on the body. For example, the genetic predisposition to store dietary fat quickly and lose it slowly is beneficial for people who go through times when food is scarce: they have a better chance of surviving because their bodies use fat efficiently and store it for later. However, in the modern world, this trait is harmful because it programs the body to gain weight quickly and lose weight

slowly. Genes increase our risk of heart attacks, trigger asthma and allergies, cause lactose intolerance, and many other disorders.

Genetic traits can affect our health. While some genetic defects cause disease in all cases, most genetic traits just increase our risk of developing a disease. For example, a person may have genes that increase their risk for diabetes. However, not everyone at risk for diabetes actually develops the disease. Furthermore, even people with a high risk of diabetes can lower their risk with the right diet and exercise plan. Other genetic traits only cause illness when they are triggered by a specific environmental feature. For example, lactose intolerance is a genetic condition that causes a person who drinks milk to have digestive issues. A lactose-intolerant person who never drinks milk will not have any symptoms.

Thanks to the latest technologies, it is now possible to test specific genes to determine if you have genetic traits that are linked to various diseases. Based on the results of the analysis, we can develop a prevention program that significantly reduces your personal disease risk and helps you stay healthy.

A healthy lifestyle will decrease your risk of many diseases whether or not you have specific information about your genetic traits. However, we provide you with additional information that may point out other changes to your lifestyle that are not part of the standard medical advice. There are many examples, but one of the traits we test for is a gene that increases your body's ability to absorb iron. If you have this trait, you must not take iron supplements as the iron would accumulate and cause a life-threatening disease called haemochromatosis.

Experts estimate that every person carries about 2,000 genetic defects, which may affect their health, and in some cases, cause illnesses. A variety of factors can cause changes in our genes (also called mutations). In a few cases, these mutations can benefit us. However, the vast majority either have no effect or have a negative impact on our health. The best-known cause of mutations is radioactivity. Radioactive rays and particles actually impact the DNA in our cells and physically alter our genes. They mostly go unnoticed or cause deadly diseases, such as cancer, or congenital abnormality in newborns. Mutations are also caused by substances in burned food. The substances enter the cells and damage our genes, which can lead to colon cancer, among other forms of cancer. UV radiation from the sun can also damage our genes and cause diseases, such as skin cancer.

External influences can affect individual genes and disrupt their function, but the majority of our defective genes are inherited from our parents. Each embryo receives half of its genes from the father and half from the mother, resulting in a new human being with some characteristics of each parent. Whether a genetic defect is passed on, is determined randomly, and it may be that some of the children carry the defective gene and others do not.

Each person is the unique product of generations of accumulation and combination of different genetic traits. Some of those traits have negative effects on our health. With the latest technology, it is now finally possible to examine genes and determine personal health risks and strengths. In many cases, taking advantage of this knowledge, and following some precautionary measures, the diseases may be prevented. This is the next step in preventive medicine and a new generation of health care.

Action index

Discuss risks marked in orange or red with your doctor. All other results do not require any further attention assuming there are no current medical conditions.

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PHARMACO GENETICS
Not ordered

ONCOLOGY
Not ordered

CARDIOVASCULAR SYSTEM
Not ordered

NEUROLOGY
Not ordered

METABOLISM

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ADDITIONAL INFORMATION

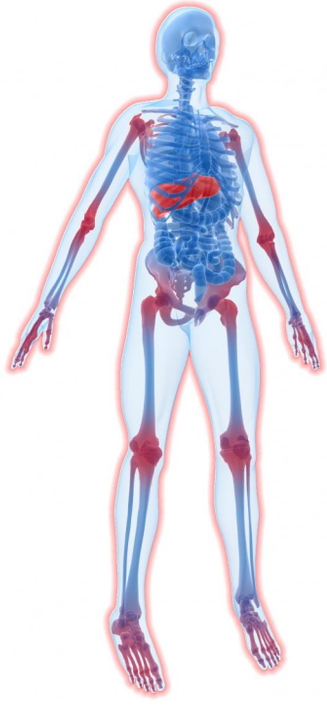
Iron Sensor

Haemochromatosis: easily prevent iron overload

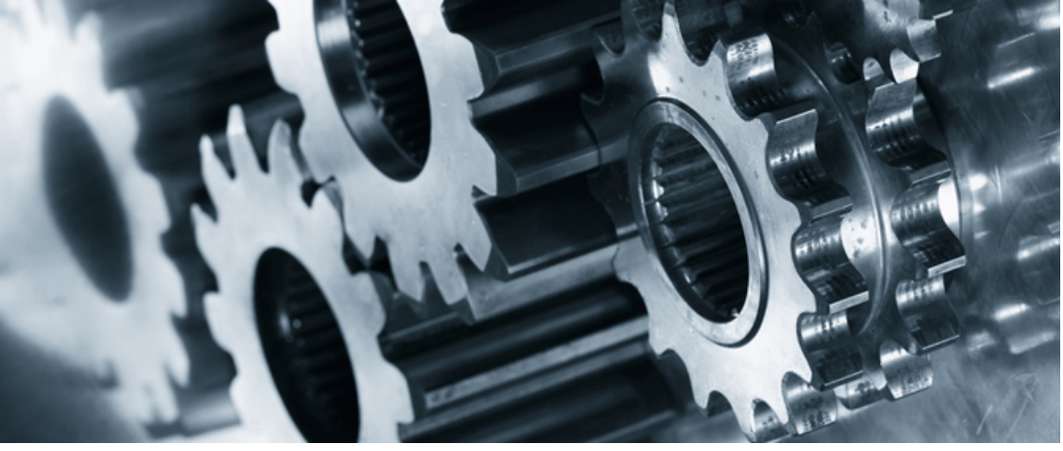
Haemochromatosis (iron overload)

The hereditary condition haemochromatosis, also called iron storage disease, is among the most common inherited metabolic diseases. It is caused by defects in the genes that are responsible for regulating the absorption of iron from food. These defects impair the proper function of these genes and lead to excessive absorption of iron, which over the years is deposited in the organs such as liver, heart, pancreas, pituitary gland and the joints, and damages them. In this case, accompanying diseases such as diabetes and liver cancer may appear.

Haemochromatosis is an "autosomal recessive" disease which usually occurs only when a person has inherited a defective iron-storage gene from both parents. People with only one defective gene have a slightly increased risk of disease, only 5-10% of people with one defective gene have elevated iron levels. The inherited form of haemochromatosis is very common. 1 in 10 persons has a single defective gene and is thus a carrier, while about 1 in 200 people has two defective genes and has a high risk of developing the iron storage disease.



Some symptoms of iron storage disease, eg. elevated liver function, are often misdiagnosed, which leads to wrong treatment and to the worsening of the symptoms. Misdiagnoses is common and, according to experts, 76% of cases are misdiagnosed. If left untreated, this disease can cause early death, but it can be treated and even prevented by regular blood donations (4-6 times per year) or through phlebotomy therapy. It is therefore, helpful to detect a genetic predisposition before symptoms appear. It may be possible to avoid symptoms with the help of preventive measures.



Genes associated with haemochromatosis

Haemochromatosis is usually a recessive disease. This means that the HFE genes from both parents must carry the same polymorphism in order for the disease to develop. In cases where only one gene exhibits a genetic variation affecting its function, the risk of elevated blood iron levels is present but the risk of developing the disease is very low. Your gene analysis shows the following:

Genetic traits

SYMBOL	rs NCBI	POLYMORPH	GENOTYPE
HFE	rs1800562	C282Y	G/G
HFE	rs1799945	H63D	C/C
HFE	rs1800730	S65C	A/A

LEGEND: rsNCBI = description of examined genetic variation, POLYMORPHISM = form of the genetic variation, GENOTYPE = personal analysis result

Summary of effects

Here you can see a summary of the influence your genetic variations have on your health:

- Your body absorbs an average proportion of the iron from your food
- You do not have an elevated risk of haemochromatosis

Your risk of haemochromatosis

LOW AVERAGE RISK HIGHER

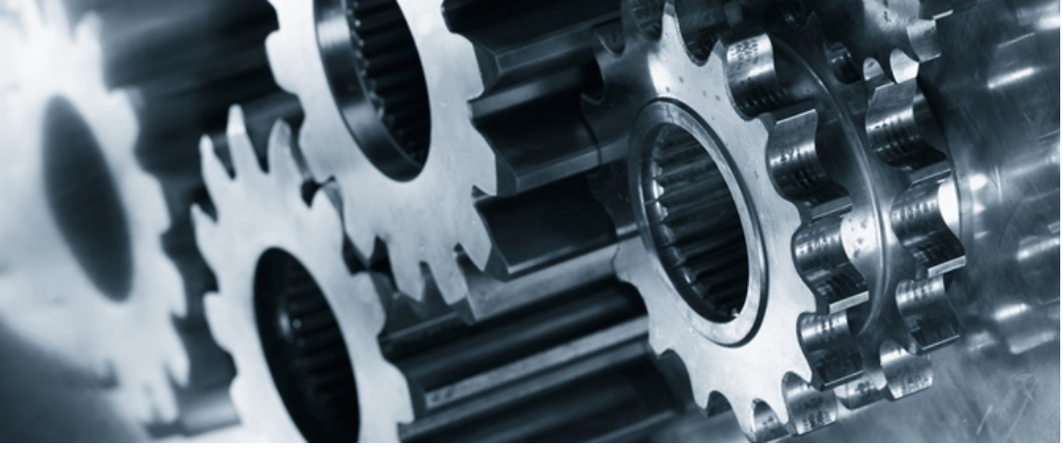
Nutritional Genes - Iron

Your nutrition is very important. Based on your genes and their associated strengths and weaknesses you should increase or decrease certain foods and nutrients. These recommendations are calculated based on your genetic profile.

Your personalized recommendations based on this section:

Iron | Alcohol

Legend: GREEN ARROWS > this nutrient or substance is classed as healthy for your genetic profile. Try to increase the intake of this substance. RED ARROWS > this substance is classed as unhealthy for your genetic profile. Try to reduce your intake of the substance. NO ARROWS > There is no effect of the nutrient on the genetics of this section. PLEASE NOTE! This interpretation only considers your genetic profile of this section.



Prevention

Your genetic analysis shows that you have no increased risk of iron storage disease. Therefore, you do not have to take any special precautions and the following section is simply general information. Affected males develop the first symptoms usually between 20-40 years of age, while women usually develop symptoms after menopause.

- People with an increased genetic risk should donate blood frequently, about 5-6 times per year. When you donate blood, you reduce the iron content in your body. If you develop symptoms of haemochromatosis your blood will not be usable for transfusions.
- Additionally, have your iron level measured twice per year and ask your doctor about how often you should donate blood. If 5-6 donations per year is not a sufficient preemptive measure, your doctor will monitor your iron levels, and if necessary, will start a phlebotomy therapy for you.
- Avoid alcohol and also multivitamins that contain iron.

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SCIENCE

This chapter shows the science behind the test.

Iron Sensor

HFE C282Y - Haemochromatosis (rs1800562)

The HFE gene encodes the hereditary haemochromatosis-protein. The protein is expressed on the cell membrane and forms a complex that enables the binding of the principal iron transport protein, transferrin. Several polymorphisms in the HFE gene are associated with the occurrence of haemochromatosis.

RES	Genotype	POP	Possible results
X	G/G	97%	No increased risk of haemochromatosis
	G/A	1%	Increased risk of haemochromatosis (in combination)
	A/A	2%	Increased risk of haemochromatosis

References

Vujić et al. Molecular basis of HFE-hemochromatosis. Front Pharmacol. 2014 Mar 11,5:42.

Carelle et al. Mutation analysis of the HLA-H gene in Italian hemochromatosis patients. Am J Hum Genet. Apr 1997, 60(4): 828–832.

Beutler E et al. HLA-H and associated proteins in patients with hemochromatosis. Molecular Medicine (Cambridge, Mass.), 3(6), 397–402.

Jouanolle A. M.et al. A candidate gene for hemochromatosis: frequency of the C282Y and H63D mutations. Human Genetics, 100(5–6), 544–7.

Moirand R et al. Haemochromatosis and HFE gene. Acta Gastroenterol Belg. 1999 Oct-Dec,62(4):403-9.

Mura C et al. HFE mutations analysis in 711 hemochromatosis probands: evidence for S65C implication in mild form of hemochromatosis. Blood. 1999 Apr 15,93(8):2502-5.

HFE H63D - Haemochromatosis (rs1799945)

The HFE gene encodes the hereditary haemochromatosis-protein. The protein is expressed on the cell membrane and forms a complex that enables the binding of the principal iron transport protein, transferrin. Several polymorphisms in the HFE gene are associated with the occurrence of haemochromatosis.

RES	Genotype	POP	Possible results
X	C/C	87%	No increased risk of haemochromatosis
	C/G	12%	Increased risk of haemochromatosis (in combination)
	G/G	1%	Increased risk of haemochromatosis

References

Vujić et al. Molecular basis of HFE-hemochromatosis. Front Pharmacol. 2014 Mar 11,5:42.

Carelle et al. Mutation analysis of the HLA-H gene in Italian hemochromatosis patients. Am J Hum Genet. Apr 1997, 60(4): 828–832.

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Mura C et al. HFE mutations analysis in 711 hemochromatosis probands: evidence for S65C implication in mild form of hemochromatosis. Blood. 1999 Apr 15,93(8):2502-5.

HFE S65C - Haemochromatosis (rs1800730)

The HFE gene encodes the hereditary haemochromatosis-protein. The protein is expressed on the cell membrane and forms a complex that enables the binding of the principal iron transport protein, transferrin. Polymorphisms in the HFE gene are associated with the occurrence of haemochromatosis.

RES	Genotype	POP	Possible results
X	A/A	97%	No increased risk of haemochromatosis
	A/T	1%	Increased risk of haemochromatosis (in combination)
	T/T	2%	Increased risk of haemochromatosis (in combination)

References

Mura et al. HFE mutations analysis in 711 hemochromatosis probands: evidence for S65C implication in mild form of hemochromatosis. Blood. 1999 Apr 15,93(8):2502-5.

De Juan et al. HFE gene mutations analysis in Basque hereditary haemochromatosis patients and controls. European Journal of Human Genetics, 9(12), 961–964.

Crownover BK et al. Hereditary hemochromatosis. Am Fam Physician. 2013 Feb 1,87(3):183-90.

Wallace DF et al. Frequency of the S65C mutation of HFE and iron overload in 309 subjects heterozygous for C282Y. J Hepatol. 2002 Apr,36(4):474-9.

Asberg A et al. Hereditary hemochromatosis: the clinical significance of the S65C mutation. Genet Test. 2002 Spring,6(1):59-62.

Mura C et al. HFE mutations analysis in 711 hemochromatosis probands: evidence for S65C implication in mild form of hemochromatosis. Blood. 1999 Apr 15,93(8):2502-5.

LEGEND: RES = your personal analysis result (marked with an X), GENOTYPE = different variations of the gene (called alleles),
POP = percent of the general population that have this genetic result,
POSSIBLE RESULTS = influence of the genetic variation.

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SCIENCE

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In this chapter you will receive useful information

Certifications

Our laboratory is one of the most modern and automated laboratories in Europe and has numerous certifications and quality assurance systems that meet, and even exceed, international standards. The various areas of business are certified separately to the highest standards.

Laboratory diagnostics, manufacturing & sales

Quality management system in accordance with ISO 9001:2015

qualityaustria
SYSTEM CERTIFIED
ISO 9001:2015 No.14365/0

Licensed for medical genetics

Approved by the Federal Ministry of Health, Austria

BUNDESMINISTERIUM FÜR GESUNDHEIT

Cosmetic/genetic diagnostics and cosmetics manufacturing

Good manufacturing practice (GMP) in accordance with ISO 22716:2007

qualityaustria
SYSTEM CERTIFIED
EN ISO 22716:2007 No.00010/0

Food supplement manufacturing

Management system for food safety in accordance with ISO 22000:2018

qualityaustria
SYSTEM CERTIFIED
ISO 22000:2018 No.00521/0

Customer Service

Questions or comments about our service?

Our customer service team is happy to help with any enquiries or problems. You can contact us in the following ways:

- Phone +41 (0) 41 525 100.1
- office.ch@progenom.com

Our team is looking forward to your call. Customer satisfaction is our first priority. If you are not fully satisfied with our service, please let us know. We will do our best to help find a satisfactory solution to your problem.

Contact | Impressum
ProGenom GmbH
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6343 Rotkreuz
SWITZERLAND

Technical details

Order number
DEMO_DS

Date of birth
01/01/1990

Established analysis methods
qRT-PCR, DNA sequencing, fragment length analysis, CNV assay, GC-MS, Immunocap ISAC, Cytolisa

Report generated
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Product codes
M3IRO

Current version
V538

Ordering company
ProGenom GmbH
Riedstrasse 1
6343 Rotkreuz
SWITZERLAND

Analyzing company
DNA Plus - Zentrum für Humangenetik
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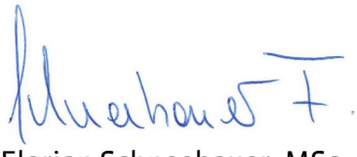
Deutschland

Laboratory Director

Dr. Daniel Wallerstorfer Bsc.

Laboratory Manager



Florian Schneebauer, MSc.

NOTES: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