ProMedicDNA

IBD Sensor

Jane Doe
DEMO_DS

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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

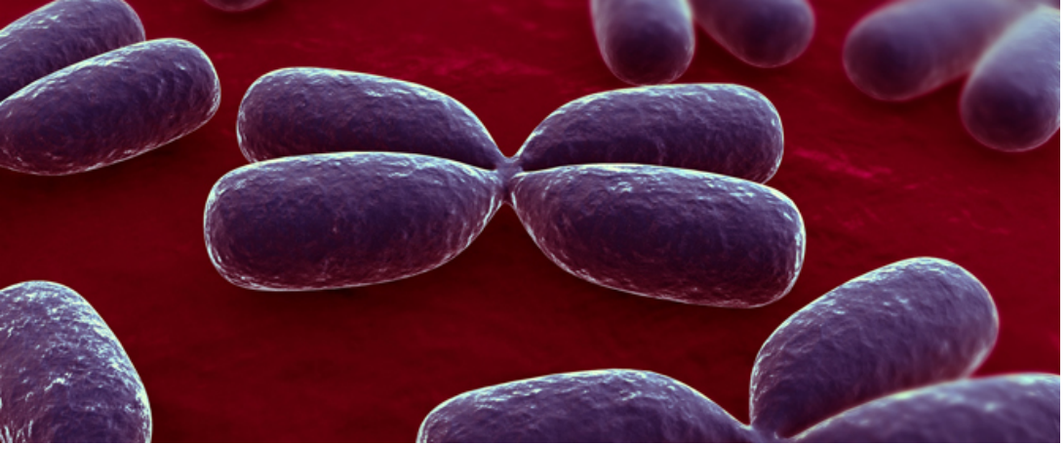
Florian Schneebauer, MSc.
Laboratory Manager

IBD Sensor

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

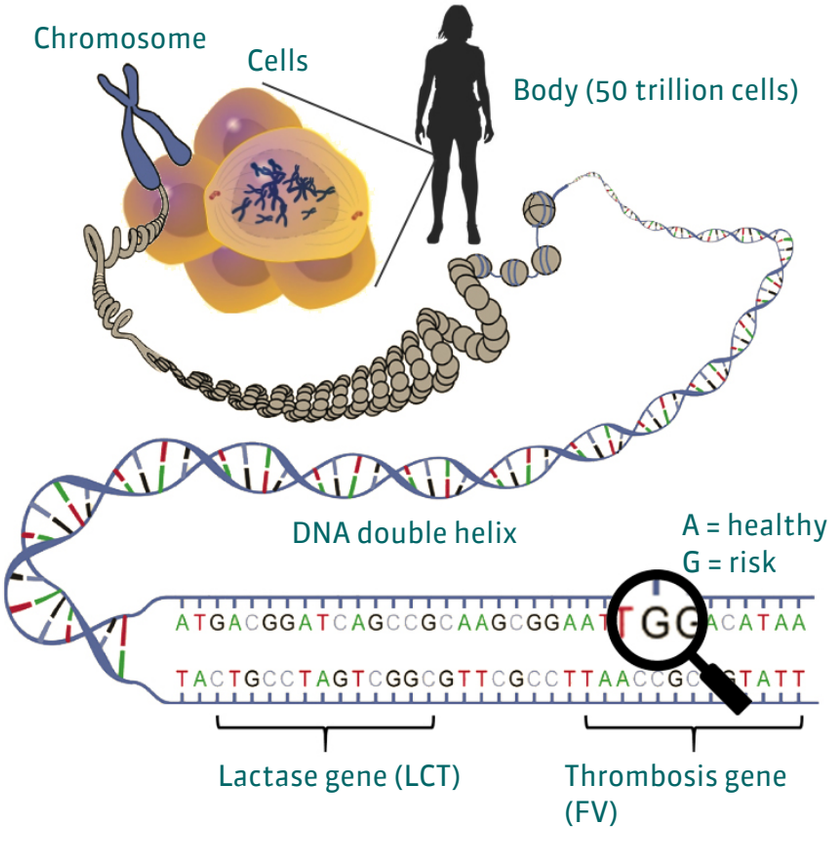
Order number:
DEMO_DS

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
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CARDIOVASCULAR SYSTEM
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NEUROLOGY
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OTHERS
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SCIENCE

ADDITIONAL INFORMATION

IBD Sensor

Crohn's disease early detection and proper treatment

Crohn's disease

The inflammatory gastro-intestinal disease known as Crohn's disease (named after Burill Bernhard Crohn, the first gastroenterologist who identified and described it) is a chronic and progressive intestinal disease that can affect the entire digestive tract. An abnormal immune reaction causes inflammation of the intestine in multiple locations, which causes digestive problems such as diarrhoea and cramps. The inflammation mostly occurs in the colon and small intestine, and more rarely the mouth and oesophagus. Damage to the intestinal tissue increases if the inflammation persists.

Approximately 1 in 700 people suffer from this inflammatory intestinal disease (Crohn's disease), which can be triggered by an inherited error in the intestinal gene 1 (NOD2). This gene is involved in the function of the immune system. Symptoms most often appear for the first time in people between the ages 16 and 35, or people over 60.

Crohn's disease is usually intermittent, with periods of remission alternating with intensive manifestation of symptoms. However, in some cases, this disease can also be chronically active. In many cases, it can take years to correctly diagnose the disease because the first symptoms are temporary digestive issues. Left untreated, the disease may lead to a variety of conditions which must be properly treated.

The cause of the disease is not fully understood. Better understanding of the disease may lead to improved treatments. Currently, the best treatment consists of alleviating symptoms and using immunosuppressants to reduce the immune reaction. Treatment is aimed at reducing the severity of episodes, preventing further attacks, and treating complications such as strictures, fistulas and perforation of the intestinal tissue. In most cases this leads to a significant improvement in the quality of life of those affected. Because many cases are not diagnosed, this genetic test is recommended for people with recurring digestive problems, as it identifies an increased risk of inflammatory bowel disease, and where applicable, the right diagnosis.

Relevant genes for Crohn's disease

The analysed genes have an influence on your risk of developing Crohn's disease and ulcerative colitis. At present, there is no way to reduce your risk of developing Crohn's disease but an accurate diagnosis and proper medical care can significantly reduce the discomfort. The main benefit of this genetic analysis is to determine your risk for Crohn's disease. Close attention to the early symptoms of the disease will help your doctor make an accurate diagnosis relatively quickly and spare you the long ordeal of searching for the correct diagnosis and treatment. These diseases can be successfully treated by an appropriate diet and genetically-tailored drug therapy.

Genetic traits

SYMBOL	rs NCBI	POLYMORPH	GENOTYPE
NOD2	rs2066844	C>T	C/C
NOD2	rs2066845	G>C	G/G
NOD2	rs2066847	del>C	del/del

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:

➤ Your risk of developing Crohn's disease or ulcerative colitis is not increased.

Risk for inflammatory bowel disease

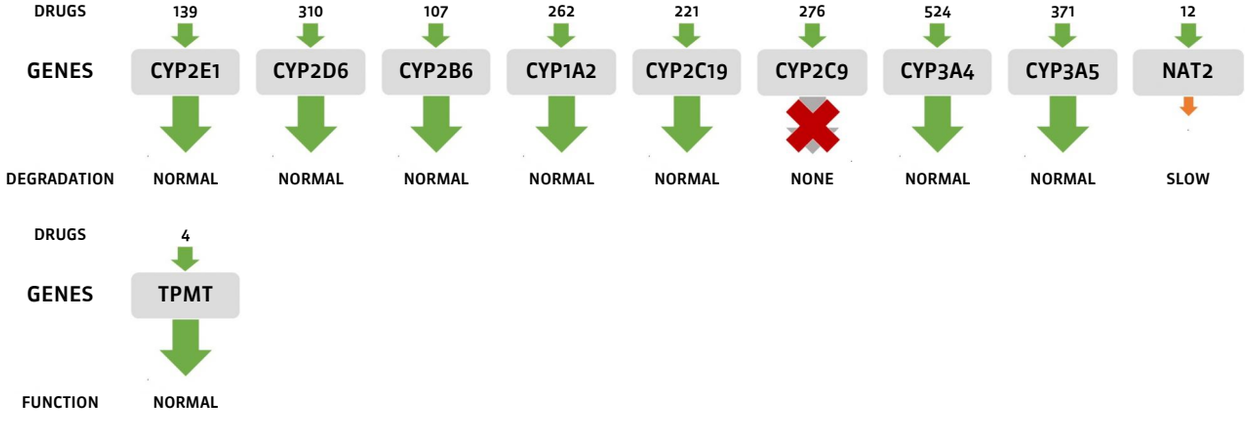
LOW AVERAGE RISK HIGHER

Prevention

Based on the genetic profile, your risk of developing Crohn's disease is not increased.

You do not need to take any special preventative or observational measures since your risk is not increased.

Drug compatibility



Effect on relevant medication

	Effect	Breakdown	Dose
Adalimumab	✓	✓	✓
Budesonide	✓	✓	✓
Corticotropin	✓	✓	✓
Diclofenac	✓	✗	✗
Infliximab	✓	✓	✓
Methylcellulose	✓	✓	✓
Olsalazine	✓	✓	✓

	Effect	Breakdown	Dose
Azathioprine	✓	✓	✓
Budesonide	✓	✓	✓
Cromoglicic Acid	✓	✓	✓
Hydrocortisone	✓	↑	↑
Mercaptopurine	✓	✓	✓
Metronidazole	✓	✓	✓
Prednisone	✓	↑	↑

	Effect	Breakdown	Dose
Balsalazide	✓	✓	✓
Ciprofloxacin	✓	✓	✓
Dexamethasone	✓	↑	↑
Ibuprofen	✓	✗	✗
Methotrexate	✓	✓	✓
Naproxen	✓	✗	✗
Sulfasalazine	✓	✓	✓

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.

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NOD2 - nucleotide-binding oligomerization domain containing 2 (rs2066844)

NOD2 (nucleotide-binding oligomerization domain-containing protein 2) is a receptor protein which recognizes bacterial molecules and activates the NF-KB signaling pathway. This is part of the immune response. NOD2 was identified as a Crohn disease associated gene.

RES	Genotype	POP	Possible results
	T/T	1%	Increased risk of Crohn's disease (OR: 2.52)
	T/C	3%	Increased risk of Crohn's disease (OR: 1.59)
X	C/C	96%	No increased risk of Crohn's disease

References

Jung et al. Genotype/phenotype analyses for 53 Crohn's disease associated genetic polymorphisms. PLoS One. 2012,7(12):e52223.

Hugot et al. Association of NOD2 leucine-rich repeat variants with susceptibility to Crohn's disease. Nature. 2001 May 31,411(6837):599-603.

Glas et al. The NOD2 single nucleotide polymorphisms rs2066843 and rs2076756 are novel and common Crohn's disease susceptibility gene variants. PLoS One. 2010 Dec 30,5(12):e14466.

Yazdanyar et al. Penetrance of NOD2/CARD15 genetic variants in the general population. CMAJ. 2010 Apr 20,182(7):661-5.

NOD2 - nucleotide-binding oligomerization domain containing 2 (rs2066845)

RES	Genotype	POP	Possible results
X	G/G	98%	No increased risk of Crohn's disease
	G/C	1%	Increased risk of Crohn's disease (OR: 1.98)
	C/C	1%	Increased risk of Crohn's disease (OR: 3.92)

References

Jung et al. Genotype/phenotype analyses for 53 Crohn's disease associated genetic polymorphisms. PLoS One. 2012,7(12):e52223.

Hugot et al. Association of NOD2 leucine-rich repeat variants with susceptibility to Crohn's disease. Nature. 2001 May 31,411(6837):599-603.

Glas et al. The NOD2 single nucleotide polymorphisms rs2066843 and rs2076756 are novel and common Crohn's disease susceptibility gene variants. PLoS One. 2010 Dec 30,5(12):e14466.

Yazdanyar et al. Penetrance of NOD2/CARD15 genetic variants in the general population. CMAJ. 2010 Apr 20,182(7):661-5.

NOD2 - nucleotide-binding oligomerization domain containing 2 (rs2066847)

RES	Genotype	POP	Possible results
	C/C	1%	Increased risk of Crohn's disease (OR: 15)
	del/C	1%	Increased risk of Crohn's disease (OR: 11)
X	del/del	98%	No increased risk of Crohn's disease

References

Jung et al. Genotype/phenotype analyses for 53 Crohn's disease associated genetic polymorphisms. PLoS One. 2012,7(12):e52223.

Hugot et al. Association of NOD2 leucine-rich repeat variants with susceptibility to Crohn's disease. Nature. 2001 May 31,411(6837):599-603.

Glas et al. The NOD2 single nucleotide polymorphisms rs2066843 and rs2076756 are novel and common Crohn's disease susceptibility gene variants. PLoS One. 2010 Dec 30,5(12):e14466.

Yazdanyar et al. Penetrance of NOD2/CARD15 genetic variants in the general population. CMAJ. 2010 Apr 20,182(7):661-5.

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

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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
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 17:14:19

Product codes
M2IBD

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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