ProMedicDNA

Skin Health 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

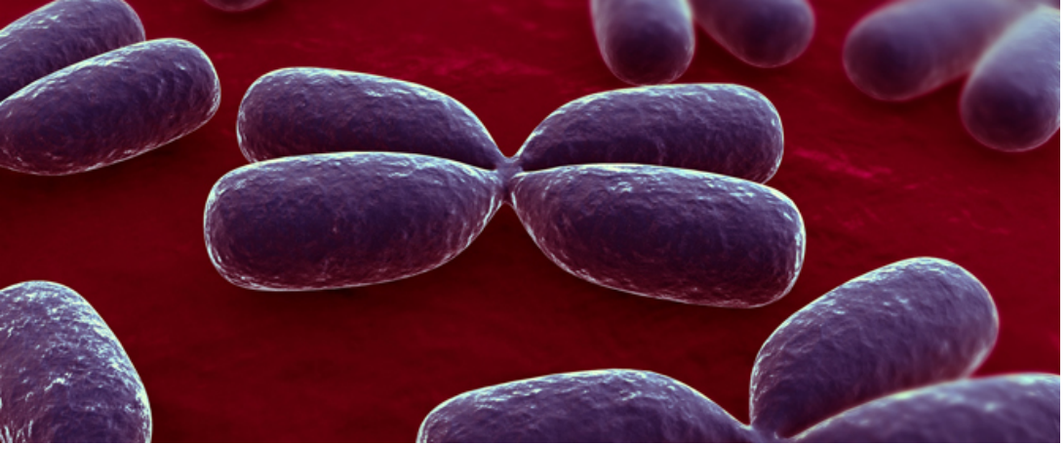
Florian Schneebauer, MSc.
Laboratory Manager

Skin Health Sensor

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

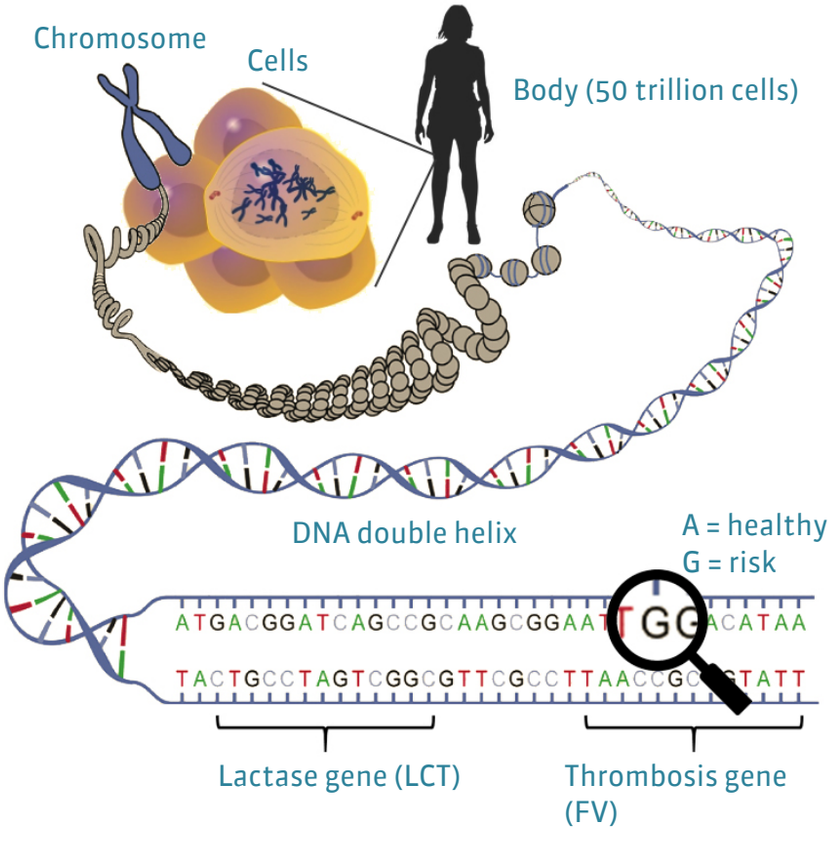
Order number:
DEMO_DS

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

CARDIOVASCULAR SYSTEM
Not ordered

NEUROLOGY
Not ordered

METABOLISM
Not ordered

MOVEMENT
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DIGESTION
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OPHTHALMOLOGY
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ODONTOLOGY
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OTHERS
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SCIENCE

ADDITIONAL INFORMATION

Skin Health Sensor

Effective prevention and early detection of skin cancer

Skin cancer

Although skin cancer is visible on the skin and can theoretically be diagnosed and treated easily, some tumours often develop inconspicuously and in concealed areas, only causing symptoms once they have already spread throughout the body.

There are basically two types of skin cancer:

Light skin cancer: This leads to a cancerous reproduction of light skin cells (basal cells or prickle cells). Although this form of cancer is common, it is usually very treatable and rarely fatal.

The black skin cancer: Black skin cancer, also called melanoma, is rarer and more dangerous. It is caused by a cancerous proliferation of pigment cells, which are usually responsible for a sun tan.

Risk factors

In addition to excessive UV radiation from the sun or a solarium, genetic factors play a significant role in skin cancer progression. Thus, individuals with an increased number of skin blemishes, family history of skin cancer, and skin types that don't tan well and easily develop mild sunburns, are at a particularly high risk.

Prevention and early detection

Protecting yourself from excessive UV radiation with sunscreen is a primary precautionary measure against skin cancer. In addition, the early detection of the disease is considerably important for successful treatment. Usually, skin cancer forms from skin blemishes, which can change visually and possibly lead to discomfort. The blemish will be examined visually by your doctor and, if necessary, also in the laboratory to diagnose possible skin cancer. In many cases, the blemish is then excised, effectively curing the cancer.

Self-examination

Regular self-examination is an essential part of early detection, especially for genetically predisposed individuals and those who have had skin cancer previously. Depending on the level of risk, you should examine your skin for any changes every 3 to 6 months (especially where the blemishes occur). If skin lesions are discovered, they should be discussed with the doctor immediately.

Genes relevant to skin cancer

Several genetic variations have been identified that are known to have an impact on the development of skin cancer. If considering these genetic variations as a whole, they can have a significant impact on the likelihood of developing the disease. The analysis of the relevant genetic variations allowed for the following conclusions:

Genetic traits

SYMBOL	rs NCBI	POLYMORPH	GENOTYPE
ASIP	rs1015362	A>G	G/A
ASIP	rs4911414	G>T	G/T
CDK10	rs258322	C>T	C/C
CLPTM1L	rs401681	T>C	C/T
MC1R	rs11547464	G>A	G/G
MC1R	rs1805005	G>T	G/G
MC1R	rs1805006	C>A	C/C
MC1R	rs1805007	C>T	C/C
MC1R	rs1805009	G>C	G/G
MC1R	rs2228479	G>A	G/G
MC1R	rs885479	G>A	G/G
MTAP	rs7023329	A>G	G/A
MYH7B	rs1885120	G>C	C/G
NCOA6	rs4911442	A>G	A/A
PARP1	rs3219090	A>G	G/G
PIGU	rs910873	G>A	A/A
SLC45A2	rs16891982	C>G	G/G
TYR	rs1393350	G>A	G/A

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

Summary of effects

Here is a summary of the impact genetic variations have on your health:

- Your risk of developing malignant melanoma ("black skin cancer") is lower than the population average.
- Your risk of developing non-malignant melanoma ("light skin cancer") is lower than the population average.

Risk of malignant melanoma ("black skin cancer")

Risk of light skin cancer (non-melanoma skin cancer)

Prevention

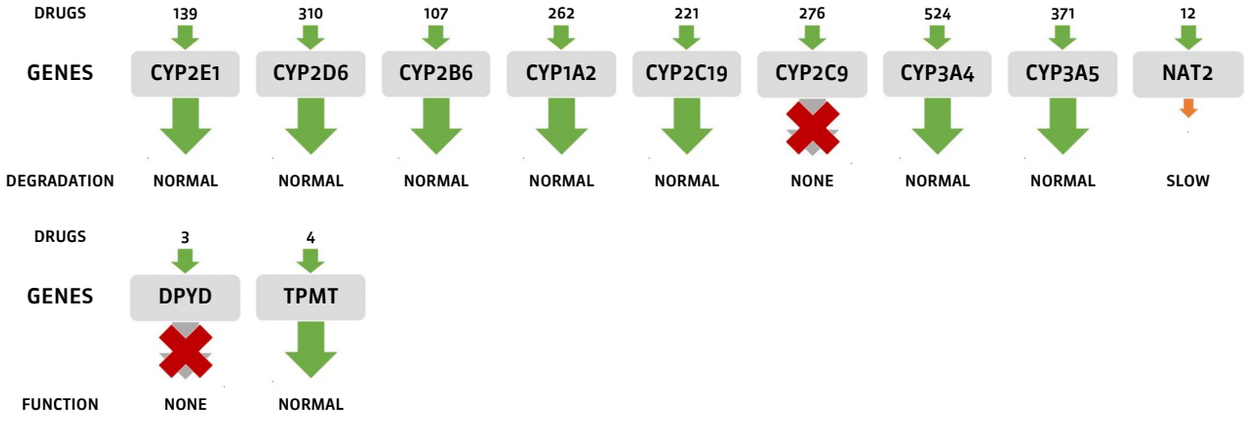
Since you do not have an increased risk of skin cancer, you will not need any precautionary measures beyond the typical recommendations for early skin cancer detection.

Recommendations for prevention and early detection

The following measures are recommended for you:

- Examine all areas of your skin visually and by palpation once a year.
- Talk to your doctor immediately if you notice changes in the skin or blemishes.
- Use sufficient amounts of sunscreen when outdoors.

Drug compatibility



Effect on relevant medication

	Effect	Breakdown	Dose
Alfentanil	✓	↑	↑
Cisplatin	✓	✓	✓
Dolasetron	✓	✓	✓
Fentanyl	✓	↑	↑
Hydrocodone	✓	✓	✓
Lidocain	✓	✓	✓
Metoclopramide	✓	✓	✓
Phenacetin	✓	✓	✓
Tramadol	✓	↑	✓

	Effect	Breakdown	Dose
Aprepitant	✓	↑	↑
Codeine	✓	✓	✓
Domperidone	✓	✓	✓
Fluorouracil	✓	✗	✗
Isoflurane	✓	✓	✓
Methadone	✓	↑	↑
Oxycodone	✓	↑	✓
Ropivacaine	✓	✓	✓
Vemurafenib	✓	✓	✓

	Effect	Breakdown	Dose
Buprenorphine	✓	↑	↑
Dacarbazine	✓	✓	✓
Enflurane	✓	✓	✓
Halothane	✓	✓	✓
Levacetylmethadol	✓	↑	↑
Methoxyflurane	✓	✓	✓
Paracetamol	✓	✓	✓
Sevoflurane	✓	✓	✓
Zolmitriptan	✓	✓	✓

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.

Skin Health Sensor

CDK10 - Cyclin dependent kinase 10 (rs258322)

The protein encoded by this gene belongs to the CDK subfamily of the Ser/Thr protein kinase family. This kinase plays a role in cell cycle regulation. Multiple transcript variants encoding different isoforms have been found for this gene. At least three alternatively spliced transcript variants encoding different isoforms have been reported, two of which contain multiple non-AUG translation initiation sites.

RES	Genotype	POP	Possible results
X	C/C	62%	No increased risk of melanoma
	C/T	28%	Increased risk of melanoma (OR: 1.64)
	T/T	10%	Increased risk of melanoma (OR: 2.69)

References

Stefanaki I et al. Replication and predictive value of SNPs associated with melanoma and pigmentation traits in a Southern European case-control study. PLoS One. 2013,8(2):e55712. doi: 10.1371/journal.pone.0055712. Epub 2013 Feb 5.

Antonopoulou K et al. Updated field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma: the MelGene database. J Invest Dermatol. 2015 Apr,135(4):1074-9.

Bishop DT et al. Genome-wide association study identifies three loci associated with melanoma risk. Nat Genet. 2009 Aug,41(8):920-5.

MTAP - Methylthioadenosine phosphorylase (rs7023329)

Methylthioadenosine Phosphorylase is an enzyme that plays a major role in polyamine biosynthesis and the methionine salvage pathway. The enzyme is deficient in many forms of cancer because the gene and the tumour suppressor p16 gene are co-deleted.

RES	Genotype	POP	Possible results
	A/A	33%	No protection against melanoma
X	A/G	45%	Protection against melanoma (OR: 0.83)
	G/G	22%	Protection against melanoma (OR: 0.69)

References

Bishop DT et al. Genome-wide association study identifies three loci associated with melanoma risk. Nat Genet. 2009 Aug,41(8):920-5. doi: 10.1038/ng.411. Epub 2009 Jul 5.

Antonopoulou K et al. Updated field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma: the MelGene database. J Invest Dermatol. 2015 Apr,135(4):1074-9.

Einfalt et al. Confirmation of single nucleotide polymorphism rs7023329 in MTAP as a melanoma risk factor in a German population.

Livia Maccioni et al. Variants at the 9p21 locus and melanoma risk. BMC Cancer. 2013 Jul 2,13:325.

MYH7B - Myosin heavy chain 7B (rs1885120)

MYH7B is belongs to the motor domain superfamily. It catalyzes the ATP hydrolysis and interacts with actin. Experiments have shown that MYH7B is not expressed in a large number of melanoma cell lines.

RES	Genotype	POP	Possible results
	G/G	98%	No increased risk of melanoma No increased risk of non-melanoma skin cancer
X	C/G	1%	Increased risk of melanoma (OR: 1.55) Increased risk of non-melanoma skin cancer (OR: 1.46)
	C/C	1%	Increased risk of melanoma (OR: 2.40) Increased risk of non-melanoma skin cancer (OR: 2.13)

References

Stefanaki I et al. Replication and predictive value of SNPs associated with melanoma and pigmentation traits in a Southern European case-control study. PLoS One. 2013,8(2):e55712. doi: 10.1371/journal.pone.0055712. Epub 2013 Feb 5.

Lin W et al. ASIP genetic variants and the number of non-melanoma skin cancers. Cancer Causes Control. 2011 Mar,22(3):495-501.

Brown KM et al. Common sequence variants on 20q11.22 confer melanoma susceptibility. Nat Genet. 2008 Jul,40(7):838-40.

Nan H et al. Melanoma susceptibility variants on chromosome 20q11.22 are associated with pigmentary traits and the risk of nonmelanoma skin cancer. Br J Dermatol. 2010 Feb 1,162(2):461-3. doi: 10.1111/j.1365-2133.2009.09579.x. Epub 2009 Nov 30.

Chatzinasiou F et al. Comprehensive field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma. J Natl Cancer Inst. 2011 Aug 17,103(16):1227-35.

Antonopoulou K et al. Updated field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma: the MelGene database. J Invest Dermatol. 2015 Apr,135(4):1074-9.

Bishop DT et al. Genome-wide association study identifies three loci associated with melanoma risk. Nat Genet. 2009 Aug,41(8):920-5.

NCOA6 - Nuclear receptor coactivator 6 (rs4911442)

Nuclear receptor coactivator 6 is a multifunctional transcription coregulator that can enhance transactivation by other transcription factors and is involved in cell survival, growth and development. The gene is amplified in several human cancers including breast cancer, colon and lung cancers.

RES	Genotype	POP	Possible results
X	A/A	93%	No increased risk of melanoma
	A/G	6%	Increased risk of melanoma (OR: 1.76)
	G/G	1%	Increased risk of melanoma (OR: 2.73)

References

Gibbs DC et al. Inherited genetic variants associated with occurrence of multiple primary melanoma. Cancer Epidemiol Biomarkers Prev. 2015 Jun,24(6):992-7.

Stefanaki I et al. Replication and predictive value of SNPs associated with melanoma and pigmentation traits in a Southern European case-control study. PLoS One. 2013,8(2):e55712. doi: 10.1371/journal.pone.0055712. Epub 2013 Feb 5.

Brown KM et al. Common sequence variants on 20q11.22 confer melanoma susceptibility. Nat Genet. 2008 Jul,40(7):838-40.

Maccioni L et al. Variants at chromosome 20 (ASIP locus) and melanoma risk. Int J Cancer. 2013 Jan 1,132(1):42-54.

PARP1 - Poly(ADP-ribose) polymerase 1 (rs3219090)

PARP1 (Poly(ADP-Ribose) Polymerase 1) is an enzyme which modifies various nuclear proteins by poly(ADP-ribosyl)ation. The protein is involved in the regulation of various cellular processes such as differentiation, proliferation and tumour transformation.

RES	Genotype	POP	Possible results
	A/A	30%	Protection against melanoma (OR: 0.74)
	A/G	43%	Protection against melanoma (OR: 0.86)
X	G/G	27%	No protection against melanoma

References

Peña-Chilet M et al. Genetic variants in PARP1 (rs3219090) and IRF4 (rs12203592) genes associated with melanoma susceptibility in a Spanish population. BMC Cancer. 2013 Mar 27,13:160.

Antonopoulou K et al. Updated field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma: the MelGene database. J Invest Dermatol. 2015 Apr,135(4):1074-9.

Law MH et al. PARP1 polymorphisms play opposing roles in melanoma occurrence and survival. Int J Cancer. 2015 May 15,136(10):2488-9.

PIGU - Phosphatidylinositol glycan anchor biosynthesis class U (rs910873)

PIGU is an integral membrane protein that plays a role in cell division control.

RES	Genotype	POP	Possible results
	G/G	97%	No increased risk of melanoma No increased risk of non-melanoma skin cancer
	G/A	2%	Increased risk of melanoma (OR: 1.81) Increased risk of non-melanoma skin cancer (OR: 1.35)
X	A/A	1%	Increased risk of melanoma (OR: 1.81) Increased risk of non-melanoma skin cancer (OR: 1.82)

References

Lin W et al. ASIP genetic variants and the number of non-melanoma skin cancers. Cancer Causes Control. 2011 Mar,22(3):495-501. doi: 10.1007/s10552-010-9724-1. Epub 2011 Jan 9.

Nan H et al. Melanoma susceptibility variants on chromosome 20q11.22 are associated with pigmentary traits and the risk of nonmelanoma skin cancer. Br J Dermatol. 2010 Feb 1,162(2):461-3. doi: 10.1111/j.1365-2133.2009.09579.x. Epub 2009 Nov 30.

Brown KM et al. Common sequence variants on 20q11.22 confer melanoma susceptibility. Nat Genet. 2008 Jul,40(7):838-40. doi: 10.1038/ng.163. Epub 2008 May 18.

Debniak T et al. Modest association of malignant melanoma with the rs910873 and rs1885120 markers on chromosome 20: a population-based study. Melanoma Res. 2010 Apr,20(2):159-60.

Chatzinasiou F et al. Comprehensive field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma. J Natl Cancer Inst. 2011 Aug 17,103(16):1227-35.

SLC45A2 - Solute carrier family 45 member 2 (rs16891982)

The SLC45A2 gene (also called MATP) provides instructions for making a protein that mediates melanin synthesis and has been found to play a role in pigmentation. It is also a melanocyte differentiation antigen that is expressed in a high percentage of melanoma cell lines.

RES	Genotype	POP	Possible results
X	G/G	22%	Protection against melanoma (OR: 0.18)
	G/C	11%	Protection against melanoma (OR: 0.42)
	C/C	67%	No protection against melanoma

References

Stefanaki I et al. Replication and predictive value of SNPs associated with melanoma and pigmentation traits in a Southern European case-control study. PLoS One. 2013,8(2):e55712. doi: 10.1371/journal.pone.0055712. Epub 2013 Feb 5.

Fernandez LP et al. SLC45A2: a novel malignant melanoma-associated gene. Hum Mutat. 2008 Sep,29(9):1161-7.

Duffy DL et al. Multiple pigmentation gene polymorphisms account for a substantial proportion of risk of cutaneous malignant melanoma. J Invest Dermatol. 2010 Feb,130(2):520-8.

López S et al. The interplay between natural selection and susceptibility to melanoma on allele 374F of SLC45A2 gene in a South European population. PLoS One. 2014 Aug 5,9(8):e104367.

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Kosiniak-Kamysz A et al. Increased risk of developing cutaneous malignant melanoma is associated with variation in pigmentation genes and VDR, and may involve epistatic effects. Melanoma Res. 2014 Aug,24(4):388-96.

Guedj M et al. Variants of the MATP/SLC45A2 gene are protective for melanoma in the French population. Hum Mutat. 2008 Sep,29(9):1154-60.

Ibarrola-Villava M et al. MC1R, SLC45A2 and TYR genetic variants involved in melanoma susceptibility in southern European populations: results from a meta-analysis. Eur J Cancer. 2012 Sep,48(14):2183-91.

Antonopoulou K et al. Updated field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma: the MelGene database. J Invest Dermatol. 2015 Apr,135(4):1074-9.

CLPTM1L - Cleft lip and palate transmembrane protein 1-like (rs401681)

CLPTM1L is a membrane protein associated with cisplatin-induced apoptosis. Polymorphisms in this gene have been associated with increased susceptibility to several cancers.

RES	Genotype	POP	Possible results
	C/C	36%	Protection against melanoma (OR: 0.74) Increased risk of non-melanoma skin cancer (OR: 1.28)
X	C/T	45%	Protection against melanoma (OR: 0.86) Increased risk of non-melanoma skin cancer (OR: 1.13)
	T/T	19%	No protection against melanoma No increased risk of non-melanoma skin cancer

References

Rafnar T et al. Sequence variants at the TERT-CLPTM1L locus associate with many cancer types. Nat Genet. 2009 Feb,41(2):221-7.

Stacey SN et al. New common variants affecting susceptibility to basal cell carcinoma. Nat Genet. 2009 Aug,41(8):909-14.

Nan H et al. Genetic variants in telomere-maintaining genes and skin cancer risk. Hum Genet. 2011 Mar,129(3):247-53.

Yang X et al. Association between TERT-CLPTM1L rs401681[C] allele and NMSC cancer risk: a meta-analysis including 45,184 subjects. Arch Dermatol Res. 2013 Jan,305(1):49-52.

TYR - Tyrosinase (rs1393350)

Tyrosinase catalyzes several steps in the conversion of tyrosine to melanin and other pigments. Polymorphisms in this gene result in skin pigmentation variation.

RES	Genotype	POP	Possible results
	G/G	85%	No increased risk of melanoma
X	G/A	14%	Increased risk of melanoma (OR: 1.21)
	A/A	1%	SP - Increased risk of melanoma (OR: 1.80)

References

Bishop DT et al. Genome-wide association study identifies three loci associated with melanoma risk. Nat Genet. 2009 Aug,41(8):920-5. doi: 10.1038/ng.411. Epub 2009 Jul 5.

Chatzinasiou F et al. Comprehensive field synopsis and systematic meta-analyses of genetic association studies in cutaneous melanoma. J Natl Cancer Inst. 2011 Aug 17,103(16):1227-35.

Kosiniak-Kamysz A et al. Increased risk of developing cutaneous malignant melanoma is associated with variation in pigmentation genes and VDR, and may involve epistatic effects. Melanoma Res. 2014 Aug,24(4):388-96.

MC1R - Melanocortin 1 receptor

The MC1R gene provides instructions for making a protein called the melanocortin 1 receptor. The G protein–coupled receptor is located on the surface of melanocytes which produce the pigment melanin through the process of melanogenesis. The MC1R score can be calculated based on the classification of MC1R variants, as implemented by Davies et al.

RES	Genotype	POP	Possible results
X	Score 0	31%	No increased risk of melanoma No increased risk of non-melanoma skin cancer
	Score 1	25%	Increased risk of melanoma (OR: 1.24) Increased risk of non-melanoma skin cancer (OR: 1.41)
	Score 2	27%	Increased risk of melanoma (OR: 1.69) Increased risk of non-melanoma skin cancer (OR: 1.81)
	Score 3	11%	Increased risk of melanoma (OR: 3.28) Increased risk of non-melanoma skin cancer (OR: 2.68)
	Score 4	5%	Increased risk of melanoma (OR: 3.12) Increased risk of non-melanoma skin cancer (OR: 2.68)

References

Pasquali E et al. MC1R variants increased the risk of sporadic cutaneous melanoma in darker-pigmented Caucasians: a pooled-analysis from the M-SKIP project. Int J Cancer. 2015 Feb 1,136(3):618-31.

Williams PF et al. Melanocortin 1 receptor and risk of cutaneous melanoma: a meta-analysis and estimates of population burden. Int J Cancer. 2011 Oct 1,129(7):1730-40.

Amos CI et al. Genome-wide association study identifies novel loci predisposing to cutaneous melanoma. Hum Mol Genet. 2011 Dec 15,20(24):5012-23.

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Kanetsky PA et al. Does MC1R genotype convey information about melanoma risk beyond risk phenotypes? Cancer. 2010 May 15,116(10):2416-28.

Raimondi S et al. MC1R variants, melanoma and red hair color phenotype: a meta-analysis. Int J Cancer. 2008 Jun 15,122(12):2753-60.

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Ferrucci LM et al. Host phenotype characteristics and MC1R in relation to early-onset basal cell carcinoma. J Invest Dermatol. 2012 Apr,132(4):1272-9.

Davies JR et al. Inherited variants in the MC1R gene and survival from cutaneous melanoma: a BioGenoMEL study. Pigment Cell Melanoma Res. 2012 May,25(3):384-94.

Scherer D et al. MC1R variants associated susceptibility to basal cell carcinoma of skin: interaction with host factors and XRCC3 polymorphism. Int J Cancer. 2008 Apr 15,122(8):1787-93.

Dwyer T et al. Does the addition of information on genotype improve prediction of the risk of melanoma and nonmelanoma skin cancer beyond that obtained from skin phenotype? Am J Epidemiol. 2004 May 1,159(9):826-33.

ASIP - Agouti signaling protein (rs4911414/rs1015362)

The agouti-signaling protein is encoded by the agouti gene and influences the distribution of black and yellow pigments in the skin and hair. A haplotype (rs4911414 T and rs1015362 G) in the proximity of ASIP is not only associated with pigmentation features, but is also involved in the development of skin cancer.

RES	Haplotype	POP	Possible results
X	-	99%	**No increased risk of melanoma** **No increased risk of non-melanoma skin cancer**
	GG_TT	1%	**Increased risk of melanoma (OR: 1.5)** **Increased risk of non-melanoma skin cancer (OR: 1.45)**

References

Gudbjartsson DF et al. ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. Nat Genet. 2008 Jul,40(7):886-91. doi: 10.1038/ng.161. Epub 2008 May 18.

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Nan H et al. Genetic variants in pigmentation genes, pigmentary phenotypes, and risk of skin cancer in Caucasians. Int J Cancer. 2009 Aug 15,125(4):909-17.

Helsing P et al. MC1R, ASIP, TYR, and TYRP1 gene variants in a population-based series of multiple primary melanomas. Genes Chromosomes Cancer. 2012 Jul,51(7):654-61.

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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.

PHARMACO GENETICS
Not ordered

ONCOLOGY

CARDIOVASCULAR SYSTEM
Not ordered

NEUROLOGY
Not ordered

METABOLISM
Not ordered

MOVEMENT
Not ordered

DIGESTION
Not ordered

OPHTHALMOLOGY
Not ordered

ODONTOLOGY
Not ordered

OTHERS
Not ordered

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
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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

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Product codes
M7SKI

Current version
V538

Ordering company
ProGenom GmbH
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6343 Rotkreuz
SWITZERLAND

Analyzing company
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83395 Freilassing
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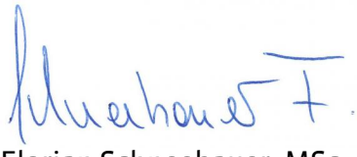
Laboratory Director

Dr. Daniel Wallerstorfer Bsc.

Laboratory Manager



Florian Schneebauer, MSc.

NOTES: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ProMedicDNA

Skin Health Sensor

Jane Doe
DEMO_DS

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