







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

AMD 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.





GENETICS

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 function. single 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.









PHARMACO GENETICS Not ordered

ONCOLOGY Not ordered

CARDIOVASCULAR SYSTEM Not ordered

NEUROLOGY Not ordered

METABOLISM Not ordered

MOVEMENT Not ordered

DIGESTION Not ordered

OPHTHALMOLOGY

ODONTOLOGY Not ordered

OTHERS Not ordered

SCIENCE

ADDITIONAL INFORMATION



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Macular Degeneration: effective prevention and early detection for best eye health



OPHTHALMOLOGY

Macular degeneration

Macular degeneration is a painless condition affecting the retina of the human eye. The condition usually begins to slowly affect individuals over 50 years of age and impairs the centre of the visual field.

The condition results in a disruptive spot in the centre of the visual field, which can make reading and recognizing details (such as faces) difficult or even impossible without impairing peripheral vision. Macular degeneration is the most common cause of blindness in industrialized countries and roughly 30 million people worldwide are estimated to suffer from the condition. Men and women are equally affected.

The layer of tissue sensitive to light in the human eye is known as the retina. The region of the retina where light is most heavily focused is called the macula. This is the point where your vision is at its highest resolution. Macular degeneration occurs when cells in the macula die with advancing age. It may also be aggravated by the formation of new blood vessels or metabolic waste products that impair macular function. Certain environmental risk factors may accelerate these processes considerably and it is therefore advisable to minimize the risks as much as possible. Risks include: smoking, heart disease and circulatory system conditions, high blood pressure, a poor diet and extreme exposure to light. Preventative measures focus mainly on minimizing such risk factors in order to delay or prevent development of the condition.

Macular degeneration advances slowly over a long period during which symptoms are initially barely noticeable but worsen gradually. Individuals affected usually first experience difficulty reading. Some letters just seem to disappear. Straight lines and edges like window frames appear wavy. This effect can be easily detected and measured by use of a simple test. This is followed by a gradual loss in sharpness of vision, increased difficulty reading, impaired contrast sensitivity and difficulty discerning colour, and increased sensitivity to glare. In advanced stages, the centre of the visual field is often only populated by gray shadows, which themselves disappear as the condition worsens further. As the disease affects only the macula, only the centre of the visual field is affected. Macular degeneration does not cause total blindness because peripheral vision and colour vision remain unaffected. Affected individuals thus retain mobility and orientation. Treatment options for advanced macular degeneration are limited and can usually only slow but not reverse the worsening of symptoms. For this reason, prevention and early detection of macular degeneration are especially important to facilitate timely treatment of the condition.



OPHTHALMOLOGY

Genes associated with macular degeneration

So far, science has identified several genes and polymorphisms linked to an increased risk of macular degeneration. By analysing all relevant polymorphisms, we are able to determine the disease risk. The following genes affect the development of macular degeneration.

Genetic traits							
SYMBOL	rs NCBI	POLYMORPH	GENOTYPE				
HTRA1	rs11200638	G>A	G/G				
CFH	rs1061170	Y402H, T>C	T/C				
LOC387715	rs10490924	G>T	G/G				

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

Summary of effects

- > You do not have an elevated risk of macular degeneration
- > Your requirement of antioxidants is average for your symptoms

Your risk of macular degeneration



Required antioxidants

NORMAL INCREASED





NUTRIGENETICS

Nutritional Genes - Eyes



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:



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

You do not have a genetic predisposition for macular degeneration. You do not need to take special preventive or observational measures because your risk is approximately equal to that of the general population. However, you can still develop macular degeneration, and if you notice symptoms you should discuss them with your doctor.

Even people who have no genetic risk can develop macular degeneration. Therefore, you should have an annual eye test after age 40 to allow for early detection and early treatment of the disease.

- ➤ High blood pressure is a risk factor for macular degeneration. Make sure your blood pressure is within the normal range. You can lower your blood pressure by getting more exercise and adopting the right diet. If diet and exercise do not lower your blood pressure, talk with your doctor about using medication to reduce it.
- Smoking is a major risk factor in the development of macular degeneration, and should be avoided.
- > Protect your eyes from direct sunlight by wearing UV-protective sunglasses or a hat.
- Make sure that your diet includes sufficient amounts of antioxidants, such as vitamins. These are found in fruit and vegetables, and are also available in concentrated form as dietary supplements.

The following sources are recommended:

- ▶ Beta carotene
- ► Copper
- ► Vitamin C
- ► Vitamin E (**a**-Tocopherol)
- ► Zinc

Macular degeneration is slow to develop and painless, therefore pay particular attention to certain symptoms because early detection plays an important role in determining the best treatment. The symptoms include, amongst others, shadowed or distorted vision (eg. window frames appear wavy), or difficulty in reading (eg. when individual letters disappear). The Amsler grid test will allow you to identify the first signs of distortion in your field of vision. The test can be found on the next page together with instructions on how to take it. If you notice any symptoms, consult your eye doctor immediately.



Instructions for macular degeneration self-examination

- > Hold the Amsler grid at a comfortable reading distance.
- Cover one eye (if you have reading glasses, please put them on).
- > Focus on the exact point in the middle with the other eye.
- > Look for wavy or blurred lines.
- > This may indicate symptoms of age-related macular degeneration.
- > Repeat the test with the other eye!
- ► If the you see the irregularities described, contact your optometrist immediately.
- Repeat this self-test once a week.



You do not have a genetic predisposition for macular degeneration. You do not need to take special preventive or observational measures because your risk is approximately equal to that of the general population. However, you can still develop macular degeneration, and if you notice symptoms you should discuss them with your doctor.







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ONCOLOGY Not ordered

CARDIOVASCULAR SYSTEM Not ordered

NEUROLOGY Not ordered

METABOLISM Not ordered

MOVEMENT Not ordered

DIGESTION Not ordered

OPHTHALMOLOGY

ODONTOLOGY Not ordered

OTHERS Not ordered

SCIENCE

ADDITIONAL INFORMATION



SCIENCE

This chapter shows the science behind the test.



SCIENCE

AMD Sensor

HTRA1 - HtrA serine peptidase 1 (rs11200638)

The polymorphism rs11200638 in the HTRA1 gene (high temperature requirement protein A1) is associated with an increased risk for age-related macular degeneration. The encoded protein, a serine protease, plays an important role in the quality control of the extracellular matrix proteins. The mutation in the promoter region of the gene leads to overexpression of the pigment epithelium and to an increased risk of disease.

RES	Genotype	POP	Possible results	
	A/A	9%	Increased risk of macular degeneration (OR: 8.6)	
	A/G	40%	Increased risk of macular degeneration (OR: 2.2)	
Х	G/G	51%	No increased risk for macular degeneration	
References				

Yang et al. A variant of the HTRA1 gene increases susceptibility to age-related macular degeneration. Science. 2006 Nov 10,314(5801):992-3.

Chen et al. Meta-analysis of the association of the HTRA1 polymorphisms with the risk of age-related macular degeneration. Exp Eye Res. 2009 Sep,89(3):292-300.

Dewan et al. HTRA1 promoter polymorphism in wet age-related macular degeneration. Science. 2006 Nov 10,314(5801):989-92.

LOC387715 - Age-related maculopathy susceptibility 2 (rs10490924)

The LOC387715 gene locus is located on chromosome 10. The rs10490924 polymorphism is associated with an increased risk of developing age-related macular degeneration.

RES	Genotype	POP	Possible results
Х	G/G	51%	No increased risk for macular degeneration
	G/T	40%	Increased risk of macular degeneration (OR: 2.69)
	T/T	9%	Increased risk of macular degeneration (OR: 8.21)

References

Fritsche et al. Age-related macular degeneration is associated with an unstable ARMS2 (LOC387715) mRNA. Nat Genet. 2008 Jul, 40(7):892-6.

Rivera et al. Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. Hum Mol Genet. 2005 Nov 1, 14(21):3227-36.

Ross et al. The LOC387715 and age-related macular degeneration: replication in three case-control samples. Invest Ophthalmol Vis Sci. 2007,48:1128–1132.



CFH - Complement factor H (rs1061170)

A defect in the CFH (complement factor H) gene is regarded, in different studies, as the primary risk for the development of AMD. The complement factor H controls the immune response against various pathogens.

RES	Genotype	POP	Possible results	
	T/T	55%	No increased risk for macular degeneration	
Х	T/C	36%	Increased risk of macular degeneration (OR: 4)	
	C/C	9%	Increased risk of macular degeneration (OR: 12)	
Defense				

References

Klein et al. Complement Factor H Polymorphism in Age-Related Macular Degeneration. Science. Apr 15, 2005, 308(5720): 385–389.

Haines et al. Complement factor H variant increases the risk of age-related macular degeneration. Science. 2005 Apr 15,308(5720):419-21.

Hageman G et al. From The Cover: A common haplotype in the complement regulatory gene factor H (HF1/CFH) predisposes individuals to age-related macular degeneration. Proceedings of the National Academy of Sciences, 102(20), 7227–7232.

Thakkinstian A et al. Systematic review and meta-analysis of the association between complement factor H Y402H polymorphisms and age-related macular degeneration. Hum Mol Genet. 2006 Sep 15,15(18):2784-90. Epub 2006 Aug 11.

Kondo N et al. Complement factor H Y402H variant and risk of age-related macular degeneration in Asians: a systematic review and meta-analysis. Ophthalmology. 2011 Feb,118(2):339-44.

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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OTHERS Not ordered

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

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



Food supplement manufacturing

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







CUSTOMER SERVICE

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 Riedstrasse 1 6343 Rotkreuz SWITZERLAND





TECHNICAL DETAILS

Technical details

Order number DEMO_DS

Established analysis methods

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

Product codes M8AMD

Ordering company

ProGenom GmbH Riedstrasse 1 6343 Rotkreuz SWITZERLAND

Laboratory Director

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Date of birth 01/01/1990

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Analyzing company

DNA Plus - Zentrum für Humangenetik Georg Wrede Strasse 13 83395 Freilassing Deutschland

Laboratory Manager

01 004

Florian Schneebauer, MSc.



NOTES:













AMD Sensor Jane Doe DEMO_DS