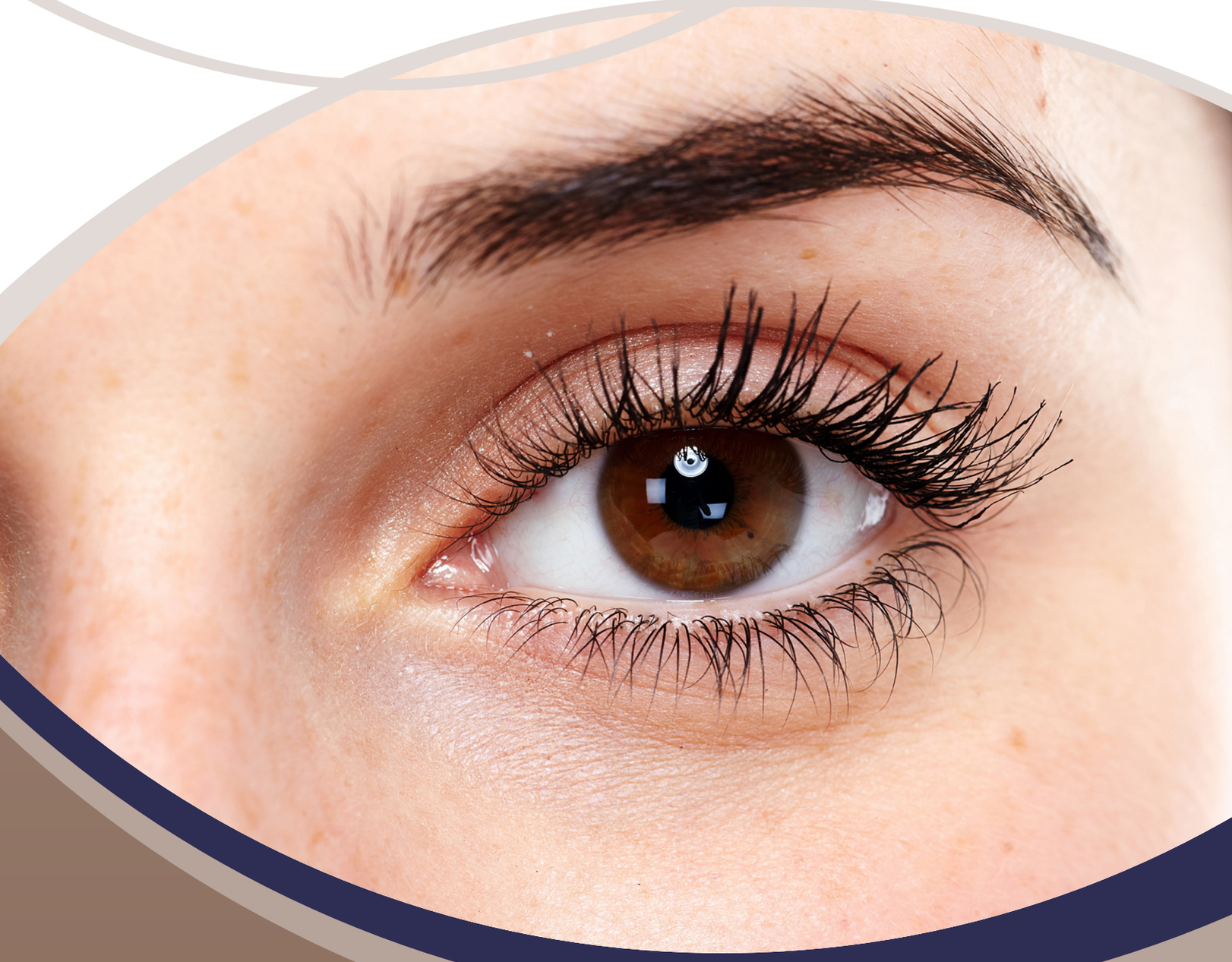
ProMedicDNA



Glaucoma Sensor

Jane Doe
DEMO_DS

ProGenom
www.progenom.com

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

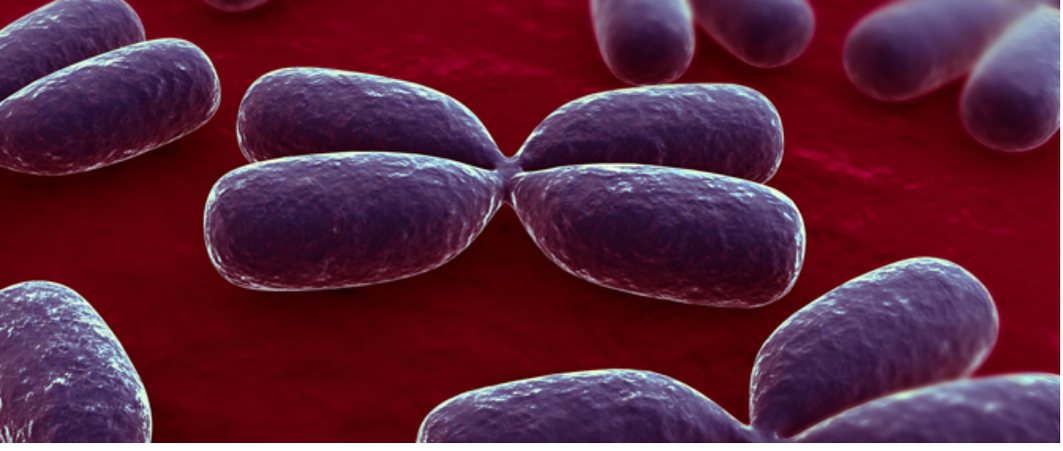
Florian Schneebauer, MSc.
Laboratory Manager

Glaucoma Sensor

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

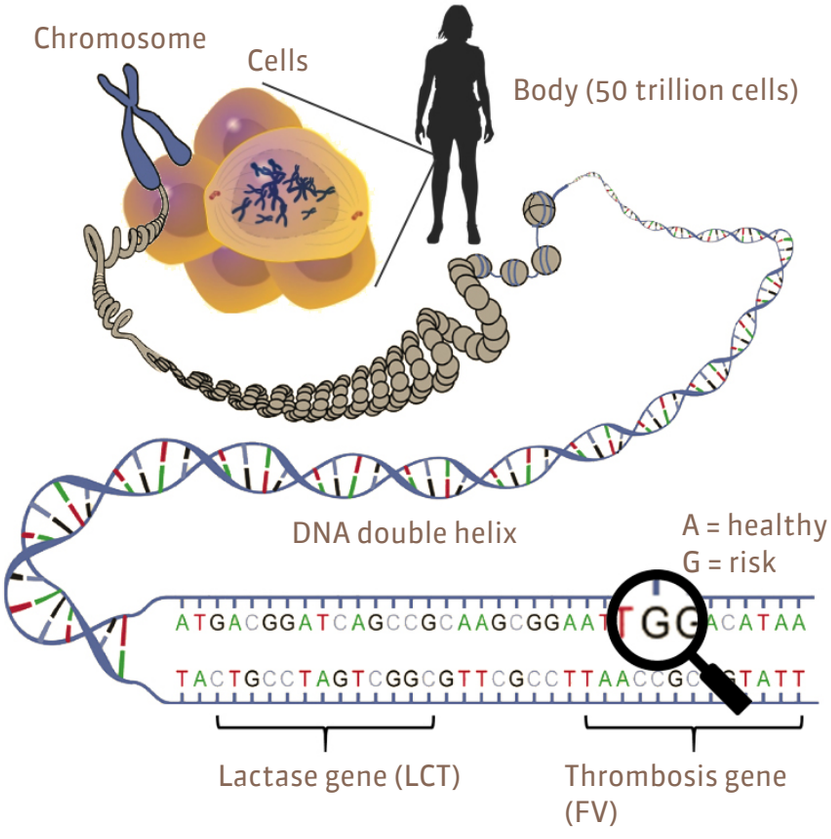
Order number:
DEMO_DS

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.

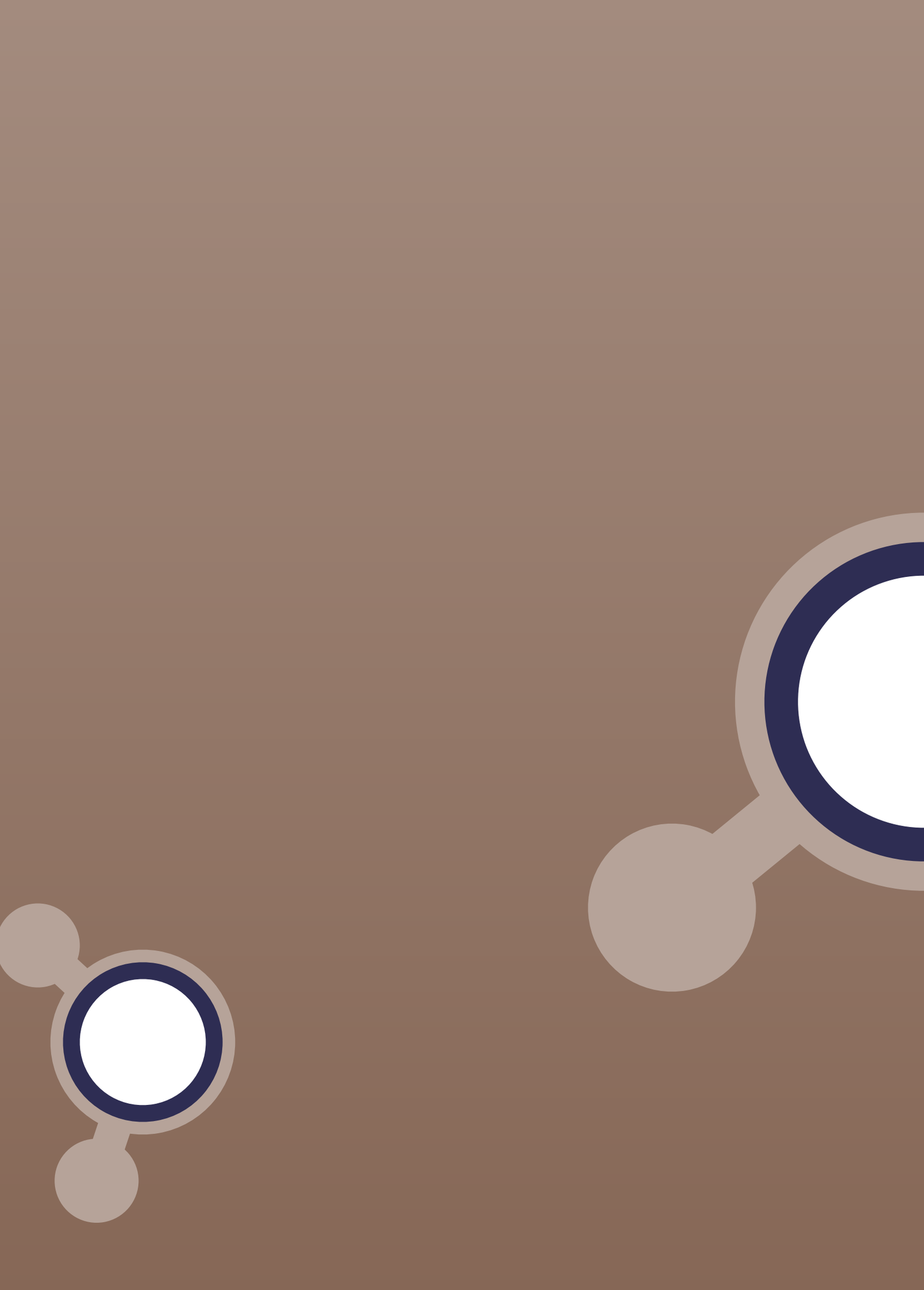
External influences can affect individual genes and disrupt their function, but the majority of our defective genes are inherited from our parents. Each embryo receives half of its genes from the father and half from the mother, resulting in a new human being with some characteristics of each parent. 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.

Glaucoma Sensor



PHARMACO GENETICS
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SCIENCE

ADDITIONAL INFORMATION

Glaucoma Sensor

Detect glaucoma at an early stage and treat it properly

Glaucoma

Glaucoma is a common eye disease and it is one of the leading causes of blindness worldwide. It is estimated that there are currently 500,000 people in Germany suffering from this disease- many unaware. Approximately 10% of them are blind.

Although the disease is easily and effectively treatable with eye drops, most people are unaware of their disease because the symptoms develop slowly and are noticeable only in the advance stages of the disease. Most cases remain untreated for a long time leading to optic nerve damage and, in severe cases, to blindness.

A continuous secretion of clear liquid takes place inside the human eye. This is produced at the posterior part of the eyeball and flows to the anterior parts through the valves. The regulation between the production and secretion creates the correct pressure within the eye, which is important for the its shape and function.
A gene that plays an important role in the function of the vent valves was identified some time ago. Unfavourable genetic variations may interfere with the function of the valves so that the produced fluid cannot be properly drained. This leads to a gradual increase in the intraocular pressure causing pressure on the blood vessels that supply the optic nerve with oxygen and nutrients, thus obstructing the blood flow. If this condition persists untreated, the nerves in the eyes start to gradually wither; in extreme cases it may lead to blindness. The brain combines the image of both eyes and thus will initially compensates for the vision impairment. The disease is usually diagnosed only when both eyes are affected and the patient is experiencing difficulties, for example: overlooking parts of words when reading or having problems while driving. By this time, the optic nerves are often severely damaged resulting in, in most cases, a permanent impairment of the visual field or leading to blindness. After diagnosis, treatment focuses on reducing the eye pressure and on preventing further damage to nerve cells. Damaged nerve cells cannot be repaired.

Preventive genetic testing for glaucoma is recommended because it determines your personal risk for glaucoma. If required, start a medical monitoring program which ensures that the first signs of the disease are immediately recognized and treated properly.

Relevant genes for glaucoma

Science can identify a gene that has an influence on the operation of the drain valves in the eye. As the disease barely manifests itself, and the first vision abnormalities occur only after approximately 95% of the ocular cells have died, it is particularly important to detect the disease as early as possible. The main benefit of this genetic analysis is therefore the recognition of one's own risk, leading to earlier and more accurate eye tests; this will allow for an early diagnosis and proper treatment.

Genetic traits			
SYMBOL	**rs NCBI**	**POLYMORPH**	**GENOTYPE**
LOXL1	rs3825942	T>C	C/C

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 impact your genetic variations have on your health:

- The risk for glaucoma is approximately 1.6 -times increased
- You should submit to regular eye examinations early on to detect the first signs of glaucoma as early as possible.

Risk for glaucoma

Prevention

Based on your genetic profile, you have a significantly increased risk of glaucoma; therefore, it is important that you take preventative measures in order to reduce your risk of developing the disease. Therefore, it would be advisable to consider the following screening measures, which will allow for early detection of the disease. With immediate treatment, permanent damage can be entirely prevented, in most cases.

For this reason, the following screening measures apply:

Get both eyes tested by an optometrist at least once a year to identify the first signs of glaucoma. A simple measurement of intraocular pressure will not detect the early stage of glaucoma. Monitoring should include the following tests:

- Intraocular pressure measurement to determine elevated pressure
- A visual field test for both eyes to detect impairments in the visual field
- Gonioscopy, an eye examination which inspects the eye where the drain valves are located
- The examination of the optic nerve in order to detect any abnormalities.

Low blood pressure contributes to the development of the disease because the flowing blood exerts less force on the already compressed blood vessels. The nerve cells located in the eyes will be even more affected by the oxygen and nutrients. Therefore, monitor your blood pressure, and if necessary, ask your doctor about appropriate treatment.
If you follow these recommendations you should be able to detect the first signs of the disease at an early stage and be properly treated before permanent damage to the optic nerve occurs.

Drug compatibility

DRUGS	139	310	107	262	221	276	524	371	12
GENES	CYP2E1	CYP2D6	CYP2B6	CYP1A2	CYP2C19	CYP2C9	CYP3A4	CYP3A5	NAT2
DEGRADATION	NORMAL	NORMAL	NORMAL	NORMAL	NORMAL	NONE	NORMAL	NORMAL	SLOW

Effect on relevant medication

	Effect	Breakdown	Dose
Acetazolamide	✓	✓	✓
Bimatoprost	✓	✓	✓
Carteolol	✓	✓	✓
Latanoprost	✓	✓	✓
Metipranolol	✓	✓	✓
Timolol	✓	✓	✓

	Effect	Breakdown	Dose
Apraclonidine	✓	✓	✓
Brimonidine	✓	✓	✓
Dorzolamide	✓	✓	✓
Levobunolol	✓	✓	✓
Pilocarpine	✓	✓	✓
Travoprost	✓	✓	✓

	Effect	Breakdown	Dose
Betaxolol	✓	✓	✓
Brinzolamide	✓	↑	↑
Epinephrine	✓	✓	✓
Methazolamide	✓	✓	✓
Tafluprost	✓	✓	✓

Please note: The right choice and dose of medication is always the responsibility of the doctor. Never make your own decision on whether to stop taking a medication or changing its dose!

Legend:

- ✓ ✓ ✓ Effect: Normal. Degredation: Normal. Recommendation: Normal dosage.
- ✓ ↓ ↓ Effect: Normal. Degradation: Slower. Recommendation: Reduce the dosage.
- ✓ ✕ ✕ Effect: Normal. Degradation: None. Recommendation: Alternative drug.
- ↓ ✓ ✓ Effect: Lower. Degradation: Normal. Recommendation: Normal dosage.
- ↓ ↓ ↓ Effect: Lower. Breakdown: Lower. Recommendation: Reduce the dosage.
- ↑ ↑ ✓ Effect: Stronger. Degradation: Stronger. Recommendation: Normal dosage.

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SCIENCE

ADDITIONAL INFORMATION

SCIENCE

This chapter shows the science behind the test.

Glaucoma Sensor

LOXL1 - lysyl oxidase-like 1 (rs3825942)

Lysyl oxidase-like 1 (LOXL1) is a copper-dependent protein that plays an important role in elastogenesis. A genetic defect in LOXL1 gene is associated with an increased risk of glaucoma.

RES	Genotype	POP	Possible results
X	C/C	73%	Increased risk of open-angle glaucoma (OR: 11.19)
	C/T	25%	Increased risk of open-angle glaucoma (OR: 1.66)
	T/T	2%	No increased risk of open-angle glaucoma

References

Chen et al. Ethnicity-based subgroup meta-analysis of the association of LOXL1 polymorphisms with glaucoma. Mol Vis. 2010 Feb 6,16:167-77.

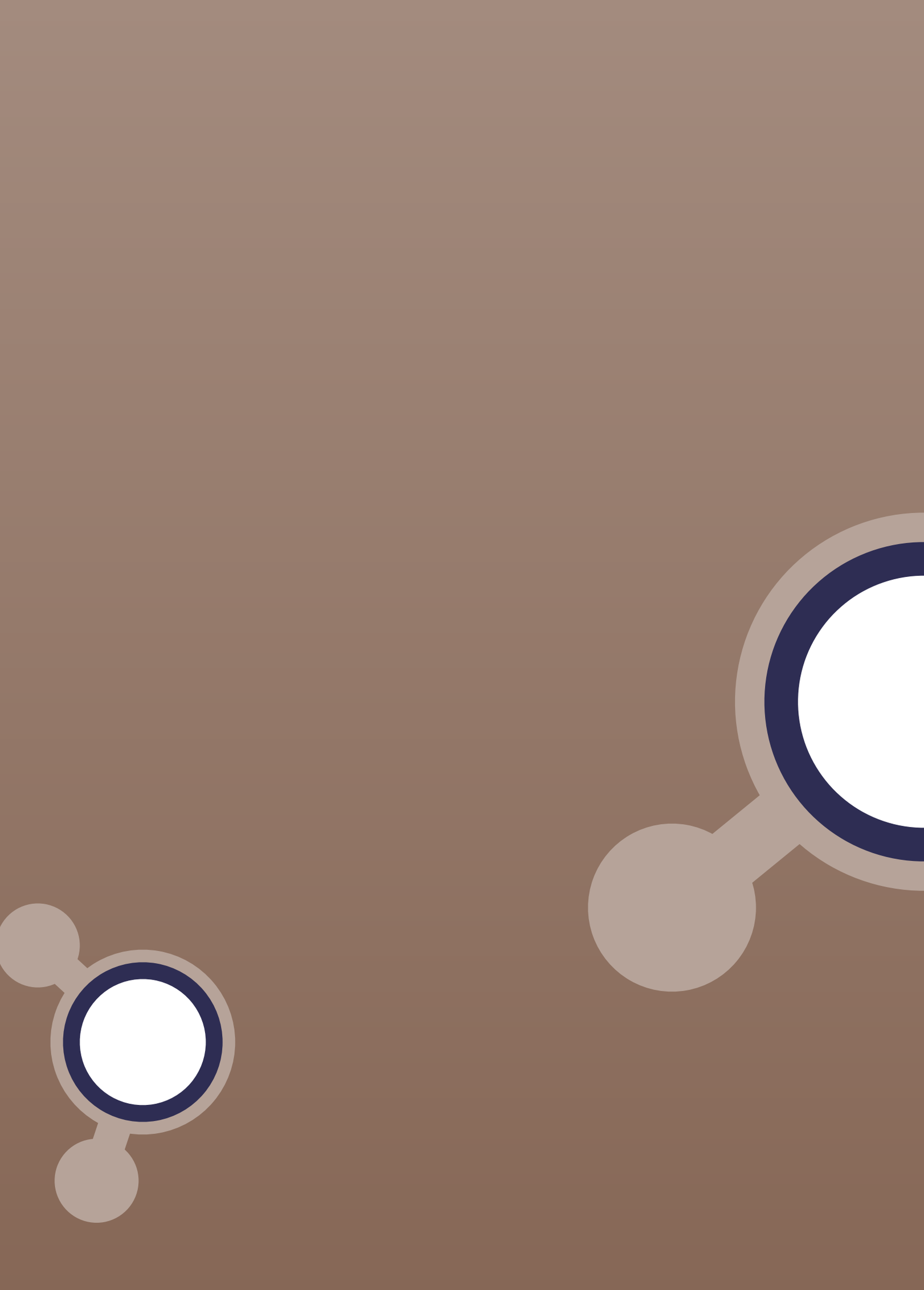
Thorleifsson et al. Common sequence variants in the LOXL1 gene confer susceptibility to exfoliation glaucoma. Science. 2007 Sep 7,317(5843):1397-400. Epub 2007 Aug 9.

Pasutto F et al. Association of LOXL1 Common Sequence Variants in German and Italian Patients with Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. Investigative Opthalmology & Visual Science, 49(4), 1459.

Wang L et al. LOXL1 Gene Polymorphism With Exfoliation Syndrome/Exfoliation Glaucoma: A Meta-Analysis. J Glaucoma. 2016 Jan,25(1):62-94.

Fan BJ et al. DNA sequence variants in the LOXL1 gene are associated with pseudoexfoliation glaucoma in a U.S. clinic-based population with broad ethnic diversity. BMC Med Genet. 2008 Feb 6,9:5.

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

ADDITIONAL INFORMATION

ADDITIONAL INFORMATION

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
Riedstrasse 1
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
19/03/2021 17:39:21

Product codes
M8GLA

Current version
V538

Ordering company
ProGenom GmbH
Riedstrasse 1
6343 Rotkreuz
SWITZERLAND

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

Laboratory Director

Dr. Daniel Wallerstorfer Bsc.

Laboratory Manager

Florian Schneebauer, MSc.

NOTES:


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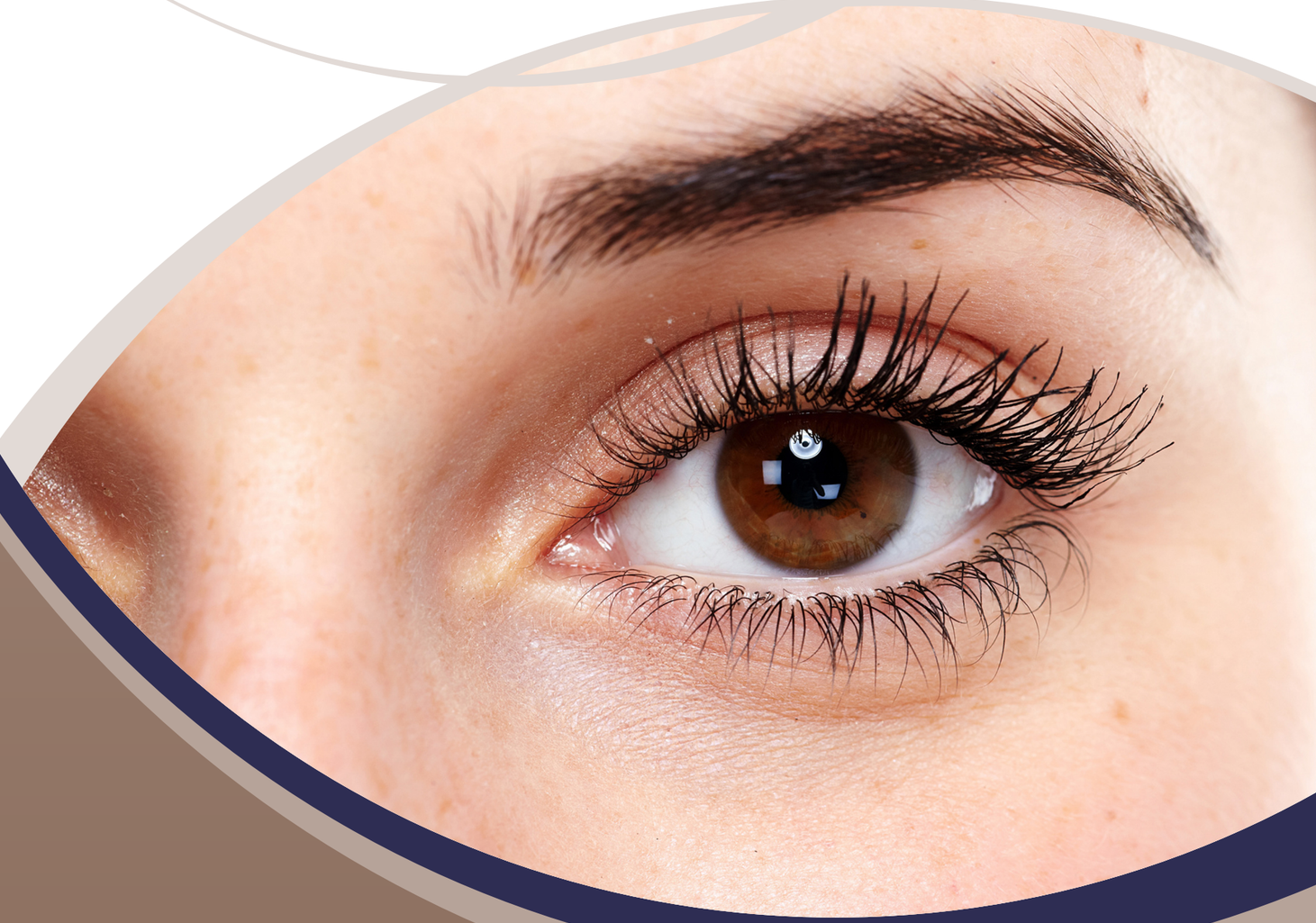

Glaucoma Sensor

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