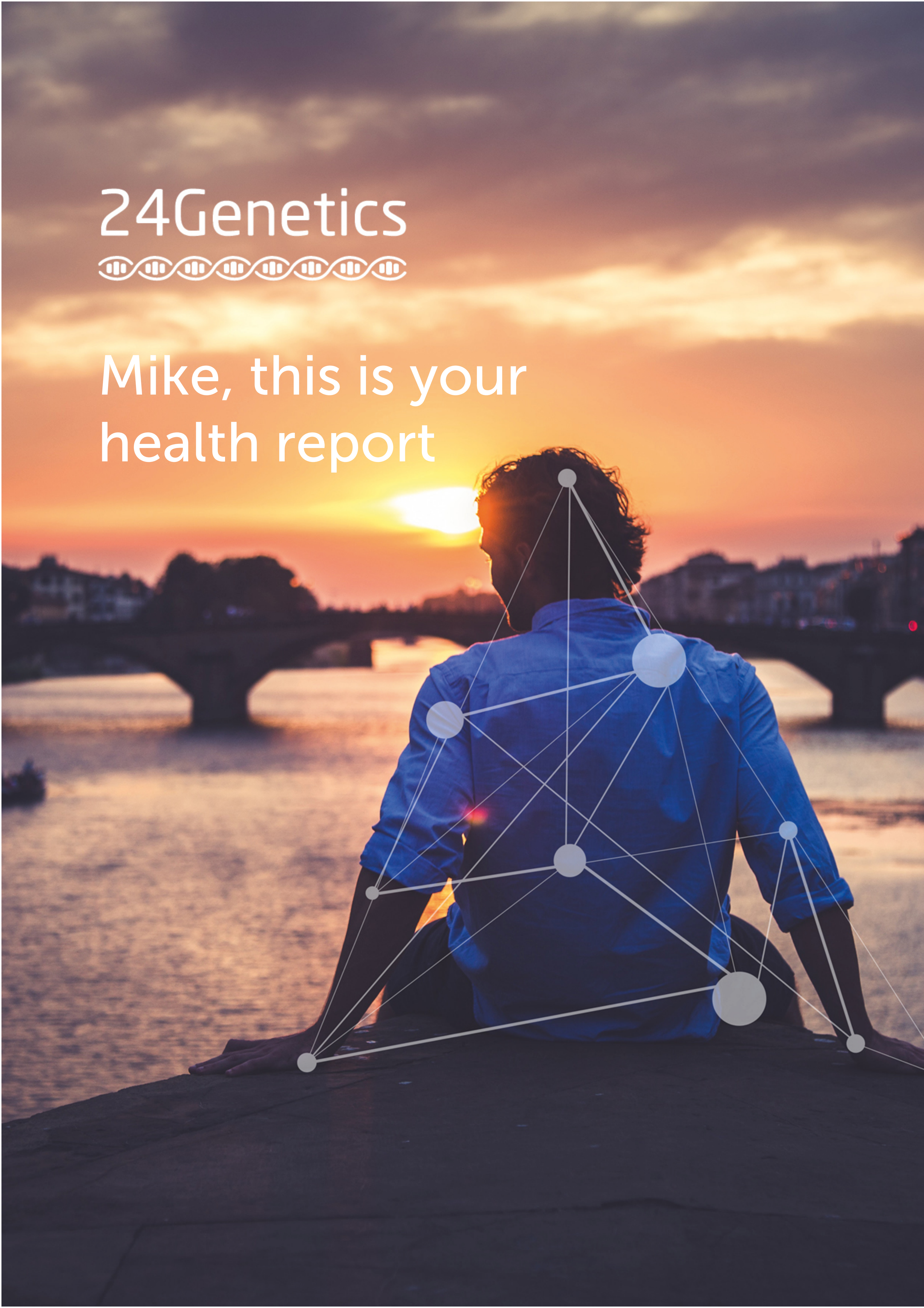
24Genetics

Mike, this is your health report



Index

1. Introduction

In this report you will see some of your genetic predispositions related to health.

As usual in our studies, in the first pages you will find an iconographic summary of each of the analyzed values, which we develop more broadly in later pages.

The report is organized in these sections.

1.1. Methodology that we use for your report

Genetic Health Risks: Gwas

In this part we apply GWAS publications, a type of study that compares the DNA markers of people with a disease or trait, to people without this disease or traits. These studies can be very useful for prevention and early diagnosis; it is not a diagnostic tool but it helps us to see in what we have to care a bit more.

The data that will give us, when applying these studies to your genetic information, is your predisposition comparing to the rest of the population. At no time does it mean that you are going to suffer the disease, it only indicates that statistically and according to this study you could have some more predisposition than the majority of the population; we indicate that you have greater predisposition when it is greater than ninety percent of the population, and smaller if your predisposition is less than ninety percent of the population.

It is important to keep in mind that complex diseases are influenced by many factors; genetic ones are only a part; lifestyle, food, etc. are in many cases the most influential factors

Genetic Health Risk: Mutations

In this section we analyze the mutations of the most important genes from the oncological point of view. We look for mutations suspected of being pathogenic, specifically those reported as pathogenic in the ClinVar database.

It is important to note that this test does not sequence the entire genome, we only analyze 700,000 of the 3.2 billion genetic links so, in the case that we do not find any mutation, that does not mean that we are not carriers, since it can be in genetic regions that we are not analyzing. In this section we analyze a small percentage of the genes classified as pathogenic in the consulted databases, so there could be pathogenic mutations in a region that we can not see in this test.

Carrier Status

Hereditary diseases are likely to be passed on to your offspring. In most of the cases we can be carriers and never suffer the disease, but there is a risk that our offspring will suffer if certain conditions. They are mostly monogenic diseases.

In this group we are looking for pathogenic mutations, or likely pathogenic mutations, in the genes involved in these diseases. We look for the mutations that are reported in some of the most important genetic databases worldwide, basically OMIM and ClinVar.

As in the previous section, we do not analyze all the genetic information related to each disease, specifically in this section we were able to analyze on average something less than half of the pathogenic markers reported in the databases consulted (ClinVar), so we could have mutations in the other half and not see them in this report.

If you need a diagnosis of a particular disease, there are genetic tests, which analyze the entire gene or genes involved in given disease, and they are valid for clinical use. If you have a family background related to a disease we recommend that you go to your doctor or geneticist to study the need for this type of test. The results of this report are personal, not applicable to studies on other members of your family.

Biomarkers, biometrics and traits

In this section we use, again, the GWAS statistical analysis to calculate your genetic predisposition to have abnormal levels of certain metabolic parameters.

As in the rest of our GWAS studies, we indicate that you have a greater predisposition when it is greater than ninety percent of the population, and lower if your predisposition is lower than that of ninety percent of the population. Due to the statistical distribution of this analysis, it is normal that several parameters will appear as high or low predisposition.

Pharmacogenomics

In this section we study your genetic predisposition towards certain medications. Depending on the drug, your genetics can affect the level of toxicity, the effectiveness of ore dose needed. Something that a doctor always has to supervise.

The results of this report are personal, and not applicable to studies about other members of your family.

These reports, as well as the scientific research in the genetics field, may vary over time. New mutations are constantly being discovered and we know better the ones we are analyzing today. At 24Genetics we make a great effort to periodically apply the consolidated scientific discoveries to our reports.

We remind you that any changes you want to make regarding your health should be guided by your doctor. From 24Genetics we recommend all our clients to get a Genetic Counseling service to ensure a better understanding of this genetic report.

1.2. Frequently Asqued Questions

If this report shows that I have a genetic predisposition to a specific disease, am I going to suffer it for sure?

Not at all, the genetic reports that we do are based on statistics. You may have genetic predisposition to a particular disease and never develop it, actually it is what it happends in most of the cases . Or you may not have a predisposition to a disease and suffer it in the future. Genetic analysis is just one more tool, the doctors and specialized health proffesionals the ones who must make the interpretation of the available set of health data.

Should I make drastic changes in my health management with the data of this test?

No at all, any changes you want to make in your health management should be analyzed by an expert geneticist and the medical specialists. Any doubts you have about any genetic test should be checked by healthcare experts in Genetic Diagnosis.

Does it all depend on my genes?

No at all, our body responds to many conditions. Our genes are certainly an important parameter. Lifestyle, sport, food, and many other circumstances influence our body. Knowing yourself certainly helps to treat our body in the most appropriate way. And this is what these genetic reports aren all about: more information.

Are all the analyzed genes listed in the sections?

We include most of the genes we analyze; in some sections we are analyzing more genes that we can't show due the to lack of space.

What is this report based on?

This test is based on different genetic studies internationally consolidated and accepted by the scientific community. There are certain scientific databases where studies are published where there is a certain level of consensus. Our genetic tests are carried out by applying these studies to the genotype of our clients. In each section you will see some of the studies publications on which it is based. There are sections where more studies are used than the ones listed.

If the report reflects that I have genetic mutations in an inherited disease, does that mean that I have that disease for sure?

No, we look for both pathogenic mutations and mutations that could be pathogenic (likely pathogenic); if you have any of these your report will indicate that we have detected it. On the other hand, this technology has a reliability greater than 99% but there is no 100% reliability in this type of genotyping technology. If you have any doubt you should talk to your doctor or geneticist.

If the report reflects that I DO NOT have genetic mutations in an inherited disease, does that mean I am free of this disease for sure?

No, our test does not analyze all the genetic zones where pathogenic mutations may exist and we do not analyze the deletions, duplications or intergenic zones. We analyze only some markers reported as pathogenic. On average our test covers just under 50% of these markers for a given disease, so there could be pathogenic markers in the other half and we would not be seeing them. There are diagnostic tests with greater coverage in certain pathologies that are valid for clinical use. If you have any doubt you should talk to your doctor or geneticist.

If I am a carrier of a mutation of a hereditary disease, how does that affect my offspring?

Almost all of us are carriers of some mutations of monogenetic diseases, it is normal to find in a person between 5 and 50 significant genetic mutations. However, the risk that our offspring suffer the disease varies greatly depending on the type of inheritance: autosomal dominant, autosomal recessive, multifactorial ..., therefore we advise always to go to your doctor or geneticist. for advice.

2. Summary

Genetic Health Risks: Gwas

- ⚪ Alopecia areata
- 🟢 Rheumatoid arthritis
- ⚪ Prostate cancer
- ⚪ Prostate cancer (early onset)
- 🟠 Upper aerodigestive tract cancers
- ⚪ Motion sickness
- ⚪ Age-related macular degeneration
- ⚪ Type 1 diabetes
- ⚪ Type 2 diabetes
- 🟠 Alzheimer's disease (late onset)
- ⚪ Parkinson's disease
- ⚪ Systemic sclerosis
- 🟠 Glioma
- ⚪ Myocardial infarction (early onset)
- ⚪ Hodgkin's lymphoma
- ⚪ Follicular lymphoma
- ⚪ Multiple myeloma
- 🟠 Osteosarcoma
- ⚪ Allergic sensitization
- ⚪ Wilms tumor
- ⚪ Intracranial aneurysm
- ⚪ Chronic bronchitis and chronic obstructive pulmonary disease
- ⚪ Prostate cancer aggressiveness
- ⚪ Bladder cancer
- ⚪ Basal cell carcinoma
- ⚪ Primary biliary cirrhosis
- ⚪ Conduct disorder
- ⚪ Type 1 diabetes nephropathy
- 🟠 Celiac disease
- ⚪ Coronary heart disease
- ⚪ Multiple sclerosis
- ⚪ Schizophrenia
- ⚪ Hypothyroidism
- ⚪ Chronic lymphocytic leukemia
- ⚪ Diffuse large B cell lymphoma
- ⚪ Myasthenia gravis
- ⚪ Neuroblastoma
- 🟢 Psoriasis
- ⚪ Testicular germ cell tumor
- ⚪ Vitiligo

Caption:

- ⚪ According to this study, you have a predisposition similar to most of the population.
- 🟢 According to this study, you are less likely to suffer from this disease than the majority of the population.
- 🟠 According to this study, you are more likely to suffer from this disease than most of the population.

Genetic Health Risks: mutations

- ⚪ APC: colorectal and pancreatic cancer
- ⚪ BRCA1: breast and ovarian cancer
- ⚪ BRIP1: breast cancer
- ⚪ CDKN2A: pancreatic cancer
- ⚪ MLH1: Lynch syndrome
- ⚪ MSH6: Lynch syndrome and colorectal cancer
- ⚪ PALB2: breast and pancreatic cancer
- ⚪ ATM: breast cancer
- ⚪ BRCA2: breast and ovarian cancer
- ⚪ CDH1: breast and gastric cancer
- ⚪ CHEK2: breast and colorectal cancer
- ⚪ MSH2: Lynch syndrome and colorectal cancer
- ⚪ MUTYH: MYH-associated polyposis and colorectal cancer
- ⚪ PMS2: Lynch syndrome and colorectal cancer

- ● PTEN: breast, uterine and colorectal cancer
- ● SMAD4: juvenile polyposis syndrome and colorectal cancer
- ● VHL: Von Hippel-Lindau syndrome
- ● SDHB: gastric cancer
- ● TP53: Li-Fraumeni syndrome, breast cancer and more
- ● RET: thyroid carcinoma

Caption:

- ● We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions.
- 🔴 We have detected at least one mutation that could be pathogenic.

Carrier Status

- ● 17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY
- ● AARSKOG-SCOTT SYNDROME; AAS
- ● LEUKEMIA, ACUTE MYELOID; AML
- ● HYPOPHOSPHATASIA, ADULT
- ● ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD
- ● ANEMIA, NONSPHEROCYTIC HEMOLYTIC, DUE TO G6PD DEFICIENCY
- ● ANTITHROMBIN III DEFICIENCY; AT3D
- ● AURICULOCONDYLAR SYNDROME 1; ARCND1
- ● BARDET-BIEDL SYNDROME 1; BBS1
- ● BETA-THALASSEMIA
- ● BRUGADA SYNDROME 1; BRGDA1
- ● CARDIOMYOPATHY, DILATED, 1S; CMD1S
- ● CEROID LIPOFUSCINOSIS, NEURONAL, 1; CLN1
- ● CHARCOT-MARIE-TOOTH DISEASE, TYPE 4C; CMT4C
- ● GRANULOMATOUS DISEASE, CHRONIC, X-LINKED; CDGX
- ● NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C; CSNB1C
- ● COSTELLO SYNDROME; CSTLO
- ● DANON DISEASE
- ● 3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY; MCC2D
- ● ACHROMATOPSIA 2; ACHM2
- ● ADRENOLEUKODYSTROPHY; ALD
- ● ALLAN-HERNDON-DUDLEY SYNDROME; AHDS
- ● AMYLOIDOSIS, HEREDITARY, TRANSTHYRETIN-RELATED
- ● ANGELMAN SYNDROME; AS
- ● ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 10; ARVD10
- ● HYPOPHOSPHATEMIC RICKETS, AUTOSOMAL DOMINANT; ADHR
- ● MUSCULAR DYSTROPHY, BECKER TYPE; BMD
- ● BLOOM SYNDROME; BLM
- ● CARDIOFACIOCUTANEOUS SYNDROME 1; CFC1
- ● CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 1; CMH1
- ● CEROID LIPOFUSCINOSIS, NEURONAL, 7; CLN7
- ● CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1
- ● ADRENAL HYPOPLASIA, CONGENITAL; AHC
- ● CORNELIA DE LANGE SYNDROME 1; CDLS1
- ● CYSTIC FIBROSIS; CF
- ● DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A

- DEAFNESS, AUTOSOMAL RECESSIVE 31; DFNB31
- DEAFNESS, AUTOSOMAL RECESSIVE 7; DFNB7
- DEAFNESS, AUTOSOMAL RECESSIVE 9; DFNB9
- MANNOSIDOSIS, ALPHA B, LYSOSOMAL; MANSA
- CARDIOMYOPATHY, DILATED, 1A; CMD1A
- DUBIN-JOHNSON SYNDROME; DJS
- EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 2; EIEE2
- MYOCLONIC EPILEPSY OF LAFORA
- ERYTHROCYTOSIS, FAMILIAL, 2; ECYT2
- FABRY DISEASE
- FAMILIAL ADENOMATOUS POLYPOSIS 1; FAP1
- CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 2; CMH2
- FAMILIAL MEDITERRANEAN FEVER; FMF
- THYROID CARCINOMA, FAMILIAL MEDULLARY; MTC
- FANCONI ANEMIA, COMPLEMENTATION GROUP O; FANCO
- NEPHROTIC SYNDROME, TYPE 1; NPHS1
- GAUCHER DISEASE, TYPE I
- GLUT1 DEFICIENCY SYNDROME 1; GLUT1DS1
- GLUTARIC ACIDEMIA I; GA1
- MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD
- GLYCOGEN STORAGE DISEASE Ia; GSD1A
- GLYCOGEN STORAGE DISEASE II; GSD2
- HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2; FHL2
- HERMANSKY-PUDLAK SYNDROME 3; HPS3
- HISTIOCYTOSIS-LYMPHADENOPATHY PLUS SYNDROME
- ECTODERMAL DYSPLASIA 1, HYPOHIDROTIC, X-LINKED; XHED
- JERVELL AND LANGE-NIELSEN SYNDROME 1; JLNS1
- JOUBERT SYNDROME 14; JBTS14
- JOUBERT SYNDROME 16; JBTS16
- JOUBERT SYNDROME 3; JBTS3
- JOUBERT SYNDROME 5; JBTS5
- JOUBERT SYNDROME 7; JBTS7
- JOUBERT SYNDROME 8; JBTS8
- JOUBERT SYNDROME 9; JBTS9
- KABUKI SYNDROME 1; KABUK1
- LEIGH SYNDROME; LS
- LEOPARD SYNDROME 1; LPRD1
- LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM
- LISSENCEPHALY 1; LIS1
- LOEYS-DIETZ SYNDROME 2; LDS2
- LONG QT SYNDROME 1; LQT1
- MAPLE SYRUP URINE DISEASE; MSUD
- MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 2; MODY2
- MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3; MODY3
- MECKEL SYNDROME, TYPE 3; MKS3
- MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA; MICPCH
- METACHROMATIC LEUKODYSTROPHY; MLD
- METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE
- METHYLMALONIC ACIDURIA, cblA TYPE
- METHYLMALONIC ACIDURIA, cblB TYPE

This report is not valid for clinical or diagnostic use.

- ⚪ MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 1; MC3DN1
- ⚪ MUCOPOLYSACCHARIDOSIS TYPE VI; MPS6
- ⚪ MUCOPOLYSACCHARIDOSIS, TYPE VII; MPS7
- ⚪ MUCOPOLYSACCHARIDOSIS, TYPE IIIA; MPS3A
- ⚪ MUCOPOLYSACCHARIDOSIS, TYPE IIIB; MPS3B
- ⚪ MUCOPOLYSACCHARIDOSIS, TYPE IVA; MPS4A
- ⚪ MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES), TYPE A, 1; MDDGA1
- ⚪ MYOPATHY, MYOFIBRILLAR, 1; MFM1
- ⚪ MYOPATHY, CENTRONUCLEAR, 1; CNM1
- ⚪ MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX
- ⚪ NEMALINE MYOPATHY 2; NEM2
- ⚪ CYSTINOSIS, NEPHROPATHIC; CTNS
- ⚪ NIEMANN-PICK DISEASE, TYPE C1; NPC1
- ⚪ NIEMANN-PICK DISEASE, TYPE A
- ⚪ NIEMANN-PICK DISEASE, TYPE B
- ⚪ NOONAN SYNDROME 1; NS1
- ⚪ NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MYELOMONOCYTIC LEUKEMIA; NSLL
- ⚪ NOONAN SYNDROME 4; NS4
- ⚪ Obesity due to melanocortin 4 receptor deficiency
- ⚪ ALBINISM, OCULOCUTANEOUS, TYPE IB; OCA1B
- ⚪ OSTEOGENESIS IMPERFECTA, TYPE III; OI3
- ⚪ DIABETES MELLITUS, PERMANENT NEONATAL; PNDM
- ⚪ PITT-HOPKINS SYNDROME; PTHS
- ⚪ POLYMICROGYRIA, BILATERAL FRONTOPARIETAL; BFPP
- ⚪ MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE; MCPH5
- ⚪ RETINITIS PIGMENTOSA; RP
- ⚪ RUBINSTEIN-TAYBI SYNDROME 1; RSTS1
- ⚪ SOTOS SYNDROME 1; SOTOS1
- ⚪ SUPRAVALVULAR AORTIC STENOSIS; SVAS
- ⚪ TAY-SACHS DISEASE; TSD
- ⚪ TUBEROUS SCLEROSIS 1; TSC1
- ⚪ TUBEROUS SCLEROSIS 2; TSC2
- ⚪ ALBINISM, OCULOCUTANEOUS, TYPE IA; OCA1A
- ⚪ TYROSINEMIA, TYPE I; TYRSN1
- ⚪ USHER SYNDROME, TYPE I; USH1
- ⚪ USHER SYNDROME, TYPE ID; USH1D
- ⚪ USHER SYNDROME, TYPE IF; USH1F
- ⚪ USHER SYNDROME, TYPE IIA; USH2A
- ⚪ USHER SYNDROME, TYPE IIC; USH2C
- ⚪ USHER SYNDROME, TYPE IID; USH2D
- ⚪ USHER SYNDROME, TYPE IIIA; USH3A
- ⚪ ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD
- ⚪ VON HIPPEL-LINDAU SYNDROME; VHL
- ⚪ WEAVER SYNDROME; WVS
- ⚪ Wilson Disease
- ⚪ AGAMMAGLOBULINEMIA, X-LINKED; XLA

Caption:

⚪ We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions.
🔴 We have detected at least one mutation that could be pathogenic.

Biomarkers

- ⚪ Adiponectin levels
- ⚪ Androgen levels in men
- ⚪ Beta-2 microglubulin plasma levels
- 🟠 Bilirubin levels
- 🟢 C-reactive protein and white blood cell count
- 🟠 Calcium levels
- ⚪ Dehydroepiandrosterone sulphate levels
- ⚪ Eosinophil counts
- ⚪ Glycated hemoglobin levels
- ⚪ Homocysteine levels
- ⚪ IgE levels
- 🟠 Liver enzyme levels (gamma-glutamyl transferase)
- 🟢 Liver enzyme levels
- ⚪ Magnesium levels
- ⚪ Monocyte count
- 🟢 Phospholipid levels (plasma)
- ⚪ Phosphorus levels
- 🟠 Plasma omega-6 polyunsaturated fatty acid levels (dihomo-gamma-linolenic acid)
- ⚪ Platelet count
- 🟢 Red blood cell count
- ⚪ Serum albumin level
- 🟢 Sex hormone levels
- ⚪ Thyroid hormone levels
- ⚪ Uric acid levels
- ⚪ Urinary uromodulin levels
- ⚪ Vitamin B levels in ischemic stroke
- 🟢 White blood cell count

Caption:

- ⚪ According to this study, you have a similar predisposition to the majority of the population to have normal levels.
- 🟢 According to this study, you have a better predisposition than the majority of the population to have normal levels.
- 🟠 According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

Biometrics

- 🟢 Aortic root size
- ⚪ Bone mineral density
- ⚪ Heart rate
- ⚪ Resting heart rate

Caption:

- ⚪ According to this study, you have a similar predisposition to the majority of the population to have normal levels.
- 🟢 According to this study, you have a better predisposition than the majority of the population to have normal levels.
- 🟠 According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

Traits

- ⚪ Alcoholism (alcohol dependence factor score)
- ⚪ Spirometric measure of pulmonary function (Forced vital capacity)
- ⚪ Smoking behavior

Caption:

- ⚪ According to this study, you have a predisposition similar to most of the population.
- 🟢 According to this study, you have less predisposition than the majority of the population.
- 🟠 According to this study, you have a greater predisposition than the majority of the population.

Pharmacogenomics: Cardiology

- 🟢 Pravastatin
- ⚪ Simvastatin

- 🟣 Warfarin

Caption:

- ⚪ We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟣 According to your genotype you are more predisposed to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🔴 According to your genotype you are more predisposed to have harmful effects on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟢 According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

Pharmacogenomics: Neurology

- 🟣 Amitriptyline
- 🟢 Bupropion
- 🟣 Antidepressants

Caption:

- ⚪ We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟣 According to your genotype you are more predisposed to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🔴 According to your genotype you are more predisposed to have harmful effects on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟢 According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

Pharmacogenomics: Oncology

- 🟣 Methotrexate
- ⚪ Fluorouracil, capecitabine, pyrimidine analogues, tegafur and Neoplasms
- 🔴 Vincristine

Caption:

- ⚪ We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟣 According to your genotype you are more predisposed to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🔴 According to your genotype you are more predisposed to have harmful effects on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟢 According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

Pharmacogenomics: Other

- 🟣 Tacrolimus
- 🟣 Sildenfail (Viagra)

Caption:

- ⚪ We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟣 According to your genotype you are more predisposed to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🔴 According to your genotype you are more predisposed to have harmful effects on you. Other non-analyzed and non-genetic genetic factors may play a role.
- 🟢 According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

Pharmacogenomics: Pain

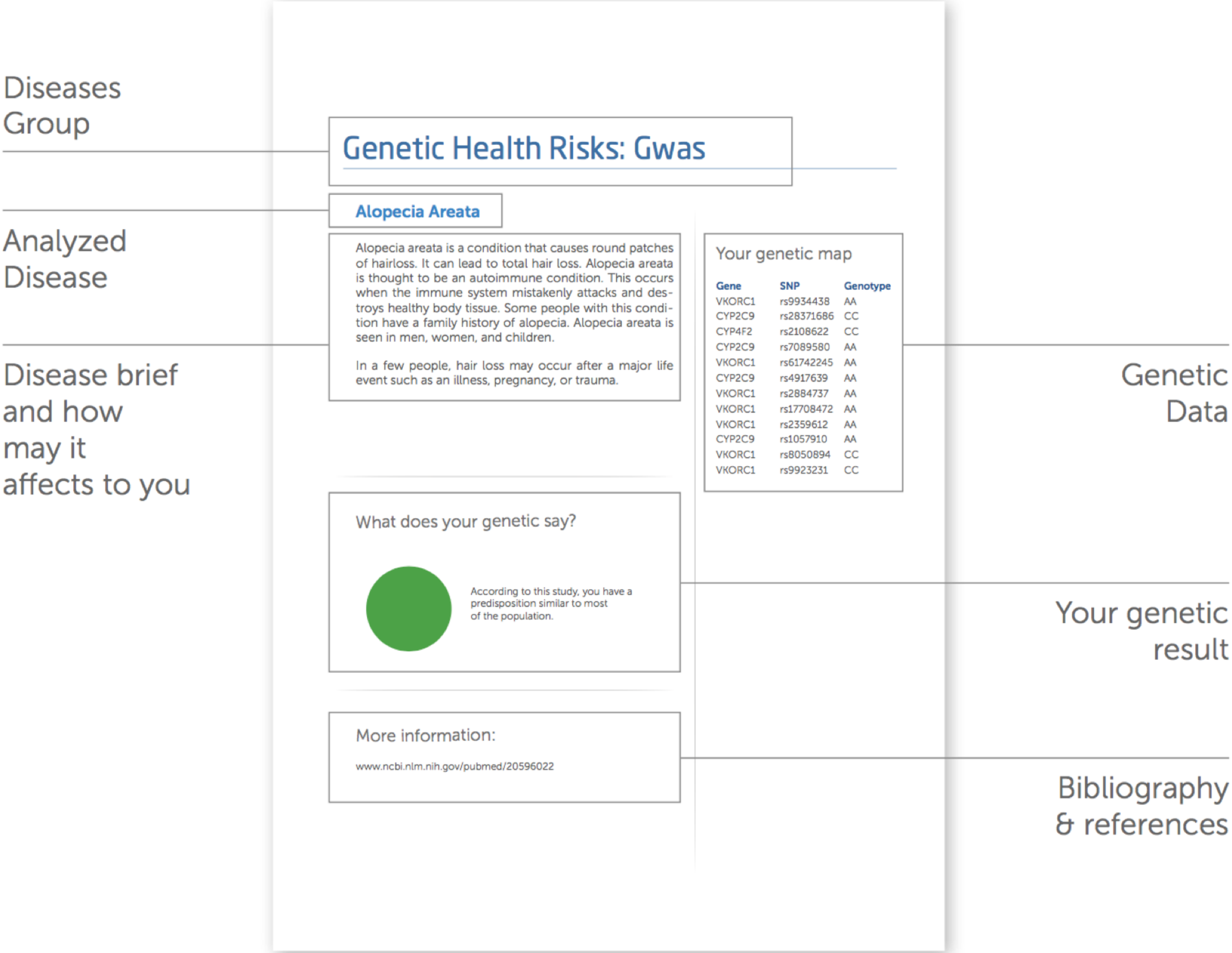
- 🟣 Meperidine
- 🟣 Pentazocine
- 🟣 Morphine
- ⚪ Aspirin

Caption:

- ● We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- ● According to your genotype you are more predisposed to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- ● According to your genotype you are more predisposed to have harmful effects on you. Other non-analyzed and non-genetic genetic factors may play a role.
- ● According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

3. Genetic Results

3.1. What information is included in the results?



Genetic Health Risks: Gwas

Alopecia Areata

Alopecia areata is a condition that causes round patches of hairloss. It can lead to total hair loss. Alopecia areata is thought to be an autoimmune condition. This occurs when the immune system mistakenly attacks and destroys healthy body tissue. Some people with this condition have a family history of alopecia. Alopecia areata is seen in men, women, and children.

In a few people, hair loss may occur after a major life event such as an illness, pregnancy, or trauma.

Your genetic map

Gene	SNP	Genotype
VKORC1	rs9934438	AA
CYP2C9	rs28371686	CC
CYP4F2	rs2108622	CC
CYP2C9	rs7089580	AA
VKORC1	rs61742245	AA
CYP2C9	rs4917639	AA
VKORC1	rs2884737	AA
VKORC1	rs17708472	AA
VKORC1	rs2359612	AA
CYP2C9	rs1057910	AA
VKORC1	rs8050894	CC
VKORC1	rs9923231	CC

What does your genetic say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20596022

3.2. Your genetic results

Genetic Health Risks: Gwas

Alopecia areata

Alopecia areata is a condition that causes round patches of hair loss. It can lead to total hair loss.

Alopecia areata is thought to be an autoimmune condition. This occurs when the immune system mistakenly attacks and destroys healthy body tissue.

Some people with this condition have a family history of alopecia. Alopecia areata is seen in men, women, and children. In a few people, hair loss may occur after a major life event such as an illness, pregnancy, or trauma.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20596022

Your genetic map

Gene	SNP	Genotype
ICOS,	rs1024161	TC
IL2, IL21	rs7682241	TG
ULBP3,	rs9479482	TC
IL2RA	rs3118470	TT
PRDX5	rs694739	GG
IKZF4	rs1701704	TT
HLA-	rs9275572	GG

Genetic Health Risks: Gwas

Intracranial aneurysm

A brain aneurysm is an abnormal bulge or "ballooning" in the wall of an artery in the brain. They are sometimes called berry aneurysms because they are often the size of a small berry. Most brain aneurysms produce no symptoms until they become large, begin to leak blood, or burst.

If a brain aneurysm presses on nerves in your brain, it can cause signs and symptoms.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20364137

Your genetic map

Gene	SNP	Genotype
SOX17	rs9298506	AA
CDKN2A,	rs1333040	CC
CNNM2	rs12413409	GG
STARD13	rs9315204	CC
RBBP8	rs11661542	AC

Genetic Health Risks: Gwas

Rheumatoid arthritis

Rheumatoid arthritis (RA) is a form of arthritis that causes pain, swelling, stiffness and loss of function in your joints. It can affect any joint but is common in the wrist and fingers.

More women than men get rheumatoid arthritis. It often starts in middle age and is most common in older people. You might have the disease for only a short time, or symptoms might come and go. The severe form can last a lifetime.

What does your genetics say?

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According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24390342

Your genetic map

Gene	SNP	Genotype
ACOXL	rs6732565	AA
ACOXL	rs6732565	AA
AFF3	rs9653442	TT
AFF3	rs9653442	TT
ANKRD55	rs7731626	AG
ARID5B	rs71508903	CC
ARID5B	rs71508903	CC
ARID5B	rs71508903	CC
ATG5	rs9372120	TT
ATG5	rs9372120	TT
BLK	rs2736337	TT
BLK	rs2736337	TT
BLK	rs2736337	TT
C1QBP	rs72634030	CC
C4orf52	rs11933540	TT
C5orf30	rs2561477	GG
C5orf30	rs2561477	GG
CCL19,	rs11574914	GG
CCL19,	rs11574914	GG
CCR6	rs1571878	TC
CCR6	rs1571878	TC
CCR6	rs1571878	TC
CD2	rs624988	CC
CD226	rs2469434	TC
CD226	rs2469434	TC
CD28	rs1980422	TT
CD28	rs1980422	TT
CD40	rs4239702	TC
CD40	rs4239702	TC
CDK6	rs4272	AA
CDK6	rs4272	AA

Genetic Health Risks: Gwas

Chronic bronchitis and chronic obstructive pulmonary disease

Chronic obstructive pulmonary disease (COPD) is a common lung disease. Having COPD makes it hard to breathe.
There are two main forms of COPD: Chronic bronchitis, which involves a long-term cough with mucus, and Emphysema, which involves damage to the lungs over time
Most people with COPD have a combination of both conditions. Smoking is the main cause of COPD. The more a person smokes, the more likely that person will develop COPD. But some people smoke for years and never get COPD. In rare cases, nonsmokers who lack a protein called alpha-1 antitrypsin can develop emphysema.

Your genetic map

Gene	SNP	Genotype
FAM13A	rs2869966	TC
IREB2	rs8042238	TC
FAM13A	rs2869967	TC
EFCAB4A	rs34391416	GG
HHIP-AS1	rs13141641	TC
CHRNA3,	rs12914385	CC
FAM13A	rs4416442	TC
CHRNA3,	rs12914385	CC
EFCAB4A	rs34391416	GG
CYS1	rs12692398	AA

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25241909

Genetic Health Risks: Gwas

Prostate cancer

The prostate is the gland below a man's bladder that produces fluid for semen. Prostate cancer is common among older men. It is rare in men younger than 40. Risk factors for developing prostate cancer include being over 65 years of age, high-fat diet, family history, and being African-American. Thanks to the early diagnosis test for blood PSA levels, the survival of men diagnosed with prostate cancer has improved in recent years. It is estimated that 10% of the cases present a hereditary component. Large-scale genetic studies have detected various susceptibility genes.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23535732

Your genetic map

Gene	SNP	Genotype
ADAM15,	rs1218582	GG
LRRN2,	rs4245739	AA
C2orf48,	rs11902236	TC
BOK-AS1,	rs3771570	CC
WDR52-	rs7611694	AC
COX18,	rs1894292	AA
BOD1,	rs6869841	TC
CYP21A2,	rs3096702	GG
LACE1,	rs2273669	AA
RGS17,	rs1933488	AA
SP8,	rs12155172	GG
EBF2	rs11135910	CC
CNNM2,	rs3850699	AA
MMP8,	rs11568818	TT
TBX5,	rs1270884	AG
DDHD1,	rs8008270	CC
RAD51B,	rs7141529	TT
DBIL5P,	rs684232	TC
NGFR,	rs11650494	GG
SALL3,	rs7241993	CC
GTPBP5,	rs2427345	CC
STMN3,	rs6062509	TT
GPR143,	rs2405942	AA

Genetic Health Risks: Gwas

Prostate cancer aggressiveness

Approximately 65% of patients suffering from prostate cancer survive more than 5 years (in developed countries). It is the third leading cause of cancer death in men. The aggressiveness of cancer, that is, tumors that progress and cause death, is partly determined by genetic factors. Large-scale association studies have identified several genes associated with the degree of aggressiveness of the disease.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25939597

Your genetic map

Gene	SNP	Genotype
RASA1,	rs35148638	AA
NAALADL	rs78943174	CC
KLK3	rs62113212	CC
LOC7276	rs4242382	GG
HNF1B	rs8064454	CC
BC03932	rs17765344	AA
BIK	rs5759167	GG
MSMB	rs10993994	TC
PRKCI	rs71277158	TT
LOC3386	rs7929962	TT
SLC22A3	rs7758229	GG
LINC005	rs17023900	AA
TERT,	rs7725218	AG
TBX5	rs10774740	TG
TET2	rs7679673	CC
NR	rs2807031	TT
NR	rs6983267	TG
NR	rs16901979	AC

Genetic Health Risks: Gwas

Prostate cancer (early onset)

Prostate cancer is a disease that primarily affects men who are older. The age of onset of prostate cancer is determined by genetic factors. 75% of the cases are people older than 65 years, although a proportion of cases is diagnosed at an early age. The risk of developing the disease at an age younger than 56 years is determined by genetic variants, as shown by a large-scale association study.

Your genetic map

Gene	SNP	Genotype
NR	rs6983267	TG
MSMB	rs10993994	TC
NR	rs7931342	GG
MYC	rs10505477	AG
KLK3	rs17632542	TT
TH	rs7126629	AA

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24740154

Genetic Health Risks: Gwas

Bladder cancer

Bladder cancer is the fourth most frequently diagnosed in men. It is much more frequent among men, being the proportion of 7 men per woman. The incidence (new cases diagnosed in one year) in our country is the highest in the world: 11% of tumors in men and 2.4% in women. 70-75% of the cases are attributed to tobacco consumption. Another risk factor is urinary tract infection. People with affected relatives are at increased risk of developing this type of tumor, suggesting that there is an underlying genetic factor. In fact, large-scale association studies have found genes predisposing to the disease.

Your genetic map

Gene	SNP	Genotype
intergeni	rs10936599	CC
LSP1	rs907611	AA
C20orf18	rs6104690	GG
NR	rs4907479	GG
UGT1A	rs11892031	AA
TP63	rs710521	TC
TMEM129	rs798766	TC
TERT,	rs401681	TT
NAT2	rs1495741	AG
PSCA	rs2204008	CC
intergeni	rs9642880	GG
SLC14A2	rs10775480	TT
CCNE1	rs8102137	TC
CBX6,	rs1014971	TT

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24163127

Genetic Health Risks: Gwas

Upper aerodigestive tract cancers

Cancer of the upper aerodigestive tract includes tumors of the oral cavity, pharynx, larynx, nasal cavity and paranasal sinuses, ear and salivary glands. Head and neck carcinoma is the most common among them and has a high mortality rate (in Spain it is 37%). Alcohol and tobacco use are the main risk factors, although human papillomavirus infection and family history also play an important role. A large-scale genetic association study has found genetic variants that increase the risk of the disease.

Your genetic map

Gene	SNP	Genotype
ADH1B	rs1229984	CC
ADH7	rs971074	TC
HEL308,	rs1494961	CC
ALDH2	rs4767364	AG

What does your genetics say?

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According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21437268

Genetic Health Risks: Gwas

Basal cell carcinoma

Nonmelanoma type tumors occur in the outermost layer of the epidermis skin, and they are about 95% of the cancers that can appear on the skin. About 20% are squamous carcinomas, which come from the malignization of the squamous cells of the skin. It is among the most common cancers among people of European descent. The main cause of occurrence is DNA damage caused by ultraviolet exposure, although large-scale genetic studies have described genetic variants of predisposition to the disease.

Your genetic map

Gene	SNP	Genotype
MYCN,	rs57244888	TT
ALS2CR1	rs13014235	GG
ZFHX4,	rs28727938	CC
GATA3,	rs73635312	GG
RCC2,	rs7538876	GG
RHOU	rs801114	TG
TERT,	rs401681	TT
KRT5	rs11170164	TC
CDKN2A,	rs2151280	GG
KLF14	rs157935	TT
TP53	rs78378222	TT
TGM3	rs214782	GG
RGS22	rs7006527	AC

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25855136

Genetic Health Risks: Gwas

Motion sickness

Motion sickness is a common problem in people traveling by car, train, airplanes, and especially boats. Anyone can get it, but it is more common in children, pregnant women, and people taking certain medicines. Motion sickness can start suddenly, with a queasy feeling and cold sweats. It can then lead to dizziness and nausea and vomiting. Your brain senses movement by getting signals from your inner ears, eyes, muscles, and joints. When it gets signals that do not match, you can get motion sickness. For example, if you are reading on your phone while riding a bus, your eyes are focused on something that is not moving, but your inner ear senses motion. Despite their high heritability, no associated genetic factors have been discovered. This section is based on a genome association study on motion sickness in 80,494 individuals who were surveyed about this pathology.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25628336

Your genetic map

Gene	SNP	Genotype
PVRL3	rs66800491	GG
GPD2	rs56051278	AG
ACO1	rs10970305	CC
AUTS2	rs1195218	GG
GPR26	rs705145	CC
CBLN4	rs6069325	TT
MUTED	rs2153535	GG
LINGO2	rs2150864	AG
CPNE4	rs9834560	AA
RWDD3	rs1858111	AA
PRDM16	rs61759167	CC
NLGN1	rs11713169	AC
HOXD	rs2551802	GG
COPS8	rs2318131	AA
TLE4	rs149951341	AA
HOXB	rs9906289	CC
ST18	rs2360806	AC
SDK1	rs4343996	AG
NR2F2	rs7170668	TC
CELF2	rs10752212	AA
CNTN1	rs7957589	AA
MCTP2	rs62018380	CC
ARAP2	rs6833641	CC
AUTS2	rs6946969	AG
RGS5	rs4076764	TC
MAP2K5	rs997295	TT
AGA	rs1378552	TT
POU6F2	rs60464047	AT
TUSC1	rs1782032	AG
GXYLT2	rs1847202	TT
SDK1	rs34912216	

Genetic Health Risks: Gwas

Primary biliary cirrhosis

The bile ducts are tubes that move bile from the liver to the small intestine. Bile is a substance that helps with digestion. All of the bile ducts together are called the biliary tract. When the bile ducts become swollen or inflamed, this blocks the flow of bile. The buildup of bile damages the liver cells and leads to scarring of the liver called cirrhosis. This is called biliary cirrhosis.

Genetic susceptibility has been suggested, as well as the influence of environmental factors (infections, smoking, exposure to chemicals).

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21399635

Your genetic map

Gene	SNP	Genotype
DENND1	rs12134279	CC
STAT4	rs10931468	CC
CD80	rs2293370	AG
NFKB1	rs7665090	AG
IL7R	rs860413	AC
ELMO1	rs6974491	GG
CXCR5	rs6421571	CC
TNFRSF1	rs1800693	CC
RAD51L1	rs911263	TT
CLEC16A	rs12924729	GG
intergeni	rs11117432	AG
MAP3K7I	rs968451	GG
IL12A	rs485499	TT
MHC	rs7774434	TC
IRF5	rs12531711	AA
ORMDL3	rs7208487	TG
SPIB	rs3745516	AA
PLCL2	rs1372072	GG
RPS6KA4	rs538147	AG
TNFAIP2	rs8017161	AG

Genetic Health Risks: Gwas

Age-related macular degeneration

Macular degeneration, or age-related macular degeneration (AMD), is a leading cause of vision loss in Americans 60 and older. It is a disease that destroys your sharp, central vision. You need central vision to see objects clearly and to do tasks such as reading and driving. AMD affects the macula, the part of the eye that allows you to see fine detail. It does not hurt, but it causes cells in the macula to die. There are two types: wet and dry. Wet AMD happens when abnormal blood vessels grow under the macula. These new blood vessels often leak blood and fluid. Wet AMD damages the macula quickly. Blurred vision is a common early symptom. Dry AMD happens when the light-sensitive cells in the macula slowly break down. Your gradually lose your central vision. A common early symptom is that straight lines appear crooked.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23455636

Your genetic map

Gene	SNP	Genotype
ARMS2,	rs10490924	TG
CFB, C2	rs429608	GG
C3	rs2230199	CG
APOE	rs4420638	AA
CETP	rs1864163	AA
VEGFA	rs943080	TC
TNFRSF1	rs13278062	TG
LIPC	rs920915	GG
CFI	rs4698775	TT
COL10A1	rs3812111	AT
FILIP1L,	rs13081855	GG
IER3,	rs3130783	AA
SLC16A8	rs8135665	TC
TGFBR1	rs334353	TT
RAD51B	rs8017304	AA
ADAMTS9	rs6795735	TC
B3GALTL	rs9542236	TC

Genetic Health Risks: Gwas

Conduct disorder

Behavioral disorder is one of the most prevalent psychiatric disorders in children. The related symptoms have an important genetic component, whose heritability is estimated at 50%, and include aggression, breaking of rules, harassment of other children, robberies, violence, etc. This disorder is a risk factor for future addictive behavior. Different genetic variants have been associated with the risk of onset of this disorder.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20585324

Your genetic map

Gene	SNP	Genotype
C1QTNF7	rs16891867	AA
PDE10A	rs7762160	TT
TOX2	rs6031252	CC
ERCC4	rs3136202	GG
LOC3430	rs4434872	CC
ARHGAP2	rs10776612	CC
intergeni	rs7950811	CC
intergeni	rs11838918	TT
intergeni	rs1256531	GG
intergeni	rs4792394	CC
intergeni	rs7950811	CC
intergeni	rs11838918	TT
intergeni	rs13398848	AA
intergeni	rs2184898	AG
intergeni	rs1550057	AA
KIAA1345	rs1861050	CC
C1QTNF7	rs16891867	AA
intergeni	rs11838918	TT
intergeni	rs13398848	AA

Genetic Health Risks: Gwas

Type 1 diabetes

Diabetes means your blood glucose, or blood sugar, levels are too high. With type 1 diabetes, your pancreas does not make insulin. Insulin is a hormone that helps glucose get into your cells to give them energy. Without insulin, too much glucose stays in your blood. Over time, high blood glucose can lead to serious problems with your heart, eyes, kidneys, nerves, and gums and teeth.

Type 1 diabetes happens most often in children and young adults but can appear at any age.

Your genetic map

Gene	SNP	Genotype
BACH2	rs11755527	GG
PRKCQ	rs947474	AA
CTSH	rs3825932	TC
C1QTNF6	rs229541	AG
PTPN22	rs6679677	CC
CTLA4	rs3087243	AG
IL2RA	rs12251307	TC
C12orf30	rs17696736	AG
ERBB3	rs2292239	GG
CLEC16A	rs12708716	AG
PTPN2	rs2542151	TT

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/18978792

Genetic Health Risks: Gwas

Type 1 diabetes nephropathy

Type 1 diabetes mellitus (DM1) is an autoimmune and metabolic disease in which the pancreas does not produce insulin, resulting in elevated blood glucose levels. Type 1 diabetes occurs most frequently in children and young adults and accounts for 13% of all cases of diabetes in countries like Spain where the number of cases for children under 15 is 11.5-27.6 cases / 100,000 inhabitants. The susceptibility to type 1 diabetes mellitus appears to be associated with multiple genetic factors, although interaction with certain environmental factors (infections, diet ...) is required for the development of the disease.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23028342

Your genetic map

Gene	SNP	Genotype
MCTP2,	rs12437854	TG
AFF3	rs7583877	TT
AFF3	rs7583877	TT
intergeni	rs878889	GG
RP11	rs4871297	GG
RNF10,	rs614226	CC
intergeni	rs13045180	TC

Genetic Health Risks: Gwas

Type 2 diabetes

Diabetes means your blood glucose, or blood sugar, levels are too high. With type 2 diabetes, the more common type, your body does not make or use insulin well. Insulin is a hormone that helps glucose get into your cells to give them energy. Without insulin, too much glucose stays in your blood. Over time, high blood glucose can lead to serious problems with your heart, eyes, kidneys, nerves, and gums and teeth. You have a higher risk of type 2 diabetes if you are older, obese, have a family history of diabetes, or do not exercise. Having prediabetes also increases your risk. Prediabetes means that your blood sugar is higher than normal but not high enough to be called diabetes.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24509480

Your genetic map

Gene	SNP	Genotype
RREB1,	rs9502570	TC
FAF1	rs17106184	AG
TCF19,	rs3132524	TC
LPP	rs6808574	TC
ARL15	rs702634	AG
MPHOSP	rs1727313	GG
PLEKHA1	rs10510110	TT
TMEM75	rs1561927	CC
VEGFA	rs9472138	TC
ETV1	rs7795991	AG
C6orf173	rs4273712	AG
TCF7L2	rs7903146	CC
CDKAL1	rs7756992	AG
GRB14	rs3923113	AC
TLE4	rs17791513	AA
CDC123	rs11257655	TC
CENTD2,	rs1552224	AA
KCNQ1	rs163184	TG
JAZF1	rs849135	AG
KCNJ11	rs5215	TC
ST64GAL	rs16861329	CC
MTNR1B	rs10830963	CG
HNF4A	rs4812829	GG
GIPR	rs8108269	TG
HMGA2	rs2261181	TC
SPRY2	rs1359790	AG
AP3S2	rs2028299	AA
FTO	rs9936385	TT
GLIS3	rs7041847	AA
IGF2BP2	rs4402960	GG
PPARG	rs1801282	CC

Genetic Health Risks: Gwas

Celiac disease

Celiac disease is an immune disease in which people can't eat gluten because it will damage their small intestine. If you have celiac disease and eat foods with gluten, your immune system responds by damaging the small intestine. Gluten is a protein found in wheat, rye, and barley. It may also be in other products like vitamins and supplements, hair and skin products, toothpastes, and lip balm. Celiac disease affects each person differently. Symptoms may occur in the digestive system, or in other parts of the body. One person might have diarrhea and abdominal pain, while another person may be irritable or depressed. Irritability is one of the most common symptoms in children. Some people have no symptoms.

What does your genetics say?

●

According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20190752

Your genetic map

Gene	SNP	Genotype
RGS1	rs2816316	AC
AHSA2,	rs13003464	GG
IL18R1,	rs917997	CC
ITGA4,	rs13010713	GG
ICOS,	rs4675374	CC
CCRL2,	rs13098911	TC
IL12A	rs17810546	AA
LPP	rs1464510	AC
IL2, IL21	rs13151961	AA
HLA-	rs2187668	CC
TNFAIP3	rs2327832	AG
SH2B3	rs653178	TC
PTPN2	rs1893217	AA
MMEL1,	rs3748816	AA
RUNX3	rs10903122	AG
intergeni	rs296547	CC
PLEK	rs17035378	TC
CD80,	rs11712165	TT
MAP3K7,	rs10806425	AA
THEMIS,	rs802734	AA
intergeni	rs9792269	AG
ZMIZ1	rs1250552	AA
ETS1	rs11221332	TC
CLEC16A,	rs12928822	CC
ICOSLG	rs4819388	TC
CD247	rs864537	AG
TNFSF18,	rs859637	CC
FRMD4B	rs6806528	CC
intergeni	rs10936599	CC
ELMO1	rs6974491	GG
intergeni	rs2762051	TC

Genetic Health Risks: Gwas

Alzheimer's disease (late onset)

Alzheimer's disease (AD) is the most common form of dementia among older people. Dementia is a brain disorder that seriously affects a person's ability to carry out daily activities. AD begins slowly. It first involves the parts of the brain that control thought, memory and language. People with AD may have trouble remembering things that happened recently or names of people they know. A related problem, mild cognitive impairment (MCI), causes more memory problems than normal for people of the same age. Many, but not all, people with MCI will develop AD. This section analyzes the predisposition to the Late Onset type of Alzheimer.

What does your genetics say?

●

According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24162737

Your genetic map

Gene	SNP	Genotype
CR1	rs6656401	AG
BIN1	rs6733839	TC
CD2AP	rs10948363	AG
EPHA1	rs11771145	GG
CLU	rs9331896	TT
MS4A6A	rs983392	AG
PICALM	rs10792832	AG
INPP5D	rs35349669	TC
MEF2C	rs190982	AA
NME8	rs2718058	AG
ZCWPW1	rs1476679	TC
CELF1	rs10838725	TT
FERMT2	rs17125944	TT
CASS4	rs7274581	TT
HLA-	rs9271192	CC
PTK2B	rs28834970	TC
SORL1	rs11218343	TT
SLC24A4,	rs10498633	GG
SQSTM1	rs72807343	CC
TREML2	rs9381040	CC
CD33	rs3865444	AC

Genetic Health Risks: Gwas

Coronary heart disease

Coronary heart disease is a narrowing of the small blood vessels that supply blood and oxygen to the heart. Coronary heart disease (CHD) is also called coronary artery disease. CHD is the leading cause of death in the United States for men and women. CHD is caused by the buildup of plaque in the arteries to your heart. This may also be called hardening of the arteries. Fatty material and other substances form a plaque buildup on the walls of your coronary arteries. The coronary arteries bring blood and oxygen to your heart.
This buildup causes the arteries to get narrow. As a result, blood flow to the heart can slow down or stop.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21378990

Your genetic map

Gene	**SNP**	**Genotype**
PCSK9	rs11206510	TT
CXCL12	rs1746048	CC
PPAP2B	rs17114036	AA
ANKS1A	rs17609940	GG
ZC3HC1	rs11556924	TC
ABO	rs579459	TC
CNNM2	rs12413409	GG
ZNF259,	rs964184	CC
COL4A1,	rs4773144	AA
HHIPL1	rs2895811	TT
ADAMTS7	rs3825807	AA
SMG6,	rs216172	GG
RASD1,	rs12936587	AG
SNF8,	rs46522	CC
SORT1	rs599839	AA
MIA3	rs17465637	CC
WDR12	rs6725887	TT
MRAS	rs2306374	TC
LPA	rs3798220	TT
CDKN2A,	rs4977574	AA
SH2B3	rs3184504	TC
LDLR	rs1122608	TT
SLC5A3,	rs9982601	CC
intergeni	rs10933436	AC
intergeni	rs7651039	TC
intergeni	rs7808424	TT
intergeni	rs1231206	GG

Genetic Health Risks: Gwas

Parkinson's disease

Parkinson's disease (PD) is a type of movement disorder. It happens when nerve cells in the brain don't produce enough of a brain chemical called dopamine. Sometimes it is genetic, but most cases do not seem to run in families. Exposure to chemicals in the environment might play a role. Symptoms begin gradually, often on one side of the body. Later they affect both sides. The contribution of genetics is increasing and is due to the identification of several genes and markers associated with family forms, which although they represent between 5 and 10% of the cases, their study is key to the knowledge of the disease .

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25064009

Your genetic map

Gene	SNP	Genotype
GBA,	rs35749011	GG
NUCKS1,	rs823118	TT
SIPA1L2	rs10797576	CC
ACMSD,	rs6430538	TT
STK39	rs1474055	TT
MCCC1	rs12637471	AG
SCARB2,	rs6812193	TC
SNCA	rs356182	AG
HLA-	rs9275326	CC
GPNMB	rs199347	GG
MIR4697	rs329648	TC
LRRK2	rs76904798	CC
CCDC62	rs11060180	AG
GCH1	rs11158026	TC
VPS13C	rs2414739	AA
BCKDK,	rs14235	AA
RIT2	rs12456492	AA
SPPL2B	rs62120679	CC

Genetic Health Risks: Gwas

Multiple sclerosis

Multiple sclerosis (MS) is a nervous system disease that affects your brain and spinal cord. It damages the myelin sheath, the material that surrounds and protects your nerve cells. This damage slows down or blocks messages between your brain and your body, leading to the symptoms of MS. They can include: Visual disturbances, Muscle weakness, Trouble with coordination and balance, Sensations such as numbness, prickling, or "pins and needles" or Thinking and memory problems No one knows what causes MS. It may be an autoimmune disease, which happens when your immune system attacks healthy cells in your body by mistake. Multiple sclerosis affects women more than men. It often begins between the ages of 20 and 40. Epidemiological studies show that genetic factors are responsible for their occurrence, which explains a higher frequency of the disease in relatives of affected people.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21833088

Your genetic map

Gene	SNP	Genotype
AGAP2,	rs12368653	AG
AHI1	rs11154801	AC
BACH2	rs12212193	GG
BATF	rs2300603	TC
C1orf106,	rs7522462	GG
CD80	rs2293370	AG
CD5,	rs650258	CC
CD58	rs1335532	AG
CD86	rs9282641	GG
CHST12	rs6952809	CC
CLECL1	rs10466829	AG
CXCR5	rs630923	AC
CYP24A1	rs2248359	TC
DDAH1	rs233100	AA
DKKL1	rs2303759	TG
DLEU1	rs806321	TC
EOMES	rs11129295	TC
EVI5	rs11810217	CC
VCAM1,	rs12048904	CC
FCRL3	rs3761959	CC
GPR65	rs2119704	CC
HHEX	rs7923837	AG
IL12A	rs2243123	TT
IL12B	rs2546890	AG
IL22RA2	rs17066096	AA
IL7R	rs6897932	TC
IRF8	rs13333054	CC
MALT1	rs7238078	TT
MAMSTR	rs281380	TC
MAPK1	rs2283792	TG
MERTK	rs17174870	CC

Genetic Health Risks: Gwas

Systemic sclerosis

Systemic sclerosis is a chronic autoimmune disease that causes an alteration of the collagen (protein of the connective tissue) and, as a consequence, the skin is sclerosis, that menas that it hardens. It can also affect other organs of the body such as the lungs, heart, kidneys, etc. although the most affected is the skin. The prognosis is highly variable from person to person. Exposure to certain toxic products (such as tobacco), excessive stress, exposure to cold and some drugs can influence the worsening symptoms. It affects one in 50,000 people and is more common in middle-aged women. It is a rare disease of unknown severely disabling origin. A large-scale study has found that different genetic variants are associated with the pathogenesis of the disease.

Your genetic map

Gene	SNP	Genotype
PSORS1C	rs3130573	AA
HLA,	rs6457617	TT
RHOB	rs13021401	TC
TNIP1	rs2233287	GG
CD247	rs2056626	TG
STAT4	rs7574865	GG
TNPO3,	rs10488631	TT

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21750679

This report is not valid for clinical or diagnostic use.

Genetic Health Risks: Gwas

Schizophrenia

Schizophrenia is a serious brain illness. People who have it may hear voices that aren't there. They may think other people are trying to hurt them. Sometimes they don't make sense when they talk. The disorder makes it hard for them to keep a job or take care of themselves. Symptoms of schizophrenia usually start between ages 16 and 30. Men often develop symptoms at a younger age than women. People usually do not get schizophrenia after age 45.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25056061

Your genetic map

Gene	SNP	Genotype
PLCH2	rs4648845	TT
KDM4A,	rs11210892	AA
LRRIQ3	rs12129573	AA
DPYD,	rs1702294	CC
FAM5B	rs6670165	TC
C1orf132,	rs7523273	AA
AKT3,	rs77149735	GG
FANCL,	rs11682175	TT
CYP26B1	rs3768644	GG
PCGEM1	rs59979824	AA
SATB2	rs6704641	AG
C2orf82,	rs6704768	GG
CNTN4	rs17194490	GG
TRANK1	rs75968099	TC
ATXN7,	rs832187	TC
MSL2,	rs7432375	GG
C4orf27,	rs10520163	TC
GPM6A	rs1106568	AA
HCN1	rs1501357	TT
ZSWIM6	rs4391122	AA
MEF2C	rs16867576	AA
MAN2A1	rs4388249	CC
CDC25C,	rs3849046	TT
GALNT10	rs11740474	AA
RIMS1	rs1339227	TT
FUT9	rs117074560	CC
GRM3	rs12704290	GG
MLL5,	rs6466055	CC
IMMP2L	rs13240464	
PODXL	rs7801375	GG
DGKI,	rs3735025	TT

Genetic Health Risks: Gwas

Glioma

Glioma is a type of neoplasm that occurs in the brain or spinal cord. It is called glioma because it arises from glial cells. Its most frequent location is the brain.

Your genetic map

Gene	SNP	Genotype
TERT	rs2736100	CC
TERT	rs2853676	CC
CCDC26	rs891835	TG
CCDC26	rs4295627	TT
CDKN2A,	rs4977756	GG
PHLDB1	rs498872	AG
RTEL1	rs6010620	GG

What does your genetics say?

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According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/19578367

Genetic Health Risks: Gwas

Hypothyroidism

Your thyroid is a butterfly-shaped gland in your neck, just above your collarbone. It is one of your endocrine glands, which make hormones. Thyroid hormones control the rate of many activities in your body. These include how fast you burn calories and how fast your heart beats. All of these activities are your body's metabolism. If your thyroid gland is not active enough, it does not make enough thyroid hormone to meet your body's needs. This condition is hypothyroidism. Hypothyroidism is more common in women, people with other thyroid problems, and those over 60 years old. Hashimoto's disease, an autoimmune disorder, is the most common cause. Other causes include thyroid nodules, thyroiditis, congenital hypothyroidism, surgical removal of part or all of the thyroid, radiation treatment of the thyroid, and some medicines.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22493691

Your genetic map

Gene	**SNP**	**Genotype**
INSR	rs4804416	TT
TRNAH,	rs10961534	AG
TNFRSF1	rs10162002	AG
HLA-C,	rs2517532	AG
MTF1	rs3748682	TC
PDE8B	rs4704397	AG
ZBTB10,	rs1051920	CC
ZNF804B	rs10248351	TT
KRT18P13	rs925489	TT
VAV3	rs4915077	TC
SH2B3	rs3184504	TC
PTPN22	rs6679677	CC
HLA-	rs3129720	CC

Genetic Health Risks: Gwas

Myocardial infarction (early onset)

Myocardial infarction has a hereditary component and is among the leading causes of death and disability worldwide. While most cases occur in individuals older than 65 years, 5 -10% occur in younger patients (men under 50 years and women under 60). These cases are associated with a substantially greater heritability, so it is important to identify the genes responsible. A large-scale association study has found several genetic variants that increase the risk of myocardial infarction in early onset.

Your genetic map

Gene	SNP	Genotype
CDKN2A,	rs4977574	AA
CELSR2,	rs646776	TT
MIA3	rs17465637	CC
CXCL12	rs1746048	CC
SLC5A3,	rs9982601	CC
WDR12	rs6725887	TT
LDLR	rs1122608	TT
PCSK9	rs11206510	TT

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/19198609

Genetic Health Risks: Gwas

Chronic lymphocytic leukemia

Leukemia is cancer of the white blood cells. White blood cells help your body fight infection. Your blood cells form in your bone marrow. In leukemia, the bone marrow produces abnormal white blood cells. These cells crowd out the healthy blood cells, making it hard for blood to do its work. In chronic lymphocytic leukemia (CLL), there are too many lymphocytes, a type of white blood cell.

CLL is the second most common type of leukemia in adults. It often occurs during or after middle age, and is rare in children.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23770605

Your genetic map

Gene	SNP	Genotype
ACOXL	rs17483466	AA
SP140	rs13397985	TT
FARP2	rs757978	CC
IRF4	rs872071	AG
HLA	rs9273363	CC
BAK1	rs210142	CC
MYC	rs2466035	TT
SCN3B	rs735665	GG
MNS1,	rs11636802	AA
RPLP1	rs7176508	GG
IRF8	rs391023	TT
BCL2	rs4987852	TT
ACTA2,	rs4406737	GG
BCL2	rs4987855	CC
TSPAN32	rs7944004	TG
LEF1	rs898518	AA
CASP8,	rs3769825	AG
AS1,	rs1679013	TC
PMAIP1	rs4368253	TC
ACOXL,	rs13401811	GG
ODF1,	rs2511714	TG

Genetic Health Risks: Gwas

Hodgkin's lymphoma

Hodgkin lymphoma is a cancer of the lymphatic system produced by the germ cells of the B lymphocytes (defensive cells of the immune system). The incidence in our country is 30 new cases per million inhabitants per year. It has a bimodal distribution, affecting either young, aged between 15 and 35 years, well over 55 years. 60-70% of the patients are asymptomatic and are usually detected by an increase in the volume of the lymph nodes. Although 45-60% of cases associated with infection of Epstein-Barr virus.

Your genetic map

Gene	SNP	Genotype
EOMES	rs3806624	AG
HBS1L,	rs7745098	CC
NR	rs1432295	GG
NR	rs501764	TT
PVT1	rs2019960	TC
NR	rs6903608	TC

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24149102

Genetic Health Risks: Gwas

Diffuse large B cell lymphoma

Diffuse large B-cell lymphoma (DLBCL) is a clinically aggressive B-cell (immune system) cancer and is the most common non-Hodgkin lymphoma. It is estimated that, in some european countries, the incidence of non-Hodgkin's lymphoma is 12,3 cases per 100.000 / year in men, whereas in women is 10,8 cases. It is a disease of the elderly, with an average diagnosis age of about 70 years. Diagnosis in the early stages may improve prognosis. Family history is a risk factor.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25261932

Your genetic map

Gene	SNP	Genotype
NCOA1,	rs79480871	CC
HLA-B	rs2523607	TT
MYC,	rs13255292	TC
MYC,	rs4733601	AA

Genetic Health Risks: Gwas

Follicular lymphoma

Follicular lymphoma is a form of non-Hodgkin's lymphoma that is characterized by a proliferation of B cells whose nodular structure of the follicular architecture is preserved. The prevalence of follicular lymphoma is estimated at about 1/3 000. The average age of diagnosis is 60-65 years. The disease is extremely rare in children. Follicular lymphoma is found mainly in lymph nodes, but can also affect spleen, bone marrow, peripheral blood and Waldeyer ring. The skin and central nervous system are affected in exceptional cases.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25279986

Your genetic map

Gene	SNP	Genotype
HLA	rs12195582	TC
CXCR5	rs4938573	TT
ETS1	rs4937362	CC
LPP	rs6444305	AG
BCL2	rs17749561	GG
PVT1	rs13254990	TC
SLC14A2	rs11082438	GG

Genetic Health Risks: Gwas

Myasthenia gravis

Myasthenia gravis is a disease that causes weakness in your voluntary muscles. These are the muscles that you control. For example, you may have weakness in the muscles for eye movement, facial expressions, and swallowing. You can also have weakness in other muscles. This weakness gets worse with activity, and better with rest.

Myasthenia gravis is an autoimmune disease. Your body's immune system makes antibodies that block or change some of the nerve signals to your muscles. This makes your muscles weaker.

Your genetic map

Gene	SNP	Genotype
PTPN22	rs2476601	GG
TNIP1	rs4958881	TT
NR	rs6719884	AC
NR	rs6719884	AC
NR	rs3130544	CC

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23055271

Genetic Health Risks: Gwas

Multiple myeloma

Multiple myeloma is a cancer that begins in plasma cells, a type of white blood cell. These cells are part of your immune system, which helps protect the body from germs and other harmful substances. In time, myeloma cells collect in the bone marrow and in the solid parts of bones.

No one knows the exact causes of multiple myeloma, but it is more common in older people and African Americans. It can run in families.

Your genetic map

Gene	SNP	Genotype
intergeni	rs10936599	CC
PSORS1C	rs2285803	TT
NR	rs11195062	CC
TNFRSF1	rs4273077	AA
CBX7	rs877529	AG

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23955597

Genetic Health Risks: Gwas

Neuroblastoma

Neuroblastoma is a cancer that forms in your nerve tissue. It usually begins in the adrenal glands, which sit atop your kidneys. It may also begin in your neck, chest or spinal cord. The cancer often begins in early childhood. Sometimes it begins before a child is born. By the time doctors find the cancer, it has usually spread to other parts of the body.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22941191

Your genetic map

Gene	SNP	Genotype
HACE1	rs4336470	TC
LIN28B	rs17065417	AA
BARD1	rs7587476	TC
LINC003	rs9295536	AC
LMO1	rs110419	AG
HSD17B1	rs11037575	TC

Genetic Health Risks: Gwas

Osteosarcoma

Osteosarcoma is a very rare type of cancerous bone tumor that usually develops in teenagers. It often occurs when a teen is growing rapidly. Osteosarcoma is the most common bone cancer in children. Average age at diagnosis is 15. Boys and girls are just as likely to develop this tumor until the late teens, when it occurs more often in boys. Osteosarcoma is also common in people over age 60.

The cause is not known. In some cases, osteosarcoma runs in families. At least one gene has been linked to an increased risk. This gene is also associated with familial retinoblastoma. This is a cancer of the eye that occurs in children.

What does your genetics say?

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According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23727862

Your genetic map

Gene	SNP	Genotype
GRM4	rs1906953	TC
AJ412031	rs573666	TC
intergeni	rs7591996	CC
ADAMTS6	rs17206779	CC

Genetic Health Risks: Gwas

Psoriasis

Psoriasis is a skin disease that causes itchy or sore patches of thick, red skin with silvery scales. You usually get the patches on your elbows, knees, scalp, back, face, palms and feet, but they can show up on other parts of your body. Some people who have psoriasis also get a form of arthritis called psoriatic arthritis. A problem with your immune system causes psoriasis. In a process called cell turnover, skin cells that grow deep in your skin rise to the surface. Normally, this takes a month. In psoriasis, it happens in just days because your cells rise too fast. The disease is not hereditary, but there is a genetic predisposition to it, and a third of those affected have direct relatives with psoriasis.

What does your genetics say?

🟢

According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25903422

Your genetic map

Gene	SNP	Genotype
TP63	rs28512356	CC
COG6	rs34394770	TC
LOC1448	rs9533962	TT
RUNX1	rs8128234	CC
TP63	rs28512356	CC
COG6	rs34394770	TC
LOC1448	rs9533962	TT
RUNX1	rs8128234	CC
CLIC6	rs9305556	GG
OSTN	rs11922372	TT
IL12B	rs7709212	TC
TNIP,	rs17728338	GG
IL12B	rs4921493	CC
IFIH1	rs3747517	CC
LCE	rs4845459	
TNFAIP3	rs643177	TC
REL	rs842625	AA
IL12B	rs2853694	TG
IFIH1	rs1990760	TT
PSMA6,	rs8016947	TT
NOS2	rs4795067	AA
IL12B	rs7709212	TC
IL12B	rs7709212	TC
LCE	rs4845459	
TNIP,	rs17728338	GG
IL12B	rs7709212	TC
TNIP,	rs17728338	GG
IFIH1	rs3747517	CC
LCE	rs4845459	
IL12B	rs4921493	CC
TNFAIP3	rs643177	TC

Genetic Health Risks: Gwas

Allergic sensitization

Allergic sensitization is the result of a complex interaction between the allergen and the host in a given environmental context. The first barrier found by an allergen on its way to sensitization is the epithelial layer of the mucosa. Allergic inflammatory diseases are accompanied by increased permeability of the epithelium, which is more susceptible to environmental triggers.

Your genetic map

Gene	SNP	Genotype
LRRC32,	rs2155219	TT
STAT6	rs1059513	TT
TSLP,	rs10056340	TG
HLA-	rs6906021	TC
IL18R1,	rs3771175	TA
FAM114A	rs17616434	TC
LPP,	rs9865818	AA
MYC	rs4410871	CC
IL2,	rs17454584	AA
MICA,	rs6932730	TC

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23817571

Genetic Health Risks: Gwas

Testicular germ cell tumor

Testicular germ cell tumors (TCC) affect 1 in 500 men and are the most common cancer in men aged 15-40 years in Western European populations. The incidence of TGCT increased dramatically during the 20th century. Known risk factors for TGCT include a history of undescended testis (UDT), testicular dysgenesis, infertility, previously diagnosed TGCT, and family history of the disease. Siblings of men with TGCT have a 8 to 10-fold risk of developing TGCT, while the relative risk for fathers and sons is 4-fold. This family relative risk is much higher than that of most other types of cancer.

Your genetic map

Gene	SNP	Genotype
SLC25A4	rs2072499	AA
UCK2	rs3790672	CC
DAZL	rs10510452	AG
CENPE	rs2720460	AA
CATSPER	rs3805663	AA
PRDM14	rs7010162	CC
HEATR3	rs8046148	AG
RAD51C,	rs9905704	TG
MCM3AP	rs2839186	TC
TERT,	rs4635969	AG
SPRY4	rs4624820	GG
BAK1	rs210138	AA
DMRT1	rs755383	TC
ATF7IP	rs2900333	CC
KITLG	rs995030	AG

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23666240

Genetic Health Risks: Gwas

Wilms tumor

Wilms tumor is a rare type of kidney cancer. It causes a tumor on one or both kidneys. It usually affects children, but can happen in adults. Having certain genetic conditions or birth defects can increase the risk of getting it. Children that are at risk should be screened for Wilms tumor every three months until they turn eight.

Symptoms include a lump in the abdomen, blood in the urine, and a fever for no reason. Tests that examine the kidney and blood are used to find the tumor.

Your genetic map

Gene	SNP	Genotype
MYCN,	rs3755132	TT
NR	rs1027643	CC
DLG2	rs790356	AG
NR	rs2283873	GG
NR	rs5955543	AA
MYCN,	rs807624	TG

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22544364

Genetic Health Risks: Gwas

Vitiligo

Vitiligo causes white patches on your skin. It can also affect your eyes, mouth, and nose. It occurs when the cells that give your skin its color are destroyed. No one knows what destroys them. It is more common in people with autoimmune diseases, and it might run in families. It usually starts before age 40.

The white patches are more common where your skin is exposed to the sun. In some cases, the patches spread. Vitiligo can cause your hair to gray early. If you have dark skin, you may lose color inside your mouth.

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22561518

Your genetic map

Gene	SNP	Genotype
IFIH1	rs2111485	GG
CD80	rs59374417	AA
CLNK	rs16872571	TT
BACH2	rs3757247	TT
SLA	rs853308	TC
CASP7	rs3814231	TC
CD44	rs10768122	AG
TYR	rs4409785	TC
IKZF4	rs2456973	AA
SH2B3	rs4766578	TA
HERC2,	rs1129038	CC
MC1R	rs9926296	AA
TICAM1	rs6510827	CC
TOB2	rs4822024	GG

Genetic Health Risks: mutations

APC: colorectal and pancreatic cancer

APC gene mutations may be related to diseases such colorectal and pancreatic cancer. There is some publication that relates it, in some cases, to gastric cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/11978510

Your genetic map

Gene	SNP	Genotype
APC	rs387906230	TT
APC	rs121913327	CC
APC	rs137854573	CC
APC	rs137854580	CC
APC	rs397514031	GG
APC	rs397515734	CC
APC	rs398123116	GG
APC	rs398123117	CC
APC	rs398123119	GG
APC	rs398123121	CC
APC	rs587779780	CC
APC	rs587779783	CC
APC	rs587779786	AA
APC	rs587779790	AA
APC	rs62619935	CC
APC	rs587781392	CC
APC	rs587782518	CC
APC	rs397514031	GG
APC	rs730881240	CC
APC	rs730881247	CC
APC	rs775126020	CC
APC	rs768922431	CC
APC	rs559510809	GG
APC	rs121913333	CC
APC	rs387906230	TT
APC	rs199740875	GG
APC	rs141576417	CC

Genetic Health Risks: mutations

ATM: breast cancer

Mutations of the ATM gene may be related to diseases such as breast cancer. There is some publication that relates this gene, to a lesser extent, to other cancers such as ovarian cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/17061036

Your genetic map

Gene	SNP	Genotype
ATM	rs28904921	TT
ATM	rs55861249	CC
ATM	rs121434219	CC
ATM	rs587776551	GG
ATM	rs121434220	CC
ATM	rs587779813	GG
ATM	rs587779815	CC
ATM	rs587779818	GG
ATM	rs587779826	TT
ATM	rs587779833	CC
ATM	rs587779836	GG
ATM	rs200976093	CC
ATM	rs587779852	GG
ATM	rs532480170	CC
ATM	rs587779856	GG
ATM	rs587779865	CC
ATM	rs587779866	AA
ATM	rs587779872	CC
ATM	rs17174393	GG
ATM	rs587780639	GG
ATM	rs371638537	AA
ATM	rs587781363	CC
ATM	rs587781545	CC
ATM	rs587781558	GG
ATM	rs377349459	GG
ATM	rs587781597	CC
ATM	rs587781672	GG
ATM	rs587781698	CC
ATM	rs587781722	CC
ATM	rs200196781	GG
ATM	rs587781911	GG

Genetic Health Risks: mutations

BRCA1: breast and ovarian cancer

Mutations of the BRCA1 gene may be related to diseases such as breast and ovarian cancer. There is some study that relates this gene, on a smaller scale, to other cancers such as colon or pancreatic cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/7907678

Your genetic map

Gene	SNP	Genotype
BRCA1	rs62625308	GG
BRCA1	rs28897686	CC
BRCA1	rs41293455	GG
BRCA1	rs62625306	CC
BRCA1	rs80357382	TT
BRCA1	rs80358158	CC
BRCA1	rs80356898	GG
BRCA1	rs80357355	TT
BRCA1	rs80358061	AA
BRCA1	rs80358163	TT
BRCA1	rs80357233	GG
BRCA1	rs80356875	CC
BRCA1	rs80356925	GG
BRCA1	rs80357251	CC
BRCA1	rs80357115	AA
BRCA1	rs397507215	GG
BRCA1	rs80357018	CC
BRCA1	rs80357318	GG
BRCA1	rs80357021	CC
BRCA1	rs80358178	CC
BRCA1	rs80358070	CC
BRCA1	rs80357259	CC
BRCA1	rs80356991	CC
BRCA1	rs80358027	CC
BRCA1	rs80357389	CC
BRCA1	rs80356988	CC
BRCA1	rs80356988	CC
BRCA1	rs80357433	GG
BRCA1	rs80358086	AA
BRCA1	rs80358053	CC
BRCA1	rs80358053	CC

Genetic Health Risks: mutations

BRCA2: breast and ovarian cancer

Mutations of the BRCA2 gene may be related to diseases such as breast and ovarian cancer. There is some study that relates this gene, on a smaller scale, to other cancers such as pancreatic cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/9497246

Your genetic map

Gene	SNP	Genotype
BRCA2	rs80359062	CC
BRCA2	rs80358785	CC
BRCA2	rs80359180	CC
BRCA2	rs81002897	GG
BRCA2	rs81002899	TT
BRCA2	rs80358474	CC
BRCA2	rs80358504	TT
BRCA2	rs397507285	TT
BRCA2	rs80358529	CC
BRCA2	rs80358532	CC
BRCA2	rs80358544	GG
BRCA2	rs80358557	CC
BRCA2	rs41293477	TT
BRCA2	rs397507303	GG
BRCA2	rs397507303	GG
BRCA2	rs80358638	GG
BRCA2	rs80358650	GG
BRCA2	rs80358663	CC
BRCA2	rs81002853	AA
BRCA2	rs80358721	CC
BRCA2	rs200265692	AA
BRCA2	rs80358785	CC
BRCA2	rs80358789	CC
BRCA2	rs41293497	CC
BRCA2	rs56253082	GG
BRCA2	rs80358831	CC
BRCA2	rs80358840	AA
BRCA2	rs80358920	CC
BRCA2	rs397507384	CC
BRCA2	rs80359011	GG
BRCA2	rs81002874	GG

Genetic Health Risks: mutations

BRIP1: breast cancer

Mutations in the BRIP1 gene may be related to diseases such as breast cancer. There is some study that relates this gene, on a smaller scale, to ovarian cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21964575

Your genetic map

Gene	SNP	Genotype
BRIP1	rs587780226	GG
BRIP1	rs587780228	CC
BRIP1	rs587780833	CC
BRIP1	rs587781292	CC
BRIP1	rs587781321	GG
BRIP1	rs587781655	CC
BRIP1	rs368796923	GG
BRIP1	rs587781786	GG
BRIP1	rs574552037	GG
BRIP1	rs587782410	AA
BRIP1	rs587782514	AA
BRIP1	rs587782539	CC
BRIP1	rs587782574	GG
BRIP1	rs730881633	GG
BRIP1	rs747604569	GG
BRIP1	rs587780875	AA
BRIP1	rs775171520	CC

Genetic Health Risks: mutations

CDH1: breast and gastric cancer

Mutations of the CDH1 gene may be related to diseases such as breast and gastric cancer. There is some study linking this gene, on a smaller scale, to ovarian and colon cancer.

Your genetic map

Gene	SNP	Genotype
CDH1	rs587780784	CC
CDH1	rs587780787	GG
CDH1	rs587782750	CC
CDH1	rs587782798	CC
CDH1	rs587783047	CC
CDH1	rs587783050	GG
CDH1	rs730881663	CC

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/11729114

Genetic Health Risks: mutations

CDKN2A: pancreatic cancer

CDKN2A gene mutations may be related to diseases such as pancreatic cancer.

Your genetic map

Gene	SNP	Genotype
CDKN2A	rs104894097	CC
CDKN2A	rs730881677	CC
CDKN2A	rs1800586	CC
CDKN2A	rs45476696	CC
CDKN2A	rs730881677	CC

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/10956390

Genetic Health Risks: mutations

CHEK2: breast and colorectal cancer

CHEK2 gene mutations may be related to diseases such as breast and colorectal cancer.

Your genetic map

Gene	SNP	Genotype
CHEK2	rs137853007	GG
CHEK2	rs121908698	CC
CHEK2	rs28909982	TT
CHEK2	rs121908698	CC
CHEK2	rs587781269	GG
CHEK2	rs587781592	GG
CHEK2	rs587781705	AA
CHEK2	rs587781836	AA
CHEK2	rs587782070	CC
CHEK2	rs730881702	CC
CHEK2	rs730881701	GG
CHEK2	rs760502479	GG
CHEK2	rs761494650	GG

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21807500

Genetic Health Risks: mutations

MLH1: Lynch syndrome

MLH1 gene mutations may be related to diseases such as Lynch syndrome.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/20301390

Your genetic map

Gene	SNP	Genotype
MLH1	rs63750198	CC
MLH1	rs63751109	CC
MLH1	rs63750710	AA
MLH1	rs63751615	CC
MLH1	rs63750206	GG
MLH1	rs63750781	CC
MLH1	rs63750899	CC
MLH1	rs63750691	CC
MLH1	rs63750217	GG
MLH1	rs63749939	GG
MLH1	rs63751194	CC
MLH1	rs63750540	AA
MLH1	rs63751221	CC
MLH1	rs193922370	GG
MLH1	rs63751715	GG
MLH1	rs63751715	GG
MLH1	rs63751715	GG
MLH1	rs63749906	TT
MLH1	rs63750580	AA
MLH1	rs587778888	AA
MLH1	rs267607706	CC
MLH1	rs267607710	GG
MLH1	rs587778894	CC
MLH1	rs63750483	CC
MLH1	rs267607713	GG
MLH1	rs63751153	CC
MLH1	rs267607825	GG
MLH1	rs587778913	CC
MLH1	rs63749795	CC
MLH1	rs587778918	AA
MLH1	rs63749923	CC

Genetic Health Risks: mutations

MSH2: Lynch syndrome and colorectal cancer

MSH2 gene mutations may be related to diseases such as Lynch syndrome and colorectal cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25070057

Your genetic map

Gene	SNP	Genotype
MSH2	rs28929483	CC
MSH2	rs63751108	CC
MSH2	rs28929484	CC
MSH2	rs63750047	CC
MSH2	rs63750875	GG
MSH2	rs63750245	CC
MSH2	rs63749932	CC
MSH2	rs193922376	AA
MSH2	rs587779063	AA
MSH2	rs63750778	CC
MSH2	rs587779065	GG
MSH2	rs63751027	GG
MSH2	rs63750396	GG
MSH2	rs587779067	CC
MSH2	rs587779067	CC
MSH2	rs587779070	AA
MSH2	rs267607940	GG
MSH2	rs63751617	AA
MSH2	rs63750558	CC
MSH2	rs63750267	CC
MSH2	rs63749849	CC
MSH2	rs587779075	CC
MSH2	rs63750302	CC
MSH2	rs63750611	CC
MSH2	rs63751412	CC
MSH2	rs63751271	CC
MSH2	rs63750006	CC
MSH2	rs63751712	GG
MSH2	rs267607949	AA
MSH2	rs63751693	CC
MSH2	rs63751646	AA

Genetic Health Risks: mutations

MSH6: Lynch syndrome and colorectal cancer

MSH6 gene mutations may be related to diseases such as Lynch syndrome and colorectal cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15236168

Your genetic map

Gene	**SNP**	**Genotype**
MSH6	rs397515875	GG
MSH6	rs267608094	CC
MSH6	rs587779208	TT
MSH6	rs63750741	TT
MSH6	rs267608046	GG
MSH6	rs587779212	CC
MSH6	rs63750564	CC
MSH6	rs267608068	TT
MSH6	rs587779232	TT
MSH6	rs63751442	CC
MSH6	rs63751127	CC
MSH6	rs587779234	GG
MSH6	rs63751321	CC
MSH6	rs587779245	TT
MSH6	rs63751017	CC
MSH6	rs587779246	CC
MSH6	rs63750140	CC
MSH6	rs63750111	CC
MSH6	rs63750258	GG
MSH6	rs63750563	CC
MSH6	rs587779252	GG
MSH6	rs267608059	GG
MSH6	rs63749999	CC
MSH6	rs63749843	CC
MSH6	rs267608084	GG
MSH6	rs267608086	GG
MSH6	rs63750356	CC
MSH6	rs587779267	GG
MSH6	rs587779279	GG
MSH6	rs267608111	AA
MSH6	rs63751058	TT

Genetic Health Risks: mutations

MUTYH: MYH-associated polyposis and colorectal cancer

MUTYH gene mutations may be related to diseases such as MYH-associated polyposis and colorectal cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23035301

Your genetic map

Gene	**SNP**	**Genotype**
MUTYH	rs34612342	TT
MUTYH	rs36053993	CC
MUTYH	rs121908380	GG
MUTYH	rs200844166	GG
MUTYH	rs200495564	GG
MUTYH	rs587780082	GG
MUTYH	rs587780751	TT
MUTYH	rs587781295	CC
MUTYH	rs587781338	GG
MUTYH	rs140342925	CC
MUTYH	rs587781628	TT
MUTYH	rs529008617	GG
MUTYH	rs587782885	GG
MUTYH	rs730881833	CC
MUTYH	rs143353451	CC
MUTYH	rs730881832	AA
MUTYH	rs374950566	GG
MUTYH	rs34126013	GG
MUTYH	rs747993448	GG
MUTYH	rs372267274	CC
MUTYH	rs765123255	GG
MUTYH	rs748170941	CC
MUTYH	rs587782228	CC
MUTYH	rs372267274	CC

Genetic Health Risks: mutations

PALB2: breast and pancreatic cancer

PALB2 gene mutations may be related to diseases such as breast and pancreatic cancer

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25099575

Your genetic map

Gene	SNP	Genotype
PALB2	rs118203998	GG
PALB2	rs180177103	CC
PALB2	rs180177083	GG
PALB2	rs180177112	CC
PALB2	rs587776417	CC
PALB2	rs587776527	GG
PALB2	rs180177100	GG
PALB2	rs118203998	GG
PALB2	rs587782050	CC
PALB2	rs180177110	GG
PALB2	rs587782446	GG
PALB2	rs587776419	CC
PALB2	rs587776419	CC
PALB2	rs730881888	AA
PALB2	rs730881905	CC
PALB2	rs730881884	CC

Genetic Health Risks: mutations

PMS2: Lynch syndrome and colorectal cancer

PMS2 gene mutations may be related to diseases such as Lynch syndrome and colorectal cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 25 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/19861671

Your genetic map

Gene	**SNP**	**Genotype**
PMS2	rs63750871	GG
PMS2	rs587778617	GG
PMS2	rs63750490	TT
PMS2	rs63751422	GG
PMS2	rs201451115	TT
PMS2	rs587779343	GG
PMS2	rs63750261	GG
PMS2	rs200640585	GG
PMS2	rs143277125	GG
PMS2	rs587780059	AA
PMS2	rs587780062	GG
PMS2	rs587780064	CC
PMS2	rs587778618	GG
PMS2	rs587781339	TT
PMS2	rs587782074	CC
PMS2	rs141577476	GG
PMS2	rs587780059	AA
PMS2	rs778531080	CC
PMS2	rs63751228	GG
PMS2	rs587780059	AA

Genetic Health Risks: mutations

PTEN: breast, uterine and colorectal cancer

PTEN gene mutations may be related to diseases such as breast, uterine and colorectal cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/22252256

Your genetic map

Gene	SNP	Genotype
PTEN	rs121909219	CC
PTEN	rs121909223	TT
PTEN	rs121909224	CC
PTEN	rs121909229	GG
PTEN	rs121909238	AA
PTEN	rs587781784	AA
PTEN	rs587782187	TT
PTEN	rs587782350	CC
PTEN	rs587782360	AA
PTEN	rs587782603	GG
PTEN	rs727504114	TT
PTEN	rs398123317	TT
PTEN	rs121913293	CC
PTEN	rs746930141	GG
PTEN	rs398123320	CC
PTEN	rs121909224	CC
PTEN	rs121913294	GG
PTEN	rs121909229	GG

Genetic Health Risks: mutations

SDHB: gastric cancer

SDHB gene mutations may be related to diseases such as gastric cancer.

Your genetic map

Gene	SNP	Genotype
SDHB	rs74315366	GG
SDHB	rs74315368	
SDHB	rs74315369	GG
SDHB	rs74315370	GG
SDHB	rs267607032	CC
SDHB	rs398122805	CC
SDHB	rs397516833	CC
SDHB	rs397516836	CC
SDHB	rs587781270	AA
SDHB	rs397516835	CC
SDHB	rs587782604	CC
SDHB	rs74315370	GG
SDHB	rs587782703	CC
SDHB	rs138996609	GG
SDHB	rs397516836	CC
SDHB	rs772551056	CC
SDHB	rs751000085	GG
SDHB	rs200245469	GG
SDHB	rs587782703	CC

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25376524

Genetic Health Risks: mutations

SMAD4: juvenile polyposis syndrome and colorectal cancer

SMAD4 gene mutations may be related to diseases such as juvenile polyposis syndrome and colorectal cancer. There is some study linking this gene, on a smaller scale, to pancreatic cancer.

Your genetic map

Gene	SNP	Genotype
SMAD4	rs80338963	CC
SMAD4	rs80338963	CC
SMAD4	rs281875324	AA
SMAD4	rs80338963	CC
SMAD4	rs377767360	CC
SMAD4	rs281875322	AA
SMAD4	rs397518413	CC
SMAD4	rs587781359	CC
SMAD4	rs730881954	CC
SMAD4	rs281875324	AA

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 10 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25645574

Genetic Health Risks: mutations

TP53: Li-Fraumeni syndrome, breast cancer and more

TP53 gene mutations may be related to diseases such Li-Fraumeni syndrome, breast, ovarian, uterine, colorectal and pancreatic cancer. There is some study that relates this gene, on a smaller scale, to gastric cancer.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/10864200

Your genetic map

Gene	SNP	Genotype
TP53	rs121912658	TT
TP53	rs121912651	GG
TP53	rs121912652	CC
TP53	rs121912653	AA
TP53	rs121912655	CC
TP53	rs121912656	CC
TP53	rs11540652	CC
TP53	rs28934873	AA
TP53	rs28934573	GG
TP53	rs28934576	CC
TP53	rs28934874	GG
TP53	rs28934874	GG
TP53	rs28934578	CC
TP53	rs121912662	AA
TP53	rs121912664	CC
TP53	rs397516436	GG
TP53	rs397516439	TT
TP53	rs483352695	TT
TP53	rs587780070	GG
TP53	rs587780071	GG
TP53	rs587780074	AA
TP53	rs587780073	TT
TP53	rs587778720	CC
TP53	rs587781288	CC
TP53	rs28934574	GG
TP53	rs587781525	TT
TP53	rs587781664	TT
TP53	rs587781702	CC
TP53	rs587782144	CC
TP53	rs587782160	TT
TP53	rs121913344	GG

Genetic Health Risks: mutations

VHL: Von Hippel-Lindau syndrome

VHL gene mutations may be related to diseases such Von Hippel-Lindau syndrome.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21386872

Your genetic map

Gene	SNP	Genotype
VHL	rs5030821	GG
VHL	rs5030818	CC
VHL	rs119103277	GG
VHL	rs5030809	TT
VHL	rs104893826	GG
VHL	rs104893830	GG
VHL	rs104893831	GG
VHL	rs5030827	GG
VHL	rs193922609	GG
VHL	rs5030826	CC
VHL	rs397516440	CC
VHL	rs5030817	GG
VHL	rs397516445	TT
VHL	rs5030804	AA
VHL	rs398123481	CC
VHL	rs398123481	CC
VHL	rs727504215	GG
VHL	rs5030826	CC
VHL	rs730882034	CC
VHL	rs119103277	GG
VHL	rs5030807	TT
VHL	rs121913346	TT
VHL	rs730882035	GG
VHL	rs5030810	CC
VHL	rs5030826	CC
VHL	rs5030804	AA
VHL	rs193922609	GG
VHL	rs730882032	GG
VHL	rs5030817	GG
VHL	rs5030817	GG
VHL	rs5030827	GG

Genetic Health Risks: mutations

RET: thyroid carcinoma

RET gene mutations may be related to diseases such thyroid carcinoma.

What does your genetics say?

We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions. In this panel we are analyzing less than 50 % of the pathogenic mutations reported in ClinVar.

More information:

https://www.ncbi.nlm.nih.gov/medgen/C1833921

Your genetic map

Gene	SNP	Genotype
RET	rs79781594	GG
RET	rs77316810	TT
RET	rs77503355	GG
RET	rs74799832	TT
RET	rs77503355	GG
RET	rs77939446	GG
RET	rs75030001	GG
RET	rs77503355	GG
RET	rs75234356	TT
RET	rs79781594	GG
RET	rs79781594	GG
RET	rs77316810	TT
RET	rs78347871	GG
RET	rs77939446	GG
RET	rs77939446	GG
RET	rs77316810	TT
RET	rs75030001	GG

Carrier Status

17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY

17-beta-hydroxysteroid dehydrogenase isozyme 3 (17betaHSD III) deficiency is a rare disorder leading to male pseudohermaphroditism (MPH), a condition characterized by incomplete differentiation of the male genitalia in 46X,Y males. The estimated incidence of this disease is 1 in 147 000 in The Netherlands. The 17betaHSD III enzyme catalyzes the conversion of androstenedione to testosterone in the testis. Lack of testosterone in the fetal testis leads to genetic males with female external genitalia. Patients usually present at birth with female or ambiguous external genitalia, characterized by clitoromegaly, posterior labioscrotal fusion and perineal blind vaginal pouch. Testes are inguinal or in the labioscrotal folds.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/264300

Your genetic map

Gene	SNP	Genotype
HSD17B3	rs119481077	GG

Carrier Status

3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY; MCC2D

17-beta-hydroxysteroid dehydrogenase isozyme 3 (17betaHSD III) deficiency is a rare disorder leading to male pseudohermaphroditism (MPH), a condition characterized by incomplete differentiation of the male genitalia in 46X,Y males. The estimated incidence of this disease is 1 in 147 000 in The Netherlands. The 17betaHSD III enzyme catalyzes the conversion of androstenedione to testosterone in the testis. Lack of testosterone in the fetal testis leads to genetic males with female external genitalia. Patients usually present at birth with female or ambiguous external genitalia, characterized by clitoromegaly, posterior labioscrotal fusion and perineal blind vaginal pouch. Testes are inguinal or in the labioscrotal folds. The internal urogenital tract (epididymides, vasa deferentia, seminal vesicles, ejaculatory ducts) is well developed; prostate and Müllerian structures are absent. Although some patients with less severe defects are brought up as males, affected males are usually brought up as females.

Your genetic map

Gene	SNP	Genotype
MCCC2	rs763293192	CC
MCCC2	rs119103219	GG
MCCC2	rs398124372	CC
MCCC2	rs727504010	CC
MCCC2	rs773774134	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/210210

Carrier Status

AARSKOG-SCOTT SYNDROME; AAS

Aarskog-Scott syndrome (AAS) is a rare developmental disorder characterized by facial, limbs and genital features, and a disproportionate acromelic short stature. AAS prevalence is not known, but less than 100 cases have been reported in the literature since the first description in 1970. However, prevalence estimates are thought to be around 1/25,000. About 40 molecularly proven cases are published worldwide.

Your genetic map

Gene	SNP	Genotype
FGD1	rs398124155	AA
FGD1	rs398124156	GG
FGD1	rs398124160	GG
FGD1	rs398124162	DD

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/305400

Carrier Status

ACHROMATOPSIA 2; ACHM2

Achromatopsia is characterized by reduced visual acuity, pendular nystagmus, increased sensitivity to light (photophobia), a small central scotoma, eccentric fixation, and reduced or complete loss of color discrimination. All individuals with achromatopsia (achromats) have impaired color discrimination along all three axes of color vision corresponding to the three cone classes: the protan or long-wavelength-sensitive cone axis (red), the deutan or middle-wavelength-sensitive cone axis (green), and the tritan or short-wavelength-sensitive cone axis (blue). Most individuals have complete achromatopsia, with total lack of function of all three types of cones. Rarely, individuals have incomplete achromatopsia, in which one or more cone types may be partially functioning. The symptoms are similar to those of individuals with complete achromatopsia, but generally less severe. Hyperopia is common in achromatopsia.

Your genetic map

Gene	**SNP**	**Genotype**
CNGA3	rs104893613	CC
CNGA3	rs104893617	CC
CNGA3	rs104893619	GG
CNGA3	rs147118493	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/216900

Carrier Status

LEUKEMIA, ACUTE MYELOID; AML

Acute myeloid leukemia (AML) is a group of neoplasms arising from precursor cells committed to the myeloid cell-line differentiation. All of them are characterized by clonal expansion of myeloid blasts. AML manifests by fever, pallor, anemia, hemorrhages and recurrent infections.Annual incidence rate of AML is estimated to be 1/33,000-1/25,000 in Europe.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601626

Your genetic map

Gene	SNP	Genotype
HRAS	rs104894229	CC
HRAS	rs104894229	CC
HRAS	rs104894230	CC
TP53	rs28934576	CC
TP53	rs28934576	CC
TP53	rs121912651	GG
TP53	rs11540652	CC
TP53	rs11540652	CC
TP53	rs11540652	CC
TP53	rs587781288	CC
TP53	rs587780070	GG
HRAS	rs104894228	CC
TP53	rs760043106	AA
HRAS	rs104894226	CC
HRAS	rs121917759	GG
NRAS	rs121913250	CC
NRAS	rs121913237	CC
NRAS	rs121434596	CC
NRAS	rs121913237	CC
NRAS	rs121913250	CC
NRAS	rs121913250	CC
NRAS	rs121913237	CC
JAK2	rs77375493	GG
PTPN11	rs121918453	GG
IDH2	rs121913502	CC

Carrier Status

ADRENOLEUKODYSTROPHY; ALD

X-linked adrenoleukodystrophy (X-ALD) affects the nervous system white matter and the adrenal cortex. Three main phenotypes are seen in affected males: The childhood cerebral form manifests most commonly between ages four and eight years. It initially resembles attention deficit disorder or hyperactivity; progressive impairment of cognition, behavior, vision, hearing, and motor function follow the initial symptoms and often lead to total disability within two years. Adrenomyeloneuropathy (AMN) manifests most commonly in the late twenties as progressive paraparesis, sphincter disturbances, sexual dysfunction, and often, impaired adrenocortical function; all symptoms are progressive over decades. "Addison disease only" presents with primary adrenocortical insufficiency between age two years and adulthood and most commonly by age 7.5 years, without evidence of neurologic abnormalit. Approximately 20% of females who are carriers develop neurologic manifestations that resemble AMN but have later onset (age ≥35 years) and milder disease than do affected males.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300100

Your genetic map

Gene	SNP	Genotype
ABCD1	rs128624218	GG
ABCD1	rs128624220	CC
ABCD1	rs387906494	II
ABCD1	rs128624224	CC
ABCD1	rs193922093	DD
ABCD1	rs193922097	GG
ABCD1	rs193922098	CC
ABCD1	rs398123100	CC
ABCD1	rs398123102	GG
ABCD1	rs398123103	GG
ABCD1	rs398123104	CC
ABCD1	rs398123105	CC
ABCD1	rs398123106	CC
ABCD1	rs398123107	GG
ABCD1	rs398123110	GG
ABCD1	rs398123112	II
ABCD1	rs398123113	CC
ABCD1	rs727503786	CC
ABCD1	rs398123108	GG

Carrier Status

HYPOPHOSPHATASIA, ADULT

Hypophosphatasia (HPP) is a rare heritable metabolic disorder characterized by defective mineralization of bone and/or teeth in the presence of reduced activity of unfractionated serum alkaline phosphatase (ALP). The clinical spectrum is extremely wide, from stillbirth at one end to fractures of the lower extremities in adulthood, at the other, or even no bone manifestations (odontohypophosphatasia).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/146300

Your genetic map

Gene	SNP	Genotype
ALPL	rs121918007	GG
ALPL	rs121918002	AA
ALPL	rs121918013	GG
ALPL	rs121918010	TT
ALPL	rs387906525	II

Carrier Status

ALLAN-HERNDON-DUDLEY SYNDROME; AHDS

Allan-Herndon-Dudley syndrome (AHDS) is an X-linked intellectual disability syndrome with neuromuscular involvement characterized by infantile hypotonia, muscular hypoplasia, spastic paraparesis with dystonic/athetoic movements, and severe cognitive deficiency. At least 132 families with 320 affected individuals have been reported in the literature to date. Although the prevalence is unknown, one study identified AHDS in 1.4% of males with intellectual disability of unknown etiology. Only males are affected.

Your genetic map

Gene	SNP	Genotype
SLC16A2	rs387906501	II
SLC16A2	rs587784386	CC
SLC16A2	rs104894936	CC
SLC16A2	rs587784382	CC
SLC16A2	rs766773277	CC
SLC16A2	rs587784383	GG
SLC16A2	rs104894936	CC
SLC16A2	rs587784384	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300523

Carrier Status

ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD

Alpha-1-antitrypsin deficiency is a hereditary disease that develops in adulthood and is characterized by chronic liver disorders (cirrhosis), respiratory disorders (emphysema), and rarely panniculitis.

Your genetic map

Gene	SNP	Genotype
SERPINA1	rs61761869	GG
SERPINA1	rs28929474	CC
SERPINA1	rs61761869	GG
SERPINA1	rs199422211	TT
SERPINA1	rs55819880	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613490

Carrier Status

AMYLOIDOSIS, HEREDITARY, TRANSTHYRETIN-RELATED

Transthyretin (TTR)-related familial amyloidotic cardiomyopathy is a hereditary TTR-related systemic amyloidosis (ATTR) with predominant cardiac involvement resulting from myocardial infiltration of abnormal amyloid protein. Prevalence is unknown. Patients present during adulthood (usually after 30 years of age) with restrictive cardiomyopathy (with varying degrees of chronic heart failure and possible brady/tachyarrhythmias).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/105210

Your genetic map

Gene	SNP	Genotype
TTR	rs76992529	GG
TTR	rs386134269	AA
TTR	rs121918076	TT
TTR	rs121918069	TT
TTR	rs121918070	AA
TTR	rs121918093	GG
TTR	rs121918098	AA

Carrier Status

ANEMIA, NONSPHEROCYTIC HEMOLYTIC, DUE TO G6PD DEFICIENCY

G6PD deficiency is the most common genetic cause of chronic and drug-, food-, or infection-induced hemolytic anemia. G6PD catalyzes the first reaction in the pentose phosphate pathway, which is the only NADPH-generation process in mature red cells; therefore, defense against oxidative damage is dependent on G6PD. The most common clinical manifestations of G6PD deficiency are neonatal jaundice and acute hemolytic anemia, which in most patients is triggered by an exogenous agent, e.g., primaquine or fava beans (see 134700). Acute hemolysis is characterized by fatigue, back pain, anemia, and jaundice. Increased unconjugated bilirubin, lactate dehydrogenase, and reticulocytosis are markers of the disorder. Although G6PD deficiency can be life-threatening, most G6PD-deficient patients are asymptomatic throughout their life. The striking similarity between the areas where G6PD deficiency is common and Plasmodium falciparum malaria (see 611162) is endemic provided evidence that G6PD deficiency confers resistance against malaria.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300908

Your genetic map

Gene	SNP	Genotype
G6PD	rs5030868	GG
G6PD	rs137852331	TT
G6PD	rs398123546	GG
G6PD	rs72554665	CC
G6PD	rs5030869	CC
G6PD	rs72554665	CC
G6PD	rs137852326	CC
G6PD	rs137852327	CC
G6PD	rs137852314	CC
G6PD	rs137852318	CC
G6PD	rs137852317	CC
G6PD	rs76723693	AA
G6PD	rs78365220	AA
G6PD	rs398123552	AA

Carrier Status

ANGELMAN SYNDROME; AS

Angelman syndrome (AS) is a neurogenetic disorder characterized by severe intellectual deficit and distinct facial dysmorphic features.Prevalence of AS is estimated to be 1/10,000 to 1/20,000 worldwide.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/105830

Your genetic map

Gene	SNP	Genotype
UBE3A	rs111033595	CC
UBE3A	rs587780570	II
UBE3A	rs587780577	AA
UBE3A	rs587781204	DD
UBE3A	rs587781208	CC
UBE3A	rs587781220	CC
UBE3A	rs587781234	II
UBE3A	rs587781238	II
UBE3A	rs587781241	GG
UBE3A	rs587784526	AA
UBE3A	rs587784520	II
UBE3A	rs587782919	TT
UBE3A	rs587784518	TT
UBE3A	rs587784516	CC
UBE3A	rs587784515	AA
UBE3A	rs587784514	CC
UBE3A	rs587784512	II
UBE3A	rs587784509	II
UBE3A	rs587784508	CC
UBE3A	rs587784533	CC
UBE3A	rs587784532	II
UBE3A	rs587784530	II
MECP2	rs28935468	GG
MECP2	rs63749748	
MECP2	rs28934906	GG
MECP2	rs61751362	GG
UBE3A	rs398124440	DD
UBE3A	rs587783097	GG
UBE3A	rs587784527	II
UBE3A	rs587784529	II

Carrier Status

ANTITHROMBIN III DEFICIENCY; AT3D

Deficiency of antithrombin III is a major risk factor for venous thromboembolic disease. Two categories of AT-III deficiency have been defined on the basis of AT-III antigen levels in the plasma of affected individuals. The majority of AT-III deficiency families belong in the type I (classic) deficiency group and have a quantitatively abnormal phenotype in which antigen and heparin cofactor levels are both reduced to about 50% of normal. The second category of AT-III deficiency has been termed type II (functional) deficiency. Affected individuals from these kindreds produce dysfunctional AT-III molecules; they have reduced heparin cofactor activity levels (about 50% of normal) but levels of AT-III antigen are often normal or nearly normal. The 2 categories of antithrombmin III deficiency have been classified further. Type I (low functional and immunologic antithrombin) has been subdivided into subtype Ia (reduced levels of normal antithrombin), and type Ib (reduced levels of antithrombin and the presence of low levels of a variant).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613118

Your genetic map

Gene	SNP	Genotype
SERPINC1	rs28929469	GG

Carrier Status

ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 10; ARVD10

Familial isolated arrhythmogenic right ventricular dysplasia (ARVC) is the familial autosomal dominant form of ARVC, a heart muscle disease characterized by life-threatening ventricular arrhythmias with left bundle branch block configuration that may manifest with palpitations, ventricular tachycardia, syncope and sudden fatal attacks, and that is due to dystrophy and fibro-fatty replacement of the right ventricular myocardium that may lead to right ventricular aneurysms.

Your genetic map

Gene	SNP	Genotype
DSG2	rs121913007	GG
DSG2	rs397516709	TT
DSG2	rs121913006	GG
DSG2	rs121913008	GG
DSG2	rs397514038	AA

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610193

Carrier Status

AURICULOCONDYLAR SYNDROME 1; ARCND1

Auriculo-condylar syndrome (ACS) presents with bilateral external ear malformations ('question mark' ears), mandibular condyle hypoplasia, microstomia, micrognathia, microglossia and facial asymmetry. Additional manifestations include hypotonia, ptosis, cleft palate, puffy cheeks, developmental delay, impaired hearing and respiratory distress.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/602483

Your genetic map

Gene	SNP	Genotype
GNAI3	rs387907178	GG
PLCB4	rs387907179	AA
PLCB4	rs397514480	AA
PLCB4	rs397514481	GG
PLCB4	rs397514482	CC
PLCB4	rs397514483	AA

Carrier Status

HYPOPHOSPHATEMIC RICKETS, AUTOSOMAL DOMINANT; ADHR

Autosomal dominant hypophosphatemic rickets (ADHR) is a hereditary renal phosphate-wasting disorder characterized by hypophosphatemia, rickets and/or osteomalacia. Less than 100 cases have been described. Clinical manifestations depend on the age of onset (childhood, adolescence, even adulthood) and on the severity of hypophosphatemia.

Your genetic map

Gene	SNP	Genotype
FGF23	rs193922701	CC
FGF23	rs193922702	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/193100

Carrier Status

BARDET-BIEDL SYNDROME 1; BBS1

Bardet-Biedl syndrome (BBS) is a ciliopathy with multisystem involvement.Its prevalence in Europe is estimated at between 1/125,000 and 1/175,000. This disorder is characterized by a combination of clinical signs: obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, and learning disabilities, many of which appear several years after disease onset.

Your genetic map

Gene	SNP	Genotype
BBS1	rs193922709	GG
BBS2	rs193922710	GG
BBS9	rs762511626	TT
BBS1	rs121917777	GG
BBS1	rs587777829	GG
BBS1	rs113624356	TT
BBS7	rs119466002	GG
BBS10	rs148374859	GG
BBS10	rs727503818	
BBS10	rs761101213	II
BBS10	rs549625604	DD
BBS2	rs193922711	II
BBS9	rs749974697	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/209900

Carrier Status

MUSCULAR DYSTROPHY, BECKER TYPE; BMD

Becker muscular dystrophy (BMD) is a neuromuscular disease characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle. BMD primarily affects males with an estimated incidence of 1/18,000 to 1/31,000 male births. Females are usually asymptomatic but a small percentage of female carriers manifest milder forms of the disease (symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers; see this term).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300376

Your genetic map

Gene	SNP	Genotype
DMD	rs104894787	GG
DMD	rs128626251	GG
DMD	rs104894797	GG
DMD	rs128627256	GG
DMD	rs398123827	GG
DMD	rs398123828	CC
DMD	rs398123830	CC
DMD	rs398123834	CC
DMD	rs398123837	II
DMD	rs398123840	CC
DMD	rs398123852	GG
DMD	rs398123854	DD
DMD	rs72468700	TT
DMD	rs398123857	II
DMD	rs398123861	GG
DMD	rs398123862	CC
DMD	rs398123863	II
DMD	rs398123865	GG
DMD	rs398123867	GG
DMD	rs398123870	GG
DMD	rs398123872	GG
DMD	rs398123875	II
DMD	rs398123882	II
DMD	rs398123883	GG
DMD	rs398123884	CC
DMD	rs398123887	CC
DMD	rs398123888	GG
DMD	rs398123895	II
DMD	rs398123903	GG
DMD	rs398123909	CC
DMD	rs398123913	II

Carrier Status

BETA-THALASSEMIA

Beta-thalassemia (BT) is characterized by deficiency (Beta+) or absence (Beta0) of synthesis of the beta globin chains of hemoglobin (Hb).Exact prevalence is unknown but annual incidence at birth of symptomatic BT is estimated at 1/100,000 worldwide. The disease was initially described in the Mediterranean basin but severe forms of BT frequently occur throughout the Middle East, South East Asia, India and China. Population migrations have lead to global distribution of the disease.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613985

Your genetic map

Gene	SNP	Genotype
HBB	rs33994806	GG
HBB	rs34305195	TT
HBB	rs34937014	
HBB	rs35703285	AA
HBB	rs33956879	AA
HBB	rs33960103	CC
HBB	rs34527846	AA
HBB	rs33941377	GG
HBB	rs33941377	GG
HBB	rs33978907	AA
HBB	rs33944208	GG
HBB	rs33941377	GG
HBB	rs34598529	TT
HBB	rs34999973	GG
HBB	rs33960103	CC
HBB	rs34451549	GG
HBB	rs35004220	CC
HBB	rs33974936	CC
HBB	rs35497102	II
HBB	rs80356820	II
HBB	rs281864901	
HBB	rs33971440	CC
HBB	rs33971440	CC
HBB	rs33915217	CC
HBB	rs33951465	AA
HBB	rs63751208	GG
HBB	rs33915217	CC
HBB	rs33941849	

Carrier Status

BLOOM SYNDROME; BLM

Bloom syndrome (BSyn) is a rare chromosomal breakage syndrome characterized by a marked genetic instability associated with pre- and postnatal growth retardation, facial sun-sensitive telangiectatic erythema, increased susceptibility to infections, and predisposition to cancer. Overall prevalence is unknown, but in the Ashkenazi Jewish population it is estimated at approximately 1/ 48,000 births.

Your genetic map

Gene	SNP	Genotype
BLM	rs367543012	DD
BLM	rs148969222	GG
BLM	rs367543014	
BLM	rs200389141	CC
BLM	rs587779884	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/210900

Carrier Status

BRUGADA SYNDROME 1; BRGDA1

Brugada syndrome (BrS) manifests with ST segment elevation in right precordial leads (V1 to V3), incomplete or complete right bundle branch block, and susceptibility to ventricular tachyarrhythmia and sudden death. BrS is an electrical disorder without overt myocardial abnormalities. As the aberrant ECG pattern is often intermittent and shows a distinct regionality, it is difficult to estimate the prevalence of the disease. The largest cohorts in far Eastern countries portray a prevalence of 1/700-1/800. The prevalence in Europe and the United States is lower: 1/3,300 to 1/10,000. Analysis of worldwide literature suggests a prevalence of the type 1 (diagnostic) ECG pattern of 1/1000.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601144

Your genetic map

Gene	SNP	Genotype
SCN5A	rs137854604	GG
SCN5A	rs28937318	CC
SCN5A	rs137854612	CC
SCN5A	rs137854601	CC
SCN5A	rs199473082	GG

Carrier Status

CARDIOFACIOCUTANEOUS SYNDROME 1; CFC1

Cardiofaciocutaneous (CFC) syndrome is a RASopathy characterized by craniofacial dysmorphology, congenital heart disease, dermatological abnormalities (most commonly hyperkeratotic skin and sparse, curly hair), growth retardation and intellectual disability. Around 300 cases have been published in the literature to date. Prevalence has been estimated at 1/810,000 people in Japan.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/115150

Your genetic map

Gene	SNP	Genotype
BRAF	rs180177039	TT
BRAF	rs180177036	CC
BRAF	rs180177034	CC
BRAF	rs180177035	TT
BRAF	rs180177040	TT
BRAF	rs180177038	CC
BRAF	rs180177037	TT
MAP2K2	rs730880517	TT

Carrier Status

CARDIOMYOPATHY, DILATED, 1S; CMD1S

Familial isolated dilated cardiomyopathy is a rare, genetically heterogeneous cardiac disease characterized by dilatation leading to systolic and diastolic dysfunction of the left and/or right ventricles, causing heart failure or arrhythmia.

Your genetic map

Gene	SNP	Genotype
MYH7	rs397516089	CC
MYH7	rs371898076	CC
TTN	rs761807131	CC
MYH7	rs121913642	AA
MYH7	rs727503253	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613426

Carrier Status

CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 1; CMH1

Hypertrophic cardiomyopathy (HCM) is typically defined by the presence of unexplained left ventricular hypertrophy (LVH). Such LVH occurs in a non-dilated ventricle in the absence of other cardiac or systemic disease capable of producing the observed magnitude of increased LV wall thickness, such as pressure overload (e.g., long-standing hypertension, aortic stenosis) or storage/infiltrative disorders (e.g., Fabry disease, amyloidosis). The clinical manifestations of HCM range from asymptomatic LVH to progressive heart failure to sudden cardiac death (SCD), and vary from individual to individual even within the same family. Common symptoms include shortness of breath (particularly with exertion), chest pain, palpitations, orthostasis, presyncope, and syncope. Most often the LVH of HCM becomes apparent during adolescence or young adulthood, although it may also develop late in life, in infancy, or in childhood.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/192600

Your genetic map

Gene	SNP	Genotype
MYBPC3	rs730880649	DD
MYBPC3	rs121909374	CC
MYBPC3	rs397515963	DD
MYH7	rs121913627	CC
MYH7	rs121913628	CC
MYH7	rs121913631	GG
MYH7	rs121913641	CC
MYH7	rs397516155	II
MYH7	rs397516161	TT
MYH7	rs121913637	GG
MYH7	rs763538103	
MYH7	rs767148171	TT
MYH7	rs730880876	CC
MYH7	rs727505202	AA
MYBPC3	rs190228518	GG
MYH7	rs121913625	GG
MYH7	rs397516153	GG
MYH7	rs121913632	CC
MYH7	rs3218714	GG
MYH7	rs36211715	CC
MYH7	rs267606908	TT
MYH7	rs3218716	CC
MYH7	rs397516209	CC
MYH7	rs727503261	AA
MYH7	rs121913638	CC
MYH7	rs121913654	AA
MYH7	rs727504299	GG
MYBPC3	rs397515970	DD
MYH7	rs397516202	CC
MYH7	rs397516212	CC
MYH7	rs121913633	CC

Carrier Status

CEROID LIPOFUSCINOSIS, NEURONAL, 1; CLN1

Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited progressive degenerative brain diseases characterized clinically by a decline of mental and other capacities, epilepsy, and vision loss through retinal degeneration, and histopathologically by intracellular accumulation of an autofluorescent material, ceroid lipofuscin, in the neuronal cells in the brain and in the retina.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/256730

Your genetic map

Gene	SNP	Genotype
PPT1	rs386833655	CC
PPT1	rs386833650	GG
PPT1	rs137852700	GG
PPT1	rs137852695	TT
PPT1	rs137852696	TT
PPT1	rs137852699	AA
PPT1	rs386833642	AA
PPT1	rs386833650	GG

Carrier Status

CEROID LIPOFUSCINOSIS, NEURONAL, 7; CLN7

The neuronal ceroid-lipofuscinoses (NCLs) are a group of inherited, neurodegenerative, lysosomal storage disorders characterized by progressive intellectual and motor deterioration, seizures, and early death. Visual loss is a feature of most forms. Clinical phenotypes have been characterized traditionally according to the age of onset and order of appearance of clinical features into infantile, late-infantile, juvenile, adult, and Northern epilepsy (also known as progressive epilepsy with mental retardation [EPMR]). There is however genetic and allelic heterogeneity; a proposed new nomenclature and classification system has been developed to take into account both the responsible gene and the age at disease onset; for example, CLN1 disease, infantile onset and CLN1 disease, juvenile onset are both caused by pathogenic variants in PPT1 but with differing age of onset. The most prevalent NCLs are CLN3 disease, classic juvenile and CLN2 disease, classic late infantile (although prevalence varies by ethnicity and country of family origin): CLN2 disease, classic late infantile. The first symptoms typically appear between age two and four years.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610951

Your genetic map

Gene	SNP	Genotype
MFSD8	rs587778809	AA
MFSD8	rs118203978	TT

Carrier Status

CHARCOT-MARIE-TOOTH DISEASE, TYPE 4C; CMT4C

Charcot-Marie-Tooth disease type 4C (CMT4C) is a subtype of Charcot-Marie-Tooth type 4 characterized by childhood or adolescent-onset of a relatively mild, demyelinating sensorimotor neuropathy that contrasts with a severe, rapidly progressing, early-onset scoliosis, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and often foot deformity). A wide spectrum of nerve conduction velocities are observed and cranial nerve involvement and kyphoscoliosis have also been reported.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601596

Your genetic map

Gene	**SNP**	**Genotype**
SH3TC2	rs80338931	GG
SH3TC2	rs80338934	GG
SH3TC2	rs80338926	GG
SH3TC2	rs80338933	GG

Carrier Status

CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1

Brachytelephalangic chondrodysplasia punctata (BCDP) is a form of nonrhizomelic chondrodysplasia punctata, a primary bone dysplasia, characterized by hypoplasia of the distal phalanges of the fingers, nasal hypoplasia, epiphyseal stippling appearing in the first year of life, and mild and nonrhizomelic shortness of the long bones.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/302950

Your genetic map

Gene	SNP	Genotype
ARSE	rs145946864	GG

Carrier Status

GRANULOMATOUS DISEASE, CHRONIC, X-LINKED; CDGX

Chronic granulomatous disease (CGD) is a rare primary immunodeficiency, mainly affecting phagocytes, which is characterized by an increased susceptibility to severe and recurrent bacterial and fungal infections, along with the development of granulomas. The average worldwide birth prevalence is estimated at 1/ 217,000. CGD can present at any age but is most commonly diagnosed before the age of 5 years.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/306400

Your genetic map

Gene	SNP	Genotype
CYBB	rs193922449	GG
CYBB	rs193922445	DD
CYBB	rs193922446	II
CYBB	rs193922448	GG

Carrier Status

ADRENAL HYPOPLASIA, CONGENITAL; AHC

X-linked adrenal hypoplasia congenita (X-linked AHC) is characterized by infantile-onset acute primary adrenal insufficiency at an average age of three weeks in approximately 60% of affected individuals. Onset in approximately 40% is in childhood. A few individuals present in adulthood with delayed-onset adrenal failure or partial hypogonadism due to partial forms of X-linked AHC. Adrenal insufficiency typically presents acutely in male infants with vomiting, feeding difficulty, dehydration, and shock caused by a salt-wasting episode. Hypoglycemia (sometimes presenting with seizures) or isolated salt loss may be the first symptom of X-linked AHC. Cortisol may be low or within the normal range, which is inappropriately low for a sick child. In older children, adrenal failure may be precipitated by intercurrent illness or stress. If untreated, adrenal insufficiency is rapidly lethal as a result of hyperkalemia, acidosis, hypoglycemia, and shock. Affected males typically have delayed puberty (onset age >14 years) or arrested puberty caused by hypogonadotropic hypogonadism (HH).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300200

Your genetic map

Gene	SNP	Genotype
NR0B1	rs386134262	AA
NR0B1	rs386134263	GG

Carrier Status

NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C; CSNB1C

Congenital stationary night blindness (CSNB) refers to a non-progressive group of retinal disorders characterized by night or dim light vision disturbance or delayed dark adaptation, poor visual acuity, nystagmus, strabismus, normal color vision and fundus abnormalities. Two forms of CSNB are recognized: complete and incomplete CSNB (CSNB1 and CSNB2 respectively).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613216

Your genetic map

Gene	SNP	Genotype
TRPM1	rs387906862	GG
TRPM1	rs778390089	II
TRPM1	rs191205969	AA
TRPM1	rs369742878	GG

Carrier Status

CORNELIA DE LANGE SYNDROME 1; CDLS1

Cornelia de Lange syndrome (CdLS) is a multisystem disorder with variable expression marked by a characteristic facial dysmorphism, variable degrees of intellectual deficit, severe growth retardation beginning before birth (2nd trimester), abnormal hands and feet, and various other malformations (heart, kidney etc.).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/122470

Your genetic map

Gene	SNP	Genotype
NIPBL	rs121918267	CC
NIPBL	rs121918269	CC
NIPBL	rs398124470	TT
NIPBL	rs80358367	CC
NIPBL	rs80358382	II
NIPBL	rs80358364	II
NIPBL	rs80358386	II
NIPBL	rs80358369	TT
NIPBL	rs80358372	II
NIPBL	rs80358380	GG
NIPBL	rs80358366	GG
NIPBL	rs80358373	AA
NIPBL	rs80358360	CC
NIPBL	rs80358363	GG
NIPBL	rs80358361	II
NIPBL	rs80358376	CC
NIPBL	rs80358370	CC
NIPBL	rs80358371	DD
NIPBL	rs587783937	GG
NIPBL	rs587784009	GG
NIPBL	rs587784011	GG
NIPBL	rs587784012	AA
NIPBL	rs587784060	II
NIPBL	rs587783886	GG
NIPBL	rs587783893	II
NIPBL	rs587783895	TT
NIPBL	rs587783917	II
NIPBL	rs587783922	AA
NIPBL	rs587783927	GG
NIPBL	rs587783928	GG
NIPBL	rs587783988	CC

Carrier Status

COSTELLO SYNDROME; CSTLO

Costello syndrome (CS) is a rare multisystemic disorder characterized by failure to thrive, short stature, developmental delay or intellectual disability, joint laxity, soft skin, and distinctive facial features. Cardiac and neurological involvement is common and there is an increased lifetime risk of certain tumors. The estimated number of patients worldwide is 300. Estimated birth prevalence has been reported to be 1/300,000 to 1/1.25 million.

Your genetic map

Gene	SNP	Genotype
HRAS	rs104894226	CC
HRAS	rs121917758	GG
HRAS	rs104894230	CC
HRAS	rs104894230	CC
HRAS	rs121917757	GG
HRAS	rs727503093	CC
HRAS	rs104894227	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/218040

Carrier Status

CYSTIC FIBROSIS; CF

Cystic fibrosis (CF) is a genetic disorder characterized by the production of sweat with a high salt content and mucus secretions with an abnormal viscosity.It is the most common genetic disorder among Caucasian children. The incidence varies between populations: the condition is considerably less common in Asian and African populations than in the white populations of Europe and North America, with variation within each country. The exact prevalence in Europe is unknown, but estimates range between 1/8,000 and 1/10,000 individuals.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/219700

Your genetic map

Gene	SNP	Genotype
CFTR	rs75541969	GG
CFTR	rs77101217	CC
CFTR	rs121908788	DD
CFTR	rs121908811	II
CFTR	rs76649725	CC
CFTR	rs267606722	GG
CFTR	rs387906361	II
CFTR	rs74767530	CC
CFTR	rs387906362	AA
CFTR	rs121908776	II
CFTR	rs121909012	CC
CFTR	rs79850223	CC
CFTR	rs121908804	II
CFTR	rs121908754	CC
CFTR	rs121909017	CC
CFTR	rs80055610	GG
CFTR	rs121909019	GG
CFTR	rs141158996	GG
CFTR	rs143570767	GG
CFTR	rs78194216	CC
CFTR	rs121908748	GG
CFTR	rs387906369	GG
CFTR	rs121909025	GG
CFTR	rs121909026	CC
CFTR	rs121908751	GG
CFTR	rs121908787	
CFTR	rs121908751	GG
CFTR	rs77409459	CC
CFTR	rs78802634	GG
CFTR	rs76554633	CC
CFTR	rs75115087	AA

Carrier Status

DANON DISEASE

Glycogen storage disease due to LAMP-2 (Lysosomal-Associated Membrane Protein 2) deficiency is a lysosomal glycogen storage disease characterised by severe cardiomyopathy and variable degrees of muscle weakness, frequently associated with intellectual deficit. More than 20 families have been described in the literature so far.

Your genetic map

Gene	SNP	Genotype
LAMP2	rs397516743	TT
LAMP2	rs727504742	CC
LAMP2	rs727504557	II
LAMP2	rs727504597	II
LAMP2	rs727504600	II
LAMP2	rs104894858	CC
LAMP2	rs397516740	CC
LAMP2	rs397516751	II
LAMP2	rs727503118	GG
LAMP2	rs730880483	GG
LAMP2	rs193922649	II

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300257

Carrier Status

DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A

Nonsyndromic hearing loss and deafness (DFNB1) is characterized by congenital non-progressive mild-to-profound sensorineural hearing impairment. No other associated medical findings are present.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/220290

Your genetic map

Gene	SNP	Genotype
GJB2	rs80338943	II
GJB2	rs104894413	CC
GJB2	rs111033296	GG
GJB2	rs772264564	AA
GJB2	rs587783646	II
GJB2	rs80338947	II
GJB2	rs111033299	CC
GJB2	rs111033294	TT
GJB2	rs143343083	GG
GJB2	rs80338948	GG
GJB2	rs104894398	CC
GJB2	rs72474224	CC
GJB2	rs80338940	CC
GJB2	rs111033253	II
GJB2	rs80338944	CC
GJB2	rs80338950	CC
GJB2	rs111033451	GG
GJB2	rs397516874	GG
GJB2	rs111033204	II
GJB2	rs111033217	TT
GJB2	rs398123814	

Carrier Status

DEAFNESS, AUTOSOMAL RECESSIVE 31; DFNB31

Mustapha et al. (2002) described a consanguineous Palestinian family from Jordan in which 6 members had profound prelingual nonsyndromic hearing loss. Tlili et al. (2005) reported a consanguineous Tunisian family in which 4 sibs had congenital profound hearing loss (greater than 90 dB) but were otherwise healthy with no dysmorphic or other abnormal findings indicative of syndromic deafness. No vestibular defects were detected.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/607084

Your genetic map

Gene	SNP	Genotype
WHRN	rs779760634	CC

Carrier Status

DEAFNESS, AUTOSOMAL RECESSIVE 7; DFNB7

Prelingual non-syndromic genetic deafness is a rare, genetically highly heterogeneous otorhinolaryngologic disease, resulting from inner and/or middle ear or hearing nerve anomalies, typically characterized by bilateral, severe to profound hearing loss (mean sensorineural hearing impairment of 60 dB or more for 500-, 1,000-, and 2,000-Hz frequency tones in the better ear) which occurs before the onset of speech development and is not associated with visible external ear abnormalities or any other medical problems. It is usually nonprogressive and impedes oral language acquisition.

Your genetic map

Gene	SNP	Genotype
TMC1	rs121908073	CC
TMC1	rs151001642	CC
TMC1	rs370088722	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/600974

Carrier Status

DEAFNESS, AUTOSOMAL RECESSIVE 9; DFNB9

Postlingual non-syndromic genetic deafness is a rare, genetically highly heterogeneous otorhinolaryngologic disease, resulting from inner and/or middle ear or hearing nerve anomalies, typically characterized by progressive, bilateral, moderate to profound hearing loss (mean sensorineural hearing impairment equal to 40 dB or more for 500-, 1,000-, and 2,000-Hz frequency tones in the better ear) which occurs after the onset of speech development and is not associated with visible external ear abnormalities or any other medical problems. Language development is not initially significantly delayed.

Your genetic map

Gene	SNP	Genotype
OTOF	rs80356590	GG
OTOF	rs80356591	II
OTOF	rs111033373	CC
OTOF	rs397515607	II
OTOF	rs80356593	GG
OTOF	rs397515591	CC
OTOF	rs199766465	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601071

Carrier Status

MANNOSIDOSIS, ALPHA B, LYSOSOMAL; MANSA

Alpha-mannosidosis is an inherited lysosomal storage disorder characterized by immune deficiency, facial and skeletal abnormalities, hearing impairment, and intellectual deficit. It occurs in approximately 1 in 500,000 live births.

Your genetic map

Gene	SNP	Genotype
MAN2B1	rs121434331	GG
MAN2B1	rs80338677	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/248500

Carrier Status

CARDIOMYOPATHY, DILATED, 1A; CMD1A

Nonsyndromic isolated dilated cardiomyopathy (DCM) is characterized by left ventricular enlargement and systolic dysfunction, a reduction in the myocardial force of contraction. DCM usually presents with any one of the following: Heart failure with symptoms of congestion (edema, orthopnea, paroxysmal nocturnal dyspnea) and/or reduced cardiac output (fatigue, dyspnea on exertion). Arrhythmias and/or conduction system disease. Thromboembolic disease (from left ventricular mural thrombus) including stroke.

Your genetic map

Gene	SNP	Genotype
LMNA	rs56984562	CC
LMNA	rs60682848	CC
LMNA	rs59026483	CC
LMNA	rs28933091	CC
LMNA	rs28933093	GG
LMNA	rs61195471	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/115200

Carrier Status

DUBIN-JOHNSON SYNDROME; DJS

Dubin-Johnson syndrome (DJS) is a benign, inherited liver disorder characterized clinically by chronic, predominantly conjugated, hyperbilirubinemia and histopathologically by black-brown pigment deposition in parenchymal liver cells. Prevalence in the general population is unknown. DJS affects individuals of all ethnic origins but is most common among Iranian or Moroccan Jews, in which, due to founder mutations, it has been reported to occur in up to 1/1,300 individuals.

Your genetic map

Gene	SNP	Genotype
ABCC2	rs72558201	AA
ABCC2	rs146405172	GG
ABCC2	rs17222547	CC
ABCC2	rs34937870	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/237500

Carrier Status

EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 2; EIEE2

Early infantile epileptic encephalopathy (EIEE), or Ohtahara syndrome, is one of the most severe forms of age-related epileptic encephalopathies, characterized by the onset of tonic spasms within the first 3 months of life that can be generalized or lateralized, independent of the sleep cycle and that can occur hundreds of times per day, leading to psychomotor impairment and death. Incidence has been estimated at 1/100 000 births in Japan and 1/50,000 births in the U.K.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300672

Your genetic map

Gene	SNP	Genotype
CDKL5	rs61753251	II
CDKL5	rs62653623	CC
CDKL5	rs267608643	CC
CDKL5	rs267608395	CC
CDKL5	rs267608493	CC
CDKL5	rs267608659	CC
CDKL5	rs267608663	CC
CDKL5	rs267608500	AA
CDKL5	rs587783406	AA
CDKL5	rs587783399	GG
CDKL5	rs587783405	CC
CDKL5	rs587783398	
CDKL5	rs587783401	
CDKL5	rs267608501	CC
CDKL5	rs267606715	GG
CDKL5	rs267608429	AA
CDKL5	rs267608653	GG
CDKL5	rs267608662	II
CDKL5	rs267608472	CC
CDKL5	rs267608497	
CDKL5	rs267608420	DD
CDKL5	rs267608532	AA
CDKL5	rs587783131	GG
CDKL5	rs587783158	CC
CDKL5	rs267608437	CC

Carrier Status

MYOCLONIC EPILEPSY OF LAFORA

Lafora disease (LD) is a rare, inherited, severe, progressive myoclonic epilepsy characterized by myoclonus and/or generalized seizures, visual hallucinations (partial occipital seizures), and progressive neurological decline.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/254780

Your genetic map

Gene	SNP	Genotype
NHLRC1	rs28940576	GG
NHLRC1	rs587776542	II
EPM2A	rs104893950	GG
NHLRC1	rs769301934	CC

Carrier Status

ERYTHROCYTOSIS, FAMILIAL, 2; ECYT2

Familial erythrocytosis-2 is an autosomal recessive disorder characterized by increased red blood cell mass, increased serum levels of erythropoietin (EPO; 133170), and normal oxygen affinity. Patients with ECYT2 carry a high risk for peripheral thrombosis and cerebrovascular events (Cario, 2005). Familial erythrocytosis-2 has features of both primary and secondary erythrocytosis. In addition to increased circulating levels of EPO, consistent with a secondary, extrinsic process, erythroid progenitors are also hypersensitive to EPO, consistent with a primary, intrinsic process.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/263400

Your genetic map

Gene	SNP	Genotype
VHL	rs104893826	GG
VHL	rs5030818	CC
VHL	rs104893830	GG
VHL	rs5030809	TT
VHL	rs5030821	GG
VHL	rs5030810	CC
VHL	rs730882035	GG

Carrier Status

FABRY DISEASE

Fabry disease (FD) is a progressive, inherited, multisystemic lysosomal storage disease characterized by specific neurological, cutaneous, renal, cardiovascular, cochleo-vestibular and cerebrovascular manifestations. Annual incidence is reported to be 1 in 80,000 live births but this figure may underestimate disease prevalence. When late-onset variants of the disease are considered, a prevalence of approximately 1 in 3,000 has been suggested. FD is pan-ethnic.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/301500

Your genetic map

Gene	SNP	Genotype
GLA	rs104894828	CC
GLA	rs727503950	AA
GLA	rs104894827	GG
GLA	rs104894835	TT
GLA	rs28935492	CC
GLA	rs28935493	CC
GLA	rs104894843	GG
GLA	rs397515870	GG
GLA	rs398123199	GG
GLA	rs398123201	AA
GLA	rs398123203	TT
GLA	rs398123205	CC
GLA	rs398123206	CC
GLA	rs398123207	CC
GLA	rs113173389	CC
GLA	rs372966991	CC
GLA	rs398123210	TT
GLA	rs398123211	TT
GLA	rs398123214	II
GLA	rs398123216	CC
GLA	rs398123217	TT
GLA	rs398123219	CC
GLA	rs398123220	CC
GLA	rs398123221	GG
GLA	rs398123222	GG
GLA	rs140329381	TT
GLA	rs398123223	AA
GLA	rs398123225	II
GLA	rs398123226	GG
GLA	rs398123227	CC
GLA	rs398123229	II

Carrier Status

FAMILIAL ADENOMATOUS POLYPOSIS 1; FAP1

Familial adenomatous polyposis (FAP) is characterized by the development of hundreds to thousands of adenomas in the rectum and colon during the second decade of life.FAP has a birth incidence of about 1/8,300, manifests equally in both sexes, and accounts for less than 1% of colorectal cancer (CRC) cases. In the EU, prevalence is estimated at 1/11.300 -1/37.600.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/175100

Your genetic map

Gene	SNP	Genotype
APC	rs121913224	
APC	rs137854568	CC
APC	rs137854573	CC
APC	rs121913333	CC
APC	rs387906230	TT
APC	rs397515732	DD
APC	rs397515733	II
APC	rs727504420	II
APC	rs559510809	GG
APC	rs137854580	CC
APC	rs397514031	GG
APC	rs587779783	CC
APC	rs730881228	II
APC	rs730881273	II
APC	rs397515734	CC
APC	rs587779352	II
APC	rs587779353	II
APC	rs398123117	CC
APC	rs587779780	CC
APC	rs62619935	CC
APC	rs587781392	CC
APC	rs587782303	II
APC	rs587782557	II
APC	rs775126020	CC
APC	rs387906238	II
APC	rs398123116	GG
APC	rs398123119	GG
APC	rs398123120	II
APC	rs398123121	CC
APC	rs398123122	DD
APC	rs587779786	AA

Carrier Status

CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 2; CMH2

Hypertrophic cardiomyopathy (HCM) is typically defined by the presence of unexplained left ventricular hypertrophy (LVH). Such LVH occurs in a non-dilated ventricle in the absence of other cardiac or systemic disease capable of producing the observed magnitude of increased LV wall thickness, such as pressure overload (e.g., long-standing hypertension, aortic stenosis) or storage/infiltrative disorders (e.g., Fabry disease, amyloidosis). The clinical manifestations of HCM range from asymptomatic LVH to progressive heart failure to sudden cardiac death (SCD), and vary from individual to individual even within the same family. Common symptoms include shortness of breath (particularly with exertion), chest pain, palpitations, orthostasis, presyncope, and syncope. Most often the LVH of HCM becomes apparent during adolescence or young adulthood, although it may also develop late in life, in infancy, or in childhood.

Your genetic map

Gene	SNP	Genotype
TNNT2	rs121964855	AA
TNNT2	rs397516463	GG
TNNT2	rs111377893	CC
TNNT2	rs121964856	CC
TNNT2	rs397516456	GG
TNNT2	rs397516457	CC
TNNT2	rs727504247	CC
TNNT2	rs397516470	II

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/115195

Carrier Status

FAMILIAL MEDITERRANEAN FEVER; FMF

Familial Mediterranean fever (FMF) is an autoinflammatory disorder characterized by recurrent short episodes of fever and serositis resulting in pain in the abdomen, chest, joints and muscles. FMF is primarily found in the south-eastern Mediterranean area. Populations having a high prevalence (1/200-1/1000) of the disease are non-Ashkenazi Jews, Turks, Armenians and Arabs. It is not considered rare in Italy, Greece or Spain.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/249100

Your genetic map

Gene	SNP	Genotype
MEFV	rs61752717	TT
MEFV	rs28940579	AA
MEFV	rs28940580	CC
MEFV	rs104895085	CC
MEFV	rs28940580	CC
MEFV	rs104895093	

Carrier Status

THYROID CARCINOMA, FAMILIAL MEDULLARY; MTC

Multiple endocrine neoplasia type 2 (MEN2) is a multiple endocrine neoplasia, a polyglandular cancer syndrome characterized by the occurrence of medullary thyroid carcinoma (MTC), pheochromocytoma (PCC; see these terms), in one variant, primary hyperparathyroidism (PHPT). There are three forms: MEN2A, MEN2B, and familial medullary thyroid carcinoma (FMTC). The total prevalence of all MEN2 variants is approximately 1/35,000. Of the three MEN2 subtypes, MEN2A accounts for about 70%-80% of cases, familial medullary thyroid carcinoma (FMTC) for 10 -20%, and MEN2B for 5%.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/155240

Your genetic map

Gene	SNP	Genotype
RET	rs75234356	TT
RET	rs77503355	GG
RET	rs79781594	GG
RET	rs75030001	GG
RET	rs77503355	GG

Carrier Status

FANCONI ANEMIA, COMPLEMENTATION GROUP O; FANCO

Fanconi anemia (FA) is a hereditary DNA repair disorder characterized by progressive pancytopenia with bone marrow failure, variable congenital malformations and predisposition to develop hematological or solid tumors.

Your genetic map

Gene	SNP	Genotype
RAD51C	rs779582317	AA
RAD51C	rs587782036	GG
RAD51C	rs267606997	GG
RAD51C	rs587782036	GG
RAD51C	rs587782170	
RAD51C	rs587782818	CC
RAD51C	rs730881940	
RAD51C	rs200293302	CC
RAD51C	rs730881931	TT
RAD51C	rs770637624	CC
RAD51C	rs779582317	AA

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613390

Carrier Status

NEPHROTIC SYNDROME, TYPE 1; NPHS1

Congenital nephrotic syndrome, Finnish type is characterised by protein loss beginning during foetal life. This type of nephrotic syndrome is more frequent in Finland (with an incidence of 1 in 8 200 births) but it is also observed in various ethnic groups worldwide.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/256300

Your genetic map

Gene	SNP	Genotype
NPHS1	rs386833895	CC
NPHS1	rs386833909	GG
NPHS1	rs386833915	GG

Carrier Status

GAUCHER DISEASE, TYPE I

Gaucher disease type 1 is the chronic non-neurological form of Gaucher disease (GD; see this term) characterized by organomegaly, bone involvement and cytopenia. It represents around 90% of all cases of GD with an estimated prevalence of 1/100,000 in the general population.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/230800

Your genetic map

Gene	SNP	Genotype
GBA	rs80356772	CC
GBA	rs80356769	CC
GBA	rs364897	TT
GBA	rs398123526	CC
GBA	rs398123528	CC
GBA	rs121908312	CC

Carrier Status

GLUT1 DEFICIENCY SYNDROME 1; GLUT1DS1

Glucose transporter type 1 (GLUT1) deficiency syndrome is characterized by an encephalopathy marked by childhood epilepsy that is refractory to treatment, deceleration of cranial growth leading to microcephaly, psychomotor retardation, spasticity, ataxia, dysarthria and other paroxysmal neurological phenomena often occurring before meals. Symptoms appear between the age of 1 and 4 months, following a normal birth and gestation.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/606777

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs587784391	II
SLC2A1	rs587784397	GG
SLC2A1	rs587784390	TT
SLC2A1	rs587784393	II
SLC2A1	rs75485205	GG
SLC2A1	rs587784396	GG
SLC2A1	rs80359823	GG

Carrier Status

GLUTARIC ACIDEMIA I; GA1

Glutaryl-CoA dehydrogenase (GCDH) deficiency (GDD) is an autosomal recessive neurometabolic disorder clinically characterized by encephalopathic crises resulting in striatal injury and a severe dystonic dyskinetic movement disorder. Worldwide prevalence is estimated at 1 in 100,000 births. GDD is more prevalent in the old order Amish community, Canadian Oji-Cree natives, Irish travelers and Lumbee Native Americans.

Your genetic map

Gene	SNP	Genotype
GCDH	rs121434369	CC
GCDH	rs121434366	TT
GCDH	rs199999619	AA
GCDH	rs149120354	TT
GCDH	rs121434371	GG
GCDH	rs121434372	GG
GCDH	rs398123194	AA
GCDH	rs398123195	GG
GCDH	rs147611168	GG
GCDH	rs141437721	AA
GCDH	rs372983141	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/231670

Carrier Status

MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD

Multiple acyl-CoA dehydrogenation deficiency (MADD) is a disorder of fatty acid and amino acid oxidation and is a clinically heterogeneous disorder ranging from a severe neonatal presentation with metabolic acidosis, cardiomyopathy and liver disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure. Birth prevalence is estimated at 1/200,000 but great variation is seen between countries/ethnicities.

Your genetic map

Gene	SNP	Genotype
ETFDH	rs377686388	TT
ETFDH	rs398124152	CC
ETFDH	rs398124151	GG
ETFDH	rs398124153	II
ETFA	rs727503918	AA
ETFDH	rs727503919	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/231680

Carrier Status

GLYCOGEN STORAGE DISEASE Ia; GSD1A

Glycogen storage disease type I (GSDI) is characterized by accumulation of glycogen and fat in the liver and kidneys, resulting in hepatomegaly and renomegaly. The two subtypes (GSDIa and GSDIb) are clinically indistinguishable. Some untreated neonates present with severe hypoglycemia; more commonly, however, untreated infants present at age three to four months with hepatomegaly, lactic acidosis, hyperuricemia, hyperlipidemia, hypertriglyceridemia, and/or hypoglycemic seizures. Affected children typically have doll-like faces with fat cheeks, relatively thin extremities, short stature, and protuberant abdomen. Xanthoma and diarrhea may be present. Impaired platelet function can lead to a bleeding tendency with frequent epistaxis. Normal growth and puberty is expected in treated children. Most affected individuals live into adulthood.

Your genetic map

Gene	SNP	Genotype
G6PC	rs104894566	TT
G6PC	rs80356484	GG
G6PC	rs1801176	GG
G6PC	rs80356483	GG
G6PC	rs104894563	CC
G6PC	rs80356488	
G6PC	rs1801175	CC
G6PC	rs80356479	
G6PC	rs80356487	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/232200

Carrier Status

GLYCOGEN STORAGE DISEASE II; GSD2

Glycogen storage disease due to acid maltase deficiency (AMD) is an autosomal recessive trait leading to metabolic myopathy that affects cardiac and respiratory muscles in addition to skeletal muscle and other tissues. AMD represents a wide spectrum of clinical presentations caused by an accumulation of glycogen in lysosomes: Glycogen storage disease due to acid maltase deficiency, infantile onset, non-classic infantile onset and adult onset. Early onset forms are more severe and often fatal.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/232300

Your genetic map

Gene	SNP	Genotype
GAA	rs28937909	GG
GAA	rs121907938	CC
GAA	rs386834236	TT
GAA	rs121907937	GG
GAA	rs28940868	CC
GAA	rs767882689	
GAA	rs140826989	GG
GAA	rs1800312	GG
GAA	rs386834235	
GAA	rs398123169	GG
GAA	rs369532274	CC
GAA	rs398123173	
GAA	rs398123174	TT
GAA	rs1800312	GG

Carrier Status

HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2; FHL2

Familial hemophagocytic lymphohistiocytosis (FHL) is characterized by proliferation and infiltration of hyperactivated macrophages and T-lymphocytes manifesting as acute illness with prolonged fever, cytopenias, and hepatosplenomegaly. Onset is typically within the first months or years of life and, on occasion, in utero, although later childhood or adult onset is more common than previously suspected. Neurologic abnormalities may be present initially or may develop later; they may include increased intracranial pressure, irritability, neck stiffness, hypotonia, hypertonia, convulsions, cranial nerve palsies, ataxia, hemiplegia, quadriplegia, blindness, and coma. Rash and lymphadenopathy are less common. Other findings include liver dysfunction and bone marrow hemophagocytosis.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/603553

Your genetic map

Gene	SNP	Genotype
PRF1	rs28933973	GG
PRF1	rs751161742	TT

Carrier Status

HERMANSKY-PUDLAK SYNDROME 3; HPS3

Hermansky-Pudlak syndrome (HPS) is a multisystem disorder characterized by: tyrosinase-positive oculocutaneous albinism; a bleeding diathesis resulting from a platelet storage pool deficiency; and, in some cases, pulmonary fibrosis, granulomatous colitis, or immunodeficiency. The albinism is characterized by: hypopigmentation of the skin and hair; and ocular findings of reduced iris pigment with iris transillumination, reduced retinal pigment, foveal hypoplasia with significant reduction in visual acuity (usually in the range of 20/50 to 20/400), nystagmus, and increased crossing of the optic nerve fibers. Hair color ranges from white to brown; skin color ranges from white to olive and is usually a shade lighter than that of other family members. The bleeding diathesis can result in easy bruising, frequent epistaxis, gingival bleeding, postpartum hemorrhage, colonic bleeding, and prolonged bleeding with menses or after tooth extraction, circumcision, and other surgeries. Pulmonary fibrosis, a restrictive lung disease, typically causes symptoms in the early thirties and can progress to death within a decade.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/614072

Your genetic map

Gene	SNP	Genotype
HPS3	rs201227603	GG

Carrier Status

HISTIOCYTOSIS-LYMPHADENOPATHY PLUS SYNDROME

Rosaï-Dorfman disease is a rare benign non-Langerhans cell histiocytosis characterized by the development of large painless histiocytic masses in the lymph nodes, predominantly of the cervical region. Extranodal involvement can also be observed, such as in the skin, respiratory tract, bones, genitourinary system, soft tissues, oral cavity, and central nervous system.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/602782

Your genetic map

Gene	SNP	Genotype
SLC29A3	rs121912583	GG
SLC29A3	rs587780462	CC
SLC29A3	rs587780463	GG
SLC29A3	rs121912584	GG

Carrier Status

ECTODERMAL DYSPLASIA 1, HYPOHIDROTIC, X-LINKED; XHED

Hypohidrotic ectodermal dysplasia (HED) is characterized by hypotrichosis (sparseness of scalp and body hair), hypohidrosis (reduced ability to sweat), and hypodontia (congenital absence of teeth). The cardinal features of classic HED become obvious during childhood. The scalp hair is thin, lightly pigmented, and slow-growing. Sweating, although present, is greatly deficient, leading to episodes of hyperthermia until the affected individual or family acquires experience with environmental modifications to control temperature. Only a few abnormally formed teeth erupt, and at a later-than-average age. Physical growth and psychomotor development are otherwise within normal limits. Mild HED is characterized by mild manifestations of any or all the characteristic features.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/305100

Your genetic map

Gene	SNP	Genotype
EDA	rs727504814	TT
EDA	rs132630312	CC
EDA	rs132630314	GG

Carrier Status

JERVELL AND LANGE-NIELSEN SYNDROME 1; JLNS1

Jervell and Lange-Nielsen syndrome (JLNS) is an autosomal recessive variant of familial long QT syndrome (see this term) characterized by congenital profound bilateral sensorineural hearing loss, a long QT interval on electrocardiogram and ventricular tachyarrhythmias. The disease is very rare. Prevalence is unknown and varies depending on the population studied (1/200,000-1/1,000,000) but is more common in countries in which consanguineous marriage is frequent.

Your genetic map

Gene	SNP	Genotype
KCNE1	rs74315445	CC
KCNQ1	rs120074190	GG
KCNQ1	rs120074189	CC
KCNQ1	rs120074186	GG
KCNQ1	rs397508118	
KCNQ1	rs397508117	
KCNQ1	rs397508110	II
KCNQ1	rs397508131	GG
KCNQ1	rs397508134	II
KCNQ1	rs397508120	II
KCNQ1	rs397508127	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/220400

Carrier Status

JOUBERT SYNDROME 14; JBTS14

Autosomal recessive development disorder characterized by severe mental retardation, cerebellar vermis hypoplasia, hypotonia, abnormal breathing pattern in infancy, and dysmorphic facial features. Additional findings may include renal cysts, abnormal eye movements, and postaxial polydactyly.

Your genetic map

Gene	SNP	Genotype
TMEM237	rs387907131	GG

What does your genetics say?

●

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/614424

Carrier Status

JOUBERT SYNDROME 16; JBTS16

Autosomal recessive development disorder characterized by the sign of the molar tooth in cerebral images, oculomotor apraxia, variable coloboma and rare renal involvement.

Your genetic map

Gene	SNP	Genotype
TMEM138	rs387907133	CC
TMEM138	rs387907132	AA

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/614465

Carrier Status

JOUBERT SYNDROME 3; JBTS3

Not many cases are known, one of the three reviews in the literature describes that multiple abnormalities of the central nervous system, such as polymicrogyria, malformations of the corpus callosum, convulsions, and spasticity, often occurred.

Your genetic map

Gene	SNP	Genotype
AHI1	rs397514726	CC
AHI1	rs121434351	CC
AHI1	rs777668842	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/608629

Carrier Status

JOUBERT SYNDROME 5; JBTS5

It is characterized mainly by the neurological and neuroradiological features of Joubert syndrome associated with severe retinal and renal involvement, but noted that the clinical spectrum was broad, including incomplete phenotypes such as cerebelloretinal and cereorothorenal syndromes. The entire JBTS5 phenotype largely coincides with the Senior-Loken syndrome (SLSN, see 266900), which is characterized by retinitis pigmentosa plus juvenile nephronoptis.

Your genetic map

Gene	SNP	Genotype
CEP290	rs137852834	TT
CEP290	rs370119681	CC
CEP290	rs62640570	
CEP290	rs62635288	CC
CEP290	rs727503853	II
CEP290	rs766608755	
CEP290	rs137852832	CC
CEP290	rs281865192	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610188

Carrier Status

JOUBERT SYNDROME 7; JBTS7

Joubert syndrome is a clinical and genetically heterogeneous group of disorders characterized by cerebellar vermis hypoplasia with the characteristic neuroradiological sign of the molar tooth and accompanying neurological symptoms, including dysregulation of the respiratory pattern and developmental delay. Other variable features include retinal dystrophy and renal abnormalities.

Your genetic map

Gene	SNP	Genotype
RPGRIP1L	rs121918204	GG
RPGRIP1L	rs121918198	TT
RPGRIP1L	rs778149316	DD
RPGRIP1L	rs532768944	GG
RPGRIP1L	rs121918203	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/611560

Carrier Status

JOUBERT SYNDROME 8; JBTS8

It is characterized by congenital malformation of the brain stem and agenesis or hypoplasia of the cerebellar vermis that leads to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia and delay in the achievement of motor milestones.

Your genetic map

Gene	SNP	Genotype
ARL13B	rs121912607	GG
ARL13B	rs121912608	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/612291

Carrier Status

JOUBERT SYNDROME 9; JBTS9

Joubert syndrome is a clinical and genetically heterogeneous group of disorders characterized by cerebellar vermis hypoplasia with the characteristic neuroradiological sign of the molar tooth and accompanying neurological symptoms, including dysregulation of the respiratory pattern and developmental delay. Other variable features include retinal dystrophy and renal abnormalities.

Your genetic map

Gene	SNP	Genotype
CC2D2A	rs118204053	CC
CC2D2A	rs764719093	CC
CC2D2A	rs118204052	CC
CC2D2A	rs200407856	GG
CC2D2A	rs386833752	CC
CC2D2A	rs386833760	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/612285

Carrier Status

KABUKI SYNDROME 1; KABUK1

Kabuki syndrome (KS) is a multiple congenital anomaly syndrome characterized by typical facial features, skeletal anomalies, mild to moderate intellectual disability and postnatal growth deficiency. KS was initially described in Japan, but has now been observed in all ethnic groups. Prevalence estimation is approximately 1:32,000.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/147920

Your genetic map

Gene	SNP	Genotype
KMT2D	rs267607237	CC
KMT2D	rs587783704	II
KMT2D	rs587783703	II
KMT2D	rs587783700	TT
KMT2D	rs587783699	GG
KMT2D	rs587783698	GG
KMT2D	rs587783697	CC
KMT2D	rs587783696	CC
KMT2D	rs587783693	II
KMT2D	rs587783692	GG
KMT2D	rs587783691	II
KMT2D	rs587783690	GG
KMT2D	rs587783688	GG
KMT2D	rs587783687	II
KMT2D	rs587783686	II
KMT2D	rs587783685	GG
KMT2D	rs587783683	II
KMT2D	rs587783682	GG
KMT2D	rs587783681	GG
KMT2D	rs587783729	GG
KMT2D	rs587783727	GG
KMT2D	rs587783725	II
KMT2D	rs556669370	GG
KMT2D	rs587783719	II
KMT2D	rs587783715	II
KMT2D	rs587783713	II
KMT2D	rs587783712	GG
KMT2D	rs587783705	CC
KMT2D	rs587783689	II
KMT2D	rs587783714	CC
KMT2D	rs587783708	CC

Carrier Status

LEIGH SYNDROME; LS

Leigh syndrome or subacute necrotizing encephalomyelopathy is a progressive neurological disease defined by specific neuropathological features associating brainstem and basal ganglia lesions. Its prevalence at birth has been estimated at approximately 1 in 36 000.

Your genetic map

Gene	SNP	Genotype
NDUFS8	rs764276946	AA

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/256000

Carrier Status

LEOPARD SYNDROME 1; LPRD1

Noonan syndrome with multiple lentigines (NSML), previously known as LEOPARD syndrome, is a rare multisystem genetic disorder characterized by lentigines, hypertrophic cardiomyopathy, short stature, pectus deformity, and dysmorphic facial features.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/151100

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918457	CC
PTPN11	rs121918468	GG
PTPN11	rs397507548	AA
PTPN11	rs121918469	GG
PTPN11	rs397507549	CC
PTPN11	rs397507542	GG

Carrier Status

LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM

A new leukoencephalopathy, the CACH syndrome (Childhood Ataxia with Central nervous system Hypomyelination) or VWM (Vanishing White Matter) was identified on clinical and MRI criteria.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/603896

Your genetic map

Gene	SNP	Genotype
EIF2B5	rs113994048	AA
EIF2B5	rs113994053	CC
EIF2B2	rs113994012	GG
EIF2B5	rs113994049	GG

Carrier Status

LISSENCEPHALY 1; LIS1

LIS1-associated lissencephaly includes Miller-Dieker syndrome (MDS), isolated lissencephaly sequence (ILS), and (rarely) subcortical band heterotopia (SBH). Lissencephaly and SBH are cortical malformations caused by deficient neuronal migration during embryogenesis. Lissencephaly refers to a "smooth brain" with absent gyri (agyria) or abnormally wide gyri (pachygyria). SBH refers to a band of heterotopic gray matter located just beneath the cortex and separated from it by a thin zone of normal white matter. MDS is characterized by lissencephaly, typical facial features, and severe neurologic abnormalities. ILS is characterized by lissencephaly and its direct sequelae: developmental delay, intellectual disability, and seizures.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/607432

Your genetic map

Gene	SNP	Genotype
PAFAH1B	rs121434487	GG
PAFAH1B	rs113994203	GG
PAFAH1B	rs113994201	
PAFAH1B	rs587784265	GG
PAFAH1B	rs587784260	CC
PAFAH1B	rs587784262	CC
PAFAH1B	rs587784272	TT
PAFAH1B	rs587784253	II
PAFAH1B	rs587784254	TT
PAFAH1B	rs587784257	GG
PAFAH1B	rs587784261	TT
PAFAH1B	rs587784263	AA
PAFAH1B	rs587784269	CC
PAFAH1B	rs587784271	II
PAFAH1B	rs587784273	CC
PAFAH1B	rs587784274	II
PAFAH1B	rs587784275	II
PAFAH1B	rs587784276	GG
PAFAH1B	rs587784277	II
PAFAH1B	rs587784278	CC
PAFAH1B	rs587784281	GG
PAFAH1B	rs587784280	GG
PAFAH1B	rs587784282	CC
PAFAH1B	rs587784284	DD
PAFAH1B	rs587784286	CC
PAFAH1B	rs587784287	AA
PAFAH1B	rs587784288	TT
PAFAH1B	rs587784289	GG
PAFAH1B	rs587784291	GG
PAFAH1B	rs587784290	GG
PAFAH1B	rs587784292	II

Carrier Status

LOEYS-DIETZ SYNDROME 2; LDS2

Loeys-Dietz syndrome (LDS) is characterized by vascular findings (cerebral, thoracic, and abdominal arterial aneurysms and/or dissections) and skeletal manifestations (pectus excavatum or pectus carinatum, scoliosis, joint laxity, arachnodactyly, talipes equinovarus). Approximately 75% of affected individuals have LDS type I with craniofacial manifestations (widely spaced eyes, bifid uvula/cleft palate, craniosynostosis); approximately 25% have LDS type II with systemic manifestations of LDSI but minimal or absent craniofacial features. LDSI and LDSII form a clinical continuum. The natural history of LDS is characterized by aggressive arterial aneurysms (mean age at death 26.1 years) and a high incidence of pregnancy-related complications, including death and uterine rupture

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610168

Your genetic map

Gene	SNP	Genotype
TGFBR2	rs104893809	CC
TGFBR2	rs104893810	CC
TGFBR2	rs104893816	GG
TGFBR2	rs104893811	CC
TGFBR2	rs104893819	CC

Carrier Status

LONG QT SYNDROME 1; LQT1

Congenital long QT syndrome (LQTS) is a hereditary cardiac disease characterized by a prolongation of the QT interval at basal ECG and by a high risk of life-threatening arrhythmias. Disease prevalence is estimated at close to 1 in 2,500 live births.

Your genetic map

Gene	SNP	Genotype
KCNQ1	rs199473457	CC
KCNQ1	rs120074181	GG
KCNQ1	rs120074182	CC
KCNQ1	rs120074180	CC
KCNQ1	rs120074184	GG
KCNQ1	rs120074179	GG
KCNQ1	rs12720459	CC
KCNQ1	rs120074178	GG
KCNQ1	rs120074193	GG
KCNQ1	rs120074194	GG
KCNQ1	rs179489	GG
KCNQ1	rs12720459	CC
KCNQ1	rs1800171	GG
KCNQ1	rs397508105	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/192500

Carrier Status

MAPLE SYRUP URINE DISEASE; MSUD

Maple syrup urine disease (MSUD) is a rare inherited disorder of branched-chain amino acid metabolism classically characterized by poor feeding, lethargy, vomiting and a maple syrup odor in the cerumen (and later in urine) noted soon after birth, followed by progressive encephalopathy and central respiratory failure if untreated. The estimated prevalence is around 1/150,000 live births, from published and unpublished newborn screening data.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/248600

Your genetic map

Gene	SNP	Genotype
BCKDHA	rs137852871	GG
BCKDHA	rs137852875	CC
DBT	rs121964999	AA
BCKDHB	rs386834234	GG
BCKDHB	rs386834233	GG
BCKDHA	rs182923857	CC
BCKDHA	rs398123490	GG
BCKDHA	rs398123491	CC
BCKDHA	rs398123492	II
BCKDHA	rs398123494	II
BCKDHA	rs398123496	GG
BCKDHA	rs398123497	CC
BCKDHA	rs398123499	CC
BCKDHA	rs398123503	CC
BCKDHA	rs375785084	CC
BCKDHA	rs373713279	CC
BCKDHA	rs398123508	GG
BCKDHA	rs398123509	AA
BCKDHA	rs398123510	II
BCKDHA	rs398123512	II
BCKDHA	rs398123513	CC
BCKDHA	rs398123515	GG
DBT	rs398123660	GG
DBT	rs398123663	AA
DBT	rs398123665	CC
DBT	rs398123667	II
DBT	rs398123668	II
DBT	rs398123669	CC
DBT	rs398123674	TT
DBT	rs398123675	GG
BCKDHB	rs398124561	CC

Carrier Status

MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 2; MODY2

MODY is a form of NIDDM (125853) characterized by monogenic autosomal dominant transmission and early age of onset. For a general phenotypic description and a discussion of genetic heterogeneity of MODY, see 606391. In a review of the various forms of MODY, Fajans et al. (2001) stated that glucokinase-related MODY2 is a common form of the disorder, especially in children with mild hyperglycemia and in women with gestational diabetes and a family history of diabetes. It has been described in persons of all racial and ethnic groups. More than 130 MODY-associated mutations have been found in the glucokinase gene.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/125851

Your genetic map

Gene	SNP	Genotype
GCK	rs193922331	AA
GCK	rs193922253	DD
GCK	rs193922255	
GCK	rs193922259	TT
GCK	rs193922260	TT
GCK	rs193922262	CC
GCK	rs193922263	GG
GCK	rs193922265	GG
GCK	rs193922267	CC
GCK	rs193922268	AA
GCK	rs193921338	GG
GCK	rs193921340	AA
GCK	rs193921340	AA
GCK	rs193922269	CC
GCK	rs193922271	GG
GCK	rs193922272	TT
GCK	rs193922273	AA
GCK	rs193922277	AA
GCK	rs193922278	AA
GCK	rs193922279	CC
GCK	rs193922281	GG
GCK	rs193922285	CC
GCK	rs193922286	GG
GCK	rs193922287	GG
GCK	rs193922290	TT
GCK	rs193922295	II
GCK	rs193922300	GG
GCK	rs193922301	TT
GCK	rs193922302	CC
GCK	rs193922303	CC
GCK	rs193922304	GG

Carrier Status

MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3; MODY3

A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/600496

Your genetic map

Gene	**SNP**	**Genotype**
HNF1A	rs193922577	TT
HNF1A	rs193922578	II
HNF1A	rs193922580	CC
HNF1A	rs193922582	DD
HNF1A	rs193922587	CC
HNF1A	rs193922588	II
HNF1A	rs193922589	AA
HNF1A	rs193922593	CC
HNF1A	rs193922594	DD
HNF1A	rs193922597	CC
HNF1A	rs150513055	CC
HNF1A	rs386134267	II
HNF1A	rs193922598	CC
HNF1A	rs193922599	II
HNF1A	rs193922600	CC
HNF1A	rs193922602	GG
HNF1A	rs193922603	GG
HNF1A	rs193922604	GG
HNF1A	rs193922605	TT

Carrier Status

MECKEL SYNDROME, TYPE 3; MKS3

Meckel syndrome is an autosomal recessive pre- or perinatal lethal malformation syndrome characterized by renal cystic dysplasia and variably associated features including developmental anomalies of the central nervous system (typically occipital encephalocele), hepatic ductal dysplasia and cysts, and postaxial polydactyly (summary by Smith et al., 2006).

For a more complete phenotypic description and information on genetic heterogeneity of Meckel syndrome, see MKS1

Your genetic map

Gene	SNP	Genotype
TMEM67	rs386834182	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/607361

Carrier Status

MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA; MICPCH

CASK-related disorders include a spectrum of phenotypes in both females and males. The two main types of clinical presentation are: Microcephaly with pontine and cerebellar hypoplasia (MICPCH), generally associated with pathogenic loss-of-function variants in CASK; and X-linked intellectual disability (XLID) with or without nystagmus, generally associated with hypomorphic CASK pathogenic variants. MICPCH is typically seen in females with moderate to severe intellectual disability, progressive microcephaly with or without ophthalmologic anomalies, and sensorineural hearing loss. To date a total of 53 females with MICPCH have been reported, the eldest of whom is 21 years old. Most are able to sit independently; 20%-25% attain the ability to walk; language is nearly absent in most.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300749

Your genetic map

Gene	SNP	Genotype
CASK	rs387906705	GG
CASK	rs587783361	GG
CASK	rs587783362	II
CASK	rs587783364	GG
CASK	rs587783366	TT
CASK	rs587783368	CC
CASK	rs587783371	GG
CASK	rs749742837	GG
CASK	rs587783360	GG
CASK	rs587783369	CC

Carrier Status

METACHROMATIC LEUKODYSTROPHY; MLD

Metachromatic leukodystrophy (MLD) is a rare lysosomal storage disorder characterized byintralysosomal accumulation of sulfatides in various tissues, leading to progressive deterioration of motor and neurocognitive function.

Your genetic map

Gene	SNP	Genotype
ARSA	rs28940893	GG
ARSA	rs74315467	GG
ARSA	rs74315470	GG
ARSA	rs398123414	II
ARSA	rs398123415	II
ARSA	rs398123416	II
ARSA	rs398123418	GG
ARSA	rs398123419	CC
ARSA	rs80338820	CC
ARSA	rs74315457	AA
ARSA	rs80338815	CC
ARSA	rs80338820	CC
ARSA	rs74315456	GG
ARSA	rs74315483	CC
ARSA	rs74315458	CC
ARSA	rs74315471	CC
ARSA	rs74315472	GG
ARSA	rs74315476	GG
ARSA	rs80338819	CC
ARSA	rs199476391	CC
ARSA	rs199476366	CC
ARSA	rs199476349	CC
ARSA	rs199476389	AA
ARSA	rs398123411	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/250100

Carrier Status

METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE

Methylmalonic acidemia with homocystinuria is an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures. Annual incidence in the USA, based on the California newborn screening program, has been estimated at 1/67,000 (for the cblC form). cblC is the most frequent type (over 550 cases)

Your genetic map

Gene	SNP	Genotype
MMACHC	rs121918241	CC
MMACHC	rs121918242	CC
MMACHC	rs370596113	CC
MMACHC	rs398124293	II
MMACHC	rs398124295	GG
MMACHC	rs398124296	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/277400

Carrier Status

METHYLMALONIC ACIDURIA, cblA TYPE

Vitamin B12-responsive methylmalonic acidemia (MA) is an inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which responds to vitamin B12. To date, over 120 patients with cblA have been reported. Prevalence of 1/48,000 -1/61,000 have been reported for MA of all causes in North America, and 1/26,000 in China.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/251100

Your genetic map

Gene	SNP	Genotype
MMAA	rs104893851	CC
MMAA	rs571038432	CC

Carrier Status

METHYLMALONIC ACIDURIA, cblB TYPE

Vitamin B12-responsive methylmalonic acidemia (MA) is an inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which responds to vitamin B12. To date, over 66 patients have been reported. Prevalence of 1/48,000-1/61,000 have been reported for MA of all causes in North America, and 1/26,000 in China.

Your genetic map

Gene	SNP	Genotype
MMAB	rs28941784	GG
MMAB	rs398124434	GG
MMAB	rs369296618	GG
MMAB	rs756414548	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/251110

Carrier Status

MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 1; MC3DN1

A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance.

Your genetic map

Gene	SNP	Genotype
BCS1L	rs121908576	CC
BCS1L	rs121908578	CC
BCS1L	rs144885874	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/124000

Carrier Status

MUCOPOLYSACCHARIDOSIS TYPE VI; MPS6

Mucopolysaccharidosis type 6 (MPS 6) is a lysosomal storage disease with progressive multisystem involvement, associated with a deficiency of arylsulfatase B (ASB) leading to the accumulation of dermatan sulfate. Birth prevalence is between 1 in 43,261 and 1 in 1,505,160 live births.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/253200

Your genetic map

Gene	SNP	Genotype
ARSB	rs201101343	TT
ARSB	rs118203941	CC
ARSB	rs118203942	CC
ARSB	rs118203943	TT
ARSB	rs118203944	TT

Carrier Status

MUCOPOLYSACCHARIDOSIS, TYPE VII; MPS7

Mucopolysaccharidosis type VII (MPS VII) is a very rare lysosomal storage disease belonging to the group of mucopolysaccharidoses. Less than 40 patients with neonatal to moderate presentation have been reported since the initial description of the disease by Sly in 1973. However, the frequency of the disease may be underestimated as the most frequent presentation is the antenatal form, which remains underdiagnosed.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/253220

Your genetic map

Gene	SNP	Genotype
GUSB	rs121918173	GG
GUSB	rs121918185	GG
GUSB	rs121918181	GG
GUSB	rs398123234	CC
GUSB	rs398123238	CC

Carrier Status

MUCOPOLYSACCHARIDOSIS, TYPE IIIA; MPS3A

Mucopolysaccharidosis type III (MPS III) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses and characterised by severe and rapid intellectual deterioration. The disorder is underdiagnosed (due to the generally very mild dysmorphism); it is the most frequent MPS in the Netherlands and Australia with respective prevalences of 1/53 0000 and 1/67 000. The frequency of the different subtypes varies between countries: subtype A is more frequent in England, the Netherlands and Australia

Your genetic map

Gene	SNP	Genotype
SGSH	rs104894636	GG
SGSH	rs104894641	CC
SGSH	rs104894637	GG
SGSH	rs104894640	CC
SGSH	rs778700037	DD
SGSH	rs104894635	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/252900

Carrier Status

MUCOPOLYSACCHARIDOSIS, TYPE IIIB; MPS3B

Mucopolysaccharidosis type III (MPS III) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses and characterised by severe and rapid intellectual deterioration. The disorder is underdiagnosed (due to the generally very mild dysmor. subtype B is more frequent in Greece and Portugal, whereas types IIIC and IIID are much less common.

Your genetic map

Gene	SNP	Genotype
NAGLU	rs104894598	GG
NAGLU	rs104894590	GG
NAGLU	rs104894598	GG
NAGLU	rs104894597	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/252920

Carrier Status

MUCOPOLYSACCHARIDOSIS, TYPE IVA; MPS4A

Mucopolysaccharidosis type IV (MPS IV) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses, and characterised by spondylo-epiphyso-metaphyseal dysplasia. It exists in two forms, A and B. Prevalence is approximately 1/250 000 for type IVA but incidence varies widely between countries. MPS IVB is even rarer.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/253000

Your genetic map

Gene	SNP	Genotype
GALNS	rs118204438	TT
GALNS	rs746756997	AA
GALNS	rs118204437	GG
GALNS	rs398123429	TT
GALNS	rs398123430	GG
GALNS	rs372893383	CC
GALNS	rs398123438	CC
GALNS	rs398123440	GG

Carrier Status

MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES), TYPE A, 1;

Congenital muscular dystrophy (CMD) is a clinically and genetically heterogeneous group of inherited muscle disorders. Muscle weakness typically presents from birth to early infancy. Affected infants typically appear "floppy" with low muscle tone and poor spontaneous movements. Affected children may present with delay or arrest of gross motor development together with joint and/or spinal rigidity. Muscle weakness may improve, worsen, or stabilize in the short term; however, with time progressive weakness and joint contractures, spinal deformities, and respiratory compromise may affect quality of life and life span.

Your genetic map

Gene	SNP	Genotype
POMT1	rs119462982	GG
POMT1	rs149682171	CC
POMT1	rs745738628	GG
POMT1	rs772370177	GG
POMT1	rs398124245	
POMT1	rs200056620	CC
POMT1	rs398124244	AA
POMT1	rs398124247	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/236670

Carrier Status

MYOPATHY, MYOFIBRILLAR, 1; MFM1

Myofibrillar myopathy is characterized by slowly progressive weakness that can involve both proximal and distal muscles. Distal muscle weakness is present in about 80% of individuals and is more pronounced than proximal weakness in about 25%. A minority of individuals experience sensory symptoms, muscle stiffness, aching, or cramps. Peripheral neuropathy is present in about 20% of affected individuals. Overt cardiomyopathy is present in 15%-30%.

Your genetic map

Gene	SNP	Genotype
DES	rs727504448	II
DES	rs397516698	GG
DES	rs121913003	CC
DES	rs121913005	CC
DES	rs62635763	CC
DES	rs267607482	AA
DES	rs267607499	AA
DES	rs267607495	CC
DES	rs62636495	CC
DES	rs150974575	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

Carrier Status

MYOPATHY, CENTRONUCLEAR, 1; CNM1

X-linked myotubular myopathy (XLMTM) is an inherited neuromuscular disorder defined by numerous centrally placed nuclei on muscle biopsy and clinical features of a congenital myopathy. The incidence of XLMTM is estimated at 1/50,000 male births.

Your genetic map

Gene	SNP	Genotype
DNM2	rs121909089	GG
DNM2	rs121909090	CC
DNM2	rs121909092	GG
DNM2	rs121909091	CC

What does your genetics say?

●

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/160150

Carrier Status

MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX

Autosomal dominant centronuclear myopathy is a congenital myopathy characterized by slowly progressive muscular weakness and wasting (Bitoun et al., 2005). The disorder involves mainly limb girdle, trunk, and neck muscles but may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life, and some affected individuals become wheelchair-bound in their fifties. Ptosis and limitation of eye movements occur frequently.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/310400

Your genetic map

Gene	SNP	Genotype
DNM2	rs121909095	CC
MTM1	rs132630302	AA
MTM1	rs587783791	
MTM1	rs132630305	CC
MTM1	rs132630306	CC
MTM1	rs587783817	TT
MTM1	rs587783823	GG
MTM1	rs587783843	GG
MTM1	rs587783844	AA
MTM1	rs587783846	GG
MTM1	rs587783857	CC
MTM1	rs587783753	CC
MTM1	rs587783788	
MTM1	rs587783796	GG
MTM1	rs587783796	GG
MTM1	rs587783803	II
MTM1	rs587783804	II
MTM1	rs587783809	CC
MTM1	rs587783814	CC
MTM1	rs587783813	AA
MTM1	rs587783812	GG
MTM1	rs587783815	II
MTM1	rs587783816	TT
MTM1	rs587783820	AA
MTM1	rs587783822	II
MTM1	rs587783824	II
MTM1	rs587783825	CC
MTM1	rs587783826	II
MTM1	rs587783828	GG
MTM1	rs587783830	GG
MTM1	rs587783831	AA

Carrier Status

NEMALINE MYOPATHY 2; NEM2

Nemaline myopathy (referred to in this entry as NM) is characterized by weakness, hypotonia, and depressed or absent deep tendon reflexes. Muscle weakness is usually most severe in the face, the neck flexors, and the proximal limb muscles. The clinical classification defines six forms of NM, which are classified by onset and severity of motor and respiratory involvement: Severe congenital (neonatal) (16% of all individuals with NM). Amish NM. Intermediate congenital (20%). Typical congenital (46%). Childhood-onset (13%). Adult-onset (late-onset) (4%). Considerable overlap occurs among the forms. There are significant differences in survival between individuals classified as having severe, intermediate, and typical congenital NM. Severe neonatal respiratory disease and the presence of arthrogryposis multiplex congenita are associated with death in the first year of life. Independent ambulation before age 18 months is predictive of survival. Most children with typical congenital NM are eventually able to walk. [from GTR]

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/256030

Your genetic map

Gene	SNP	Genotype
NEB	rs398124167	CC

Carrier Status

CYSTINOSIS, NEPHROPATHIC; CTNS

Cystinosis is a metabolic disease characterized by an accumulation of cystine inside the lysosomes, causing damage in different organs and tissues, particularly in the kidneys and eyes. The incidence of cystinosis is estimated at around 1/100,000- 1/200,000 live births.

Your genetic map

Gene	SNP	Genotype
CTNS	rs113994205	GG
CTNS	rs121908127	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/219800

Carrier Status

NIEMANN-PICK DISEASE, TYPE C1; NPC1

Niemann-Pick disease type C (NP-C) is a lysosomal lipid storage disease characterized by variable clinical signs, depending on the age of onset, such as prolonged unexplained neonatal jaundice or cholestasis, isolated unexplained splenomegaly, and progressive, often severe neurological symptoms such as cognitive decline, cerebellar ataxia, vertical supranuclear gaze palsy (VSPG), dysarthria, dysphagia, dystonia, seizures, gelastic cataplexy, and psychiatric disorders.

What does your genetics say?

●

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/257220

Your genetic map

Gene	SNP	Genotype
NPC1	rs80358257	GG
NPC1	rs80358252	CC
NPC1	rs483352886	CC
NPC1	rs369368181	GG
NPC1	rs372030650	TT
NPC1	rs80358254	CC
NPC1	rs80358259	AA
NPC1	rs80358254	CC
NPC1	rs120074135	CC
NPC1	rs28942107	GG
NPC1	rs28942108	GG
NPC1	rs80358254	CC
NPC1	rs398123284	DD
NPC1	rs543206298	GG

Carrier Status

NIEMANN-PICK DISEASE, TYPE A

Niemann-Pick disease type A is a very severe subtype of Niemann-Pick disease, an autosomal recessive lysosomal disease, and is characterized clinically by onset in infancy or early childhood with failure to thrive, hepatosplenomegaly, and rapidly progressive neurodegenerative disorders.

Your genetic map

Gene	SNP	Genotype
SMPD1	rs120074122	GG
SMPD1	rs727504166	TT
SMPD1	rs120074128	CC
SMPD1	rs182812968	CC
SMPD1	rs398123474	GG
SMPD1	rs398123479	GG
SMPD1	rs281860677	DD
SMPD1	rs727504165	II
SMPD1	rs120074126	CC
SMPD1	rs120074117	GG
SMPD1	rs120074124	TT
SMPD1	rs387906289	

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/257200

Carrier Status

NIEMANN-PICK DISEASE, TYPE B

Niemann-Pick disease type B is a mild subtype of Niemann-Pick disease, an autosomal recessive lysosomal disease, and is characterized clinically by onset in childhood with hepatosplenomegaly, growth retardation, and lung disorders such as infections and dyspnea

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/607616

Your genetic map

Gene	SNP	Genotype
SMPD1	rs769904764	CC
SMPD1	rs398123475	TT
SMPD1	rs398123478	CC
SMPD1	rs120074117	GG
SMPD1	rs727504167	
SMPD1	rs120074118	

Carrier Status

NOONAN SYNDROME 1; NS1

Noonan Syndrome (NS) is characterised by short stature, typical facial dysmorphism and congenital heart defects. The incidence of NS is estimated to be between 1:1000 and 1:2500 live births.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/163950

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918463	TT
PTPN11	rs397507509	GG
PTPN11	rs397507529	AA
NRAS	rs267606921	GG
BRAF	rs387906660	GG
PTPN11	rs121918454	CC
PTPN11	rs121918453	GG
PTPN11	rs28933386	AA
PTPN11	rs121918455	AA
PTPN11	rs121918460	TT
PTPN11	rs121918461	AA
PTPN11	rs121918459	AA
PTPN11	rs121918462	CC
PTPN11	rs121918466	AA
PTPN11	rs397507509	GG
PTPN11	rs397507520	GG
NRAS	rs267606920	CC
BRAF	rs606231228	CC

Carrier Status

NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MYELOMONOCYTIC LEUKEMIA; NSLL

A syndrome characterized by a phenotype reminiscent of Noonan syndrome. Clinical features are highly variable, including facial dysmorphism, short neck, developmental delay, hyperextensible joints and thorax abnormalities with widely spaced nipples. The facial features consist of triangular face with hypertelorism, large low-set ears, ptosis, and flat nasal bridge. Some patients manifest cardiac defects. Some have an increased risk for certain malignancies, particularly juvenile myelomonocytic leukemia.

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918456	AA
CBL	rs397517076	GG
CBL	rs727504504	CC
CBL	rs267606704	AA
CBL	rs267606708	GG
CBL	rs397517076	GG
CBL	rs397517077	II

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613563

Carrier Status

NOONAN SYNDROME 4; NS4

Noonan Syndrome (NS) is characterised by short stature, typical facial dysmorphism and congenital heart defects. The incidence of NS is estimated to be between 1:1000 and 1:2500 live births. The main facial features of NS are hypertelorism with down-slanting palpebral fissures, ptosis and low-set posteriorly rotated ears with a thickened helix. The cardiovascular defects most commonly associated with this condition are pulmonary stenosis and hypertrophic cardiomyopathy. Other associated features are webbed neck, chest deformity, mild intellectual deficit, cryptorchidism, poor feeding in infancy, bleeding tendency and lymphatic dysplasia.

Your genetic map

Gene	SNP	Genotype
SOS1	rs137852813	AA
SOS1	rs267607079	CC
SOS1	rs267607080	AA
SOS1	rs137852813	AA
SOS1	rs397517154	CC
SOS1	rs137852812	GG
SOS1	rs137852814	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610733

Carrier Status

Obesity due to melanocortin 4 receptor deficiency

Melanocortin 4 receptor (MC4R) deficiency is the commonest form of monogenic obesity identified so far. MC4R deficiency is characterised by severe obesity, an increase in lean body mass and bone mineral density, increased linear growth in early childhood, hyperphagia beginning in the first year of life and severe hyperinsulinaemia, in the presence of preserved reproductive function. The prevalence in the general population is probably around 1 in 2000. The prevalence of MC4R mutations has been estimated at between 0.5 and 1% in obese adults (body mass index >30) with higher values among populations with severe childhood-onset obesity and variability between ethnic groups.

Your genetic map

Gene	SNP	Genotype
LEPR	rs193922650	CC
MC4R	rs193922685	AA
MC4R	rs79783591	AA
MC4R	rs193922687	DD
MC4R	rs52804924	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601665

This report is not valid for clinical or diagnostic use.

Carrier Status

ALBINISM, OCULOCUTANEOUS, TYPE IB; OCA1B

Type 1 oculocutaneous albinism (OCA1) describes a group of tyrosine related OCAs that includes OCA1A, OCA1B, type 1 minimal pigment oculocutaneous albinism (OCA1-MP) and type 1 temperature sensitive oculocutaneous albinism (OCA1-TS). The worldwide prevalence of OCA1 is estimated at 1/40,000.

Your genetic map

Gene	SNP	Genotype
TYR	rs28940876	CC
TYR	rs104894314	GG
TYR	rs121908011	GG
TYR	rs61753180	GG
TYR	rs28940881	AA
TYR	rs104894313	CC
TYR	rs61754388	CC
TYR	rs61754381	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/606952

Carrier Status

OSTEOGENESIS IMPERFECTA, TYPE III; OI3

Osteogenesis imperfecta type III is a severe type of osteogenesis imperfecta a genetic disorder characterized by increased bone fragility, low bone mass and susceptibility to bone fractures. The main signs of type III include very short stature, a triangular face, severe scoliosis, grayish sclera, and dentinogenesis imperfecta. The overall prevalence of OI is estimated at between 1/10,000 and 1/20,000 but the prevalence of type III is unknown.

Your genetic map

Gene	**SNP**	**Genotype**
COL1A2	rs72658151	GG
COL1A2	rs72658161	GG
COL1A2	rs768171831	CC
COL1A1	rs72645357	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/259420

Carrier Status

DIABETES MELLITUS, PERMANENT NEONATAL; PNDM

Permanent neonatal diabetes mellitus (PNDM) is a monogenic form of neonatal diabetes characterized by persistent hyperglycemia within the first 12 months of life in general, requiring continuous insulin treatment. The incidence of NDM is estimated to be 1/95,000 to 1/150,000 live births. The condition has been reported in all ethnic groups and affects male and female infants equally.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/606176

Your genetic map

Gene	SNP	Genotype
KCNJ11	rs80356616	CC
KCNJ11	rs80356624	CC
KCNJ11	rs80356625	GG
KCNJ11	rs193929355	CC
KCNJ11	rs193929356	TT
INS	rs80356669	GG

Carrier Status

PITT-HOPKINS SYNDROME; PTHS

Pitt-Hopkins syndrome (PHS) is characterized by the association of intellectual deficit, characteristic facial dysmorphism and problems of abnormal and irregular breathing. About 50 cases have been reported worldwide. Males and females are equally affected.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/610954

Your genetic map

Gene	SNP	Genotype
TCF4	rs398123561	
TCF4	rs121909123	CC
TCF4	rs587784462	CC
TCF4	rs587784460	CC
TCF4	rs587784459	CC
TCF4	rs587784458	CC
TCF4	rs587784469	CC
TCF4	rs587784468	II
TCF4	rs587784466	CC
TCF4	rs587784463	II
TCF4	rs727504175	GG
TCF4	rs727504174	II
TCF4	rs121909121	CC
TCF4	rs121909122	GG
TCF4	rs398123560	CC
TCF4	rs121909123	CC
TCF4	rs587784464	GG

Carrier Status

POLYMICROGYRIA, BILATERAL FRONTOPARIETAL; BFPP

Bilateral frontoparietal polymicrogyria (BFPP) is a sub-type of polymicrogyria, a cerebral cortical malformation characterized by excessive cortical folding and abnormal cortical layering, that involves the frontoparietal region of the brain and that presents with hypotonia, developmental delay, moderate to severe intellectual disability, pyramidal signs, epileptic seizures, non progressive cerebellar ataxia, dysconjugate gaze and/or strabismus.

Your genetic map

Gene	SNP	Genotype
ADGRG1	rs587783658	CC
ADGRG1	rs146278035	CC
ADGRG1	rs587783660	GG
ADGRG1	rs532188689	GG
ADGRG1	rs587783652	CC
ADGRG1	rs587783653	TT
ADGRG1	rs587783655	TT
ADGRG1	rs587783656	GG
ADGRG1	rs587783657	GG
ADGRG1	rs121908464	CC
ADGRG1	rs587783654	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/606854

Carrier Status

MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE; MCPH5

Autosomal recessive primary microcephaly (MCPH) is a rare genetically heterogeneous disorder of neurogenic brain development characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment. Exact prevalence of non-syndromic microcephaly is not known. MCPH is more common in Asian and Middle Eastern populations than in Caucasians, in whom an annual incidence of 1/1,000,000 is reported. It is more common in specific populations, e.g. northern Pakistanis. Consanguinity appears to play a role in incidence.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/608716

Your genetic map

Gene	SNP	Genotype
ASPM	rs199422146	
ASPM	rs137852997	AA
ASPM	rs140602858	GG
ASPM	rs199422165	GG
ASPM	rs199422134	GG
ASPM	rs199422189	GG
ASPM	rs587783220	II
ASPM	rs587783221	
ASPM	rs587783227	GG
ASPM	rs587783228	II
ASPM	rs587783230	AA
ASPM	rs587783238	CC
ASPM	rs587783239	II
ASPM	rs587783247	AA
ASPM	rs587783248	GG
ASPM	rs587783259	II
ASPM	rs587783268	GG
ASPM	rs587783269	II
ASPM	rs587783272	GG
ASPM	rs587783275	GG
ASPM	rs587783277	II
ASPM	rs587783278	II
ASPM	rs587783282	GG
ASPM	rs587783285	CC
ASPM	rs587783287	GG
ASPM	rs587783288	AA
ASPM	rs587783289	II
ASPM	rs759632528	DD
ASPM	rs199422147	II
ASPM	rs199422161	CC
ASPM	rs199422173	

Carrier Status

RETINITIS PIGMENTOSA; RP

Retinitis pigmentosa (RP) is an inherited retinal dystrophy leading to progressive loss of the photoreceptors and retinal pigment epithelium and resulting in blindness usually after several decades. Prevalence of RP is reported to be 1/3,000 to 1/5,000. No ethnic specificities have been reported although founder effects are possible.

Your genetic map

Gene	SNP	Genotype
USH2A	rs80338903	II
IFT140	rs779007169	CC
PDE6B	rs727504075	GG
USH2A	rs397518039	TT

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/268000

Carrier Status

RUBINSTEIN-TAYBI SYNDROME 1; RSTS1

Rubinstein-Taybi syndrome is a rare malformation syndrome characterized by congenital anomalies (microcephaly, specific facial characteristics, broad thumbs and halluces and postnatal growth retardation), short stature, intellectual disability and behavioural characteristics. Birth prevalence is estimated at around 1/ 100,000 to 125,000.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/180849

Your genetic map

Gene	SNP	Genotype
CREBBP	rs587783510	GG
CREBBP	rs587783508	II
CREBBP	rs587783507	II
CREBBP	rs587783505	GG
CREBBP	rs587783503	AA
CREBBP	rs587783500	II
CREBBP	rs587783499	II
CREBBP	rs587783497	TT
CREBBP	rs587783496	TT
CREBBP	rs147688139	AA
CREBBP	rs587783494	TT
CREBBP	rs587783493	GG
CREBBP	rs587783492	AA
CREBBP	rs587783491	CC
CREBBP	rs587783490	GG
CREBBP	rs587783489	GG
CREBBP	rs587783488	CC
CREBBP	rs587783486	TT
CREBBP	rs200782888	CC
CREBBP	rs587783482	CC
CREBBP	rs587783481	TT
CREBBP	rs587783480	CC
CREBBP	rs587783479	GG
CREBBP	rs587783475	GG
CREBBP	rs587783473	II
CREBBP	rs587783471	GG
CREBBP	rs587783470	II
CREBBP	rs587783469	II
CREBBP	rs587783467	II
CREBBP	rs587783465	II
CREBBP	rs587783464	GG

Carrier Status

SOTOS SYNDROME 1; SOTOS1

Sotos syndrome is a rare multisystemic genetic disorder characterized by a typical facial appearance, overgrowth of the body in early life with macrocephaly, and mild to severe intellectual disability.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/117550

Your genetic map

Gene	SNP	Genotype
NSD1	rs587784068	II
NSD1	rs587784071	GG
NSD1	rs587784073	II
NSD1	rs587784078	II
NSD1	rs587784079	II
NSD1	rs587784080	
NSD1	rs587784081	II
NSD1	rs587784084	CC
NSD1	rs587784085	II
NSD1	rs201327209	CC
NSD1	rs587784086	II
NSD1	rs587784088	CC
NSD1	rs587784089	II
NSD1	rs587784093	II
NSD1	rs587784094	II
NSD1	rs587784095	CC
NSD1	rs587784098	CC
NSD1	rs587784099	II
NSD1	rs587784100	II
NSD1	rs587784101	II
NSD1	rs587784103	II
NSD1	rs587784105	GG
NSD1	rs587784109	GG
NSD1	rs587784111	TT
NSD1	rs587784115	GG
NSD1	rs587784118	CC
NSD1	rs587784119	CC
NSD1	rs587784120	AA
NSD1	rs587784121	II
NSD1	rs587784122	CC
NSD1	rs587784125	II

Carrier Status

SUPRAVALVULAR AORTIC STENOSIS; SVAS

SupraValvar Aortic Stenosis (SVAS) is characterized by the narrowing of the aorta lumen (close to its origin) or other arteries (branch pulmonary arteries, coronary arteries). This narrowing of the aorta or pulmonary branches may impede blood flow, resulting in heart murmur and ventricular hypertrophy (in case of aorta involvement). The narrowing results from a thickening of the artery wall, which is not related to atherosclerosis. The incidence of SVAS is estimated at approximately 1 in 25 000 births and the mean prevalence in the general population at 1/7 500.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/185500

Your genetic map

Gene	SNP	Genotype
ELN	rs727503782	II
ELN	rs727503022	DD
ELN	rs727503023	II
ELN	rs727503024	II
ELN	rs727503026	II
ELN	rs727503027	AA
ELN	rs727503029	GG
ELN	rs727503031	II
ELN	rs727503033	TT
ELN	rs727503035	GG
ELN	rs727504581	II
ELN	rs730880355	DD
ELN	rs137854452	CC
ELN	rs397516433	CC
ELN	rs727503028	DD
ELN	rs727503030	GG

Carrier Status

TAY-SACHS DISEASE; TSD

GM2 gangliosidosis, variant B or Tay-Sachs disease is marked by accumulation of G2 gangliosides due to hexosaminidase A deficiency. The prevalence of the disease is 1 case per 320 000 live births.

Your genetic map

Gene	SNP	Genotype
HEXA	rs121907966	GG
HEXA	rs121907954	CC
HEXA	rs28942071	GG
HEXA	rs770932296	CC
HEXA	rs121907955	CC
HEXA	rs28941770	CC
HEXA	rs121907972	GG
HEXA	rs587779406	GG
HEXA	rs370266293	CC
HEXA	rs147324677	CC
HEXA	rs76173977	CC

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/272800

Carrier Status

TUBEROUS SCLEROSIS 1; TSC1

Tuberous sclerosis complex (TSC) is a neurocutaneous disorder characterized by multisystem hamartomas and associated with neuropsychiatric features. The prevalence is estimated to be 1/25,000-1/11,300 in Europe.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/191100

Your genetic map

Gene	SNP	Genotype
TSC1	rs118203682	GG
TSC1	rs118203478	
TSC1	rs118203434	GG
TSC1	rs118203506	II
TSC1	rs118203527	
TSC1	rs118203603	
TSC1	rs118203352	TT
TSC1	rs118203360	II
TSC1	rs118203423	CC
TSC1	rs118203479	
TSC1	rs397514842	CC
TSC1	rs397514867	GG
TSC1	rs397514875	II
TSC1	rs118203726	
TSC1	rs118203595	
TSC1	rs118203427	GG
TSC1	rs118203474	GG

Carrier Status

TUBEROUS SCLEROSIS 2; TSC2

Tuberous sclerosis complex (TSC) is a neurocutaneous disorder characterized by multisystem hamartomas and associated with neuropsychiatric features. The prevalence is estimated to be 1/25,000-1/11,300 in Europe. TSC is characterized by multisystem hamartomas, most commonly skin, brain, kidney, lung and heart, appearing at different ages. Skin involvement includes: hypomelanotic macules (ash leaf) present within the first years of life; angiofibromas that appear at age 3-4 years as erythematous and papulonodular lesions; ungual fibromas; cephalic and lumbar (shagreen patch) fibrous plaques; and "confetti" skin lesions appearing in childhood to early adolescence. Brain is involved in almost all cases of TSC, with the presence of different neuropathological lesions, such as cortico/subcortical tubers, radial migration lines, subependymal nodules, SEGA. SEGA can cause hydrocephalus (growth risk higher in the first 3 decades).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/613254

Your genetic map

Gene	SNP	Genotype
TSC2	rs45517182	GG
TSC2	rs397515297	GG
TSC2	rs45451497	CC
TSC2	rs45517412	CC
TSC2	rs45517395	GG
TSC2	rs137854250	
TSC2	rs45517096	AA
TSC2	rs45517229	AA
TSC2	rs45491698	GG
TSC2	rs137854368	II
TSC2	rs45517118	GG
TSC2	rs45488893	GG
TSC2	rs137854317	II
TSC2	rs45517386	
TSC2	rs45517337	CC
TSC2	rs137854155	CC
TSC2	rs45517174	AA
TSC2	rs137854298	TT
TSC2	rs45517213	GG
TSC2	rs45517246	AA
TSC2	rs45517252	GG
TSC2	rs137854261	
TSC2	rs45517395	GG
TSC2	rs45472701	CC
TSC2	rs137854249	II
TSC2	rs137854359	
TSC2	rs45479192	CC
TSC2	rs45517222	CC
TSC2	rs45517159	CC
TSC2	rs397515226	II
TSC2	rs45517258	CC

Carrier Status

ALBINISM, OCULOCUTANEOUS, TYPE IA; OCA1A

Type 1 oculocutaneous albinism (OCA1) describes a group of tyrosine related OCAs that includes OCA1A, OCA1B, type 1 minimal pigment oculocutaneous albinism (OCA1-MP) and type 1 temperature sensitive oculocutaneous albinism (OCA1-TS). The worldwide prevalence of OCA1 is estimated at 1/40,000.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/203100

Your genetic map

Gene	**SNP**	**Genotype**
TYR	rs758115945	GG
TYR	rs61754380	GG
TYR	rs151206295	CC
TYR	rs61753185	GG
TYR	rs28940880	GG
TYR	rs63159160	CC
TYR	rs61754375	GG
TYR	rs104894317	GG
TYR	rs62645917	CC
TYR	rs61754365	GG
TYR	rs61754371	CC

Carrier Status

TYROSINEMIA, TYPE I; TYRSN1

Tyrosinemia type 1 (HTI) is an inborn error of tyrosine catabolism caused by defective activity of fumarylacetoacetate hydrolase (FAH) and is characterized by progressive liver disease, renal tubular dysfunction, porphyria-like crises and a dramatic improvement in prognosis following treatment with nitisinone. Birth incidence is 1/100,000 in most areas but is more common is some regions, notably in Québec, Canada.

Your genetic map

Gene	SNP	Genotype
FAH	rs11555096	CC
FAH	rs80338901	GG
FAH	rs80338895	GG
FAH	rs80338900	GG
FAH	rs80338894	GG
FAH	rs80338898	CC
FAH	rs370686447	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/276700

Carrier Status

USHER SYNDROME, TYPE I; USH1

Usher syndrome (US) is characterized by the association of sensorineural deafness (usually congenital) with retinitis pigmentosa and progressive vision loss. Prevalence is estimated at 1/30,000. US is the most common cause of hereditary combined deafness-blindness.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/276900

Your genetic map

Gene	SNP	Genotype
MYO7A	rs397516281	TT
MYO7A	rs397516283	GG
MYO7A	rs111033206	GG
MYO7A	rs111033206	GG
MYO7A	rs111033389	GG
MYO7A	rs111033426	GG
MYO7A	rs111033180	CC
MYO7A	rs111033510	DD
MYO7A	rs397516291	CC
MYO7A	rs111033404	GG
MYO7A	rs111033404	GG
MYO7A	rs397516294	II
MYO7A	rs111033290	GG
MYO7A	rs111033433	II
MYO7A	rs111033239	II
MYO7A	rs111033482	AA
MYO7A	rs111033390	DD
MYO7A	rs397516301	GG
MYO7A	rs111033181	TT
MYO7A	rs111033202	II
MYO7A	rs397516310	TT
MYO7A	rs397516312	GG
MYO7A	rs397516315	TT
MYO7A	rs111033448	
MYO7A	rs111033182	CC
MYO7A	rs397516316	AA
MYO7A	rs397516320	DD
MYO7A	rs397516321	CC
MYO7A	rs397516322	GG
MYO7A	rs199606180	CC
MYO7A	rs397516323	TT

Carrier Status

USHER SYNDROME, TYPE ID; USH1D

USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/601067

Your genetic map

Gene	SNP	Genotype
CDH23	rs111033270	GG
PCDH15	rs111033260	GG
CDH23	rs727502931	GG
CDH23	rs397517313	II
CDH23	rs397517323	CC
CDH23	rs397517326	CC
CDH23	rs397517331	II
CDH23	rs397517337	CC
CDH23	rs397517341	GG
CDH23	rs397517342	GG
CDH23	rs397517346	GG
CDH23	rs183431253	GG
CDH23	rs111033473	II
CDH23	rs397517350	II
CDH23	rs397517353	GG
CDH23	rs397517354	GG
CDH23	rs397517362	CC
CDH23	rs397517367	II
CDH23	rs727502919	GG
CDH23	rs727504761	II
CDH23	rs397517327	CC
CDH23	rs397517329	CC
CDH23	rs727503841	GG

Carrier Status

USHER SYNDROME, TYPE IF; USH1F

Usher syndrome type I is characterized by congenital, bilateral, profound sensorineural hearing loss, vestibular areflexia, and adolescent-onset retinitis pigmentosa. Unless fitted with a cochlear implant, individuals do not typically develop speech. Retinitis pigmentosa (RP), a progressive, bilateral, symmetric degeneration of rod and cone functions of the retina, develops in adolescence, resulting in progressively constricted visual fields and impaired visual acuity.

Your genetic map

Gene	SNP	Genotype
PCDH15	rs137853001	GG
PCDH15	rs397517452	TT
PCDH15	rs202033121	GG
PCDH15	rs727504301	GG
PCDH15	rs137853003	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/602083

Carrier Status

USHER SYNDROME, TYPE IIA; USH2A

Usher syndrome type II is characterized by: Congenital, bilateral sensorineural hearing loss that is mild to moderate in the low frequencies and severe to profound in the higher frequencies; Intact vestibular responses; and Retinitis pigmentosa (RP). RP is progressive, bilateral, symmetric retinal degeneration that begins with night blindness and constricted visual fields (tunnel vision) and eventually includes decreased central visual acuity; the rate and degree of vision loss vary within and among families.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/276901

Your genetic map

Gene	SNP	Genotype
USH2A	rs146733615	GG
USH2A	rs397517978	TT
USH2A	rs397518003	TT
USH2A	rs397518008	II
USH2A	rs111033264	AA
USH2A	rs202175091	GG
USH2A	rs111033265	CC
USH2A	rs111033418	GG
USH2A	rs111033414	CC
USH2A	rs111033382	CC
USH2A	rs397517973	II
USH2A	rs397517974	CC
USH2A	rs397517976	CC
USH2A	rs397517977	CC
USH2A	rs397517979	CC
USH2A	rs111033526	CC
USH2A	rs397517981	AA
USH2A	rs111033417	CC
USH2A	rs397517988	DD
USH2A	rs397517989	CC
USH2A	rs397517994	GG
USH2A	rs397518011	GG
USH2A	rs397518012	II
USH2A	rs397518018	DD
USH2A	rs375668376	CC
USH2A	rs397518021	GG
USH2A	rs397518023	CC
USH2A	rs111033386	CC
USH2A	rs397518029	GG
USH2A	rs397518036	GG
USH2A	rs397518041	CC

Carrier Status

USHER SYNDROME, TYPE IIC; USH2C

Usher syndrome type II is characterized by: Congenital, bilateral sensorineural hearing loss that is mild to moderate in the low frequencies and severe to profound in the higher frequencies; Intact vestibular responses; and Retinitis pigmentosa (RP). RP is progressive, bilateral, symmetric retinal degeneration that begins with night blindness and constricted visual fields (tunnel vision) and eventually includes decreased central visual acuity; the rate and degree of vision loss vary within and among families.

Your genetic map

Gene	SNP	Genotype
ADGRV1	rs376689763	CC
ADGRV1	rs371981035	AA
ADGRV1	rs397517426	II
ADGRV1	rs373780305	CC
ADGRV1	rs397517429	DD
ADGRV1	rs397517436	GG
ADGRV1	rs397517441	II
ADGRV1	rs727504644	II

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/605472

Carrier Status

USHER SYNDROME, TYPE IID; USH2D

Usher syndrome type II is characterized by: Congenital, bilateral sensorineural hearing loss that is mild to moderate in the low frequencies and severe to profound in the higher frequencies; Intact vestibular responses; and Retinitis pigmentosa (RP). RP is progressive, bilateral, symmetric retinal degeneration that begins with night blindness and constricted visual fields (tunnel vision) and eventually includes decreased central visual acuity; the rate and degree of vision loss vary within and among families.

Your genetic map

Gene	SNP	Genotype
WHRN	rs397517255	GG
WHRN	rs397517258	II

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/611383

Carrier Status

USHER SYNDROME, TYPE IIIA; USH3A

Usher syndrome type III is characterized by postlingual, progressive hearing loss, variable vestibular dysfunction, and onset of retinitis pigmentosa symptoms, including nyctalopia, constriction of the visual fields, and loss of central visual acuity, usually by the second decade of life (Karjalainen et al., 1985; Pakarinen et al., 1995). For a discussion of phenotypic heterogeneity of Usher syndrome, see USH1 (276900). Genetic Heterogeneity of Usher syndrome Type III Usher syndrome type IIIB (614504) is caused by mutation in the HARS gene (142810) on chromosome 5q31.3.

Your genetic map

Gene	SNP	Genotype
CLRN1	rs121908140	AA
CLRN1	rs111033267	GG
CLRN1	rs111033434	CC
CLRN1	rs397517932	II
CLRN1	rs374963432	GG

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/276902

This report is not valid for clinical or diagnostic use.

Carrier Status

ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (VLCADD) is an inherited disorder of mitochondrial long-chain fatty acid oxidation with a variable presentation including: cardiomyopathy, hypoketotic hypoglycemia, liver disease, exercise intolerance and rhabdomyolysis. Over 400 cases have been reported worldwide. Prevalence in Germany is of 1/50, 000.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/201475

Your genetic map

Gene	SNP	Genotype
ACADVL	rs751995154	GG
ACADVL	rs387906251	
ACADVL	rs118204016	GG
ACADVL	rs113994170	CC
ACADVL	rs727503794	GG
ACADVL	rs140629318	GG
ACADVL	rs779901247	CC
ACADVL	rs200771970	GG
ACADVL	rs113690956	GG
ACADVL	rs118204014	CC
ACADVL	rs387906249	
ACADVL	rs2309689	GG
ACADVL	rs113994171	GG
ACADVL	rs113994168	CC
ACADVL	rs113994167	TT
ACADVL	rs398123080	TT
ACADVL	rs369560930	GG
ACADVL	rs398123092	AA
ACADVL	rs387906252	
ACADVL	rs753108198	II
ACADVL	rs545215807	GG
ACADVL	rs112406105	GG

Carrier Status

VON HIPPEL-LINDAU SYNDROME; VHL

Von Hippel-Lindau disease (VHL) is a familial cancer predisposition syndrome associated with a variety of malignant and benign neoplasms, most frequently retinal, cerebellar, and spinal hemangioblastoma, renal cell carcinoma (RCC), and pheochromocytoma. Prevalence is estimated at 1/53,000 and annual birth incidence at 1/36,000. Men and women are equally affected. Mean age at diagnosis is 26 years (range: infancy - 7th decade).

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/193300

Your genetic map

Gene	SNP	Genotype
VHL	rs193922613	AA
VHL	rs730882034	CC
VHL	rs5030807	TT
VHL	rs730882020	II
VHL	rs193922609	GG
VHL	rs193922609	GG
VHL	rs5030804	AA
VHL	rs398123481	CC
VHL	rs119103277	GG
VHL	rs121913346	TT
VHL	rs5030827	GG
VHL	rs193922610	CC
VHL	rs193922611	TT
VHL	rs397516440	CC
VHL	rs397516442	II
VHL	rs5030817	GG
VHL	rs397516445	TT
VHL	rs398123481	CC
VHL	rs398123482	TT
VHL	rs727504215	GG
VHL	rs5030826	CC

Carrier Status

WEAVER SYNDROME; WVS

Weaver syndrome (WVS) is a rare, multisystem disorder characterized by tall stature, a typical facial appearance (hypertelorism, retrognathia) and variable intellectual disability. Additional features may include camptodactyly, soft doughy skin, umbilical hernia, and a low hoarse cry. Around 50 cases of Weaver syndrome have been reported to date. Precise prevalence and incidence rates are not available.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/277590

Your genetic map

Gene	SNP	Genotype
EZH2	rs587783627	TT
EZH2	rs587783626	GG
EZH2	rs587783625	CC
EZH2	rs775407864	AA

Carrier Status

Wilson Disease

Wilson disease is a very rare inherited multisystemic disease presenting non-specific neurological, hepatic, psychiatric or osseo-muscular manifestations due to excessive copper deposition in the body.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/277900

Your genetic map

Gene	SNP	Genotype
ATP7B	rs121907992	CC
ATP7B	rs28942075	CC
ATP7B	rs121907998	AA
ATP7B	rs121908000	AA
ATP7B	rs121908001	CC
ATP7B	rs193922102	AA
ATP7B	rs193922108	CC
ATP7B	rs193922111	II
ATP7B	rs137853283	CC
ATP7B	rs372436901	TT
ATP7B	rs587783306	CC
ATP7B	rs587783307	TT
ATP7B	rs587783318	CC
ATP7B	rs184388696	CC
ATP7B	rs749085322	TT
ATP7B	rs768729972	DD
ATP7B	rs76151636	GG
ATP7B	rs28942074	CC
ATP7B	rs121907996	CC
ATP7B	rs121907999	GG
ATP7B	rs72552255	GG
ATP7B	rs193922107	GG
ATP7B	rs193922109	GG
ATP7B	rs193922110	CC
ATP7B	rs398123137	AA
ATP7B	rs201738967	TT
ATP7B	rs191312027	CC
ATP7B	rs121907990	TT
ATP7B	rs60431989	AA

Carrier Status

AGAMMAGLOBULINEMIA, X-LINKED; XLA

X-linked agammaglobulinemia (XLA) is a clinically variable form of isolated agammaglobulinemia, an inherited immunodeficiency disorder (see this term), and is characterized in affected males by recurrent bacterial infections during infancy. Estimated prevalence is 1/350,000 to 1/700,000. Annual incidence is not known. The disorder has been reported in various ethnic groups worldwide. Only males are affected and females are asymptomatic carriers.

What does your genetics say?

We have not detected any pathogenic mutation, but you might have some in non analyzed genetic regions.

More information:

https://www.omim.org/entry/300755

Your genetic map

Gene	SNP	Genotype
BTK	rs128620183	CC
BTK	rs128620187	GG
BTK	rs193922124	GG
BTK	rs193922125	TT
BTK	rs193922126	II
BTK	rs193922128	II
BTK	rs193922131	CC
BTK	rs193922132	TT
BTK	rs193922133	TT

Biomarkers

Adiponectin levels

Circulating levels of adiponectin, a hormone produced predominantly by adipocytes, are highly heritable and are inversely associated with type 2 diabetes mellitus (T2D) and other metabolic traits.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22479202

Your genetic map

Gene	SNP	Genotype
LOC1027	rs3001032	TT
LOC6467	rs1515110	TG
GNL3	rs1108842	
ADIPOQ	rs182052	AG
ARL15	rs6450176	GG
VEGFA -	rs998584	AA
LOC6454	rs668459	TT
TRIB1 -	rs2980879	TA
ADRB1 -	rs10885531	TC
PDE3B	rs11023332	GG
LOC1053	rs7955516	AC
ATP6V0A	rs6488898	AA
CDH13	rs12051272	GG
PEPD	rs731839	AA
PBRM1	rs2590838	AG
LOC1027	rs6810075	TC
LOC6454	rs592423	CC
TRIB1 -	rs2980879	TA
KNTC1 -	rs601339	AA
CMIP	rs2925979	TT
CDH13	rs12051272	GG
PEPD	rs4805885	CC

Biomarkers

Androgen levels in men

Circulating androgen levels are often used as indicators of physiological or pathological conditions. More than half of the variance for circulating androgen levels is thought to be genetically influenced. This item is valid only for men.

Your genetic map

Gene	SNP	Genotype
REEP3	rs10822184	TT
SHBG -	rs727428	TC
LOC1053	rs5934505	CC
SHBG -	rs727428	TC
ATP1B2	rs72829446	TC
ATP1B2	rs72829446	TC

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22936694

Biomarkers

Beta-2 microglubulin plasma levels

Beta-2 microglobulin (B2M) is a component of the major histocompatibility complex (MHC) class I molecule and has been studied as a biomarker of kidney function, cardiovascular diseases and mortality.

Your genetic map

Gene	SNP	Genotype
TRIM31-	rs2023472	GG
HLA-B	rs2523608	AG
LOC1019	rs16899524	CC
SH2B3	rs3184504	TC

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23417110

Biomarkers

Bilirubin levels

Variation in serum bilirubin is associated with altered cardiovascular disease risk and drug metabolism.

Your genetic map

Gene	SNP	Genotype
UGT1A8,	rs6742078	TT
HIST1H1T	rs12206204	CC
ARHGEF7	rs4773330	GG
SLCO1B1	rs4149056	TT

What does your genetics say?

●

According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/19414484

Biomarkers

C-reactive protein and white blood cell count

C-reactive protein (CRP) and white blood cell (WBC) have been utilized as critical markers contributing to acute and chronic inflammation.

What does your genetics say?

🟢

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22788528

Your genetic map

Gene	SNP	Genotype
DPF3,	rs2526932	AA
FLJ20021	rs6846071	TT
DOCK4	rs10255299	AG
LOC1053	rs6904416	TC
KCNE4 -	rs960246	GG
HNF1A	rs2393791	TT
LOC1053	rs7600502	AA
PSMD3 -	rs8078723	TT
LOC1005	rs16993221	AA

Biomarkers

Calcium levels

Calcium is vital to the normal functioning of multiple organ systems and its serum concentration is tightly regulated.

What does your genetics say?

●

According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/24068962

Your genetic map

Gene	SNP	Genotype
CASR	rs1801725	TG
DGKD	rs1550532	GG
GCKR	rs780094	TC
LOC1019	rs10491003	TC
CARS	rs7481584	GG
LOC1053	rs7336933	GG
CYP24A1	rs1570669	AG
WDR81	rs12150338	CC

Biomarkers

Dehydroepiandrosterone sulphate levels

Dehydroepiandrosterone sulphate (DHEAS) is the most abundant circulating steroid secreted by adrenal glands--yet its function is unknown. Its serum concentration declines significantly with increasing age, which has led to speculation that a relative DHEAS deficiency may contribute to the development of common age-related diseases or diminished longevity.

Your genetic map

Gene	SNP	Genotype
ZKSCAN5	rs11761528	CC
SULT2A1	rs2637125	AG
SRP14-	rs7181230	AA
HHEX -	rs2497306	AC
LOC1079	rs2185570	TT
TRIM4	rs17277546	AG
BCL2L11	rs6738028	CC
ARPC1A	rs740160	CC

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/21533175

Biomarkers

Eosinophil counts

Eosinophils are pleiotropic multifunctional leukocytes involved in initiation and propagation of inflammatory responses and thus have important roles in the pathogenesis of inflammatory diseases

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/19198610

Your genetic map

Gene	SNP	Genotype
IL1RL1	rs1420101	TC
LOC1027	rs12619285	AG
TMED10P	rs4857855	CC
SH2B3	rs3184504	TC
IRF1 - IL5	rs4143832	GG
WDR36	rs2416257	CC
TNXB	rs2269426	GG

Biomarkers

Glycated hemoglobin levels

Glycated hemoglobin A1c (HbA1c) is used as a measure of glycemic control and also as a diagnostic criterion for diabetes.

Your genetic map

Gene	SNP	Genotype
TMEM79,	rs6684514	GG
LOC1079	rs9399137	TT
FADS2	rs174570	TC
PIEZO1	rs9933309	CC
MYO9B	rs11667918	TC
ANK1	rs4737009	AG
FN3KRP	rs1046875	AG
ABCB11	rs3755157	CC
CDKAL1	rs7772603	TC
GCK -	rs1799884	CC
LOC1053	rs13266634	TC

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/24647736

Biomarkers

Homocysteine levels

Homocysteine (HC) is a sulfur amino acid important in the transfer of methyl groups in cell metabolism, this has been considered an influential factor in the development of cardiovascular and cerebrovascular diseases.

Recent studies have focused on the analysis of the relationship between hyperhomocysteinemia (increased plasma homocysteine concentration) and damage to neuronal cells; in neurotoxic mechanisms such as: increase of oxidative stress and generation of homocysteine derivatives as well as the increase in the toxicity of β-amyloid protein, among others.

Homocysteine is synthesized as an intermediate product of the metabolism of methionine by the action of the enzyme methionine adenosyl transferase.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23824729

Your genetic map

Gene	SNP	Genotype
MTHFR	rs1801133	GG
MTR	rs2275565	GG
EEF1A1P4	rs9369898	
NOX4	rs7130284	CC
DPEP1 -	rs154657	AG
CBS	rs234709	TC
PRDX1	rs4660306	TC
SLC17A3	rs548987	GG
LOC1079	rs42648	AG
RPL12P33	rs2251468	AA
FGF21	rs838133	AG
C1orf167,	rs12134663	
TRDMT1	rs12780845	AA
NOX4	rs957140	GG
CBS	rs2851391	TC

Biomarkers

IgE levels

Atopy and plasma IgE concentration are genetically complex traits, and the specific genetic risk factors that lead to IgE dysregulation and clinical atopy are an area of active investigation

Your genetic map

Gene	SNP	Genotype
FCER1A	rs2251746	TT
STAT6	rs1059513	TT
IL13	rs20541	AG
LOC1053	rs2523809	GG
HLA-W	rs2571391	
ACKR1	rs13962	
MTCO3P	rs2858331	GG
OR10J7P	rs4656784	AA
LPP	rs9290877	TT

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22075330

Biomarkers

Liver enzyme levels (gamma-glutamyl transferase)

Concentrations of liver enzymes in plasma are widely used as indicators of liver disease.

Your genetic map

Gene	SNP	Genotype
PNPLA3	rs738409	CG
NBPF3	rs1976403	AC
RNU6	rs6984305	TT
LOC1053	rs10819937	GG
ABO -	rs579459	TC
JMJD1C	rs7923609	AA
FADS2	rs174601	TT
ST3GAL4	rs2236653	TC
ASGR1 -	rs314253	TC
ABHD12	rs7267979	AG
LOC1019	rs1497406	AA
CEPT1	rs1335645	AA
EFHD1	rs2140773	AC
SLC2A2	rs10513686	AG
HPRT1P2	rs6888304	AA
MLXIPL	rs17145750	CC
DLG5	rs754466	AA
HNF1A	rs7310409	GG
EXOC3L4	rs944002	AG
RORA,	rs339969	AA
CD276	rs8038465	TT
LOC1027	rs4581712	CC
SOX9-	rs9913711	CC
FUT2,	rs516246	TC
MICAL3	rs1076540	CC
GGT1	rs2073398	GC

What does your genetics say?

●

According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22001757

Biomarkers

Liver enzyme levels

Plasma liver-enzyme tests are widely used in the clinic for the diagnosis of liver diseases and for monitoring the response to drug treatment. There is considerable evidence that human genetic variation influences plasma levels of liver enzymes

Your genetic map

Gene	**SNP**	**Genotype**
JMJD1C	rs12355784	CC
JMJD1C	rs10761779	AA
LINC0136	rs9803659	TT
ADAMTS1	rs4962153	AA
PNPLA3	rs2281135	GG
NBPF3 -	rs1780324	GG
	rs657152	AC
GPLD1	rs9467160	GG
GGT1	rs4820599	GG

What does your genetics say?

●

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/18940312

Biomarkers

Magnesium levels

Magnesium, potassium, and sodium, cations commonly measured in serum, are involved in many physiological processes including energy metabolism, nerve and muscle function, signal transduction, and fluid and blood pressure regulation.

Your genetic map

Gene	SNP	Genotype
MUC1	rs4072037	TC
SHROOM	rs13146355	GG
LOC1079	rs7965584	AA
LOC1019	rs3925584	TT
HOXD9 -	rs2592394	AG
MECOM	rs448378	GG

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20700443

Biomarkers

Monocyte count

Monocytes are a type of agranulocyte white blood cells. It is the largest leukocyte.

With white blood cell count emerging as an important risk factor for chronic inflammatory diseases, genetic associations of differential leukocyte types, specifically monocyte count, are providing novel candidate genes and pathways to further investigate. Circulating monocytes play a critical role in vascular diseases such as in the formation of atherosclerotic plaque

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23314186

Your genetic map

Gene	SNP	Genotype
ITGA4	rs2124440	GG
LINC0156	rs2712381	CC
ACKR2	rs2228467	TC
PTGR1	rs2273788	CC
IRF8	rs424971	CC

Biomarkers

Phospholipid levels (plasma)

Long-chain n-3 polyunsaturated fatty acids (PUFAs) can derive from diet or from α-linolenic acid (ALA) by elongation and desaturation

What does your genetics say?

🟢

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/21829377

Your genetic map

Gene	SNP	Genotype
TMEM25	rs102275	CC
MYRF	rs174536	CC
RPLP0P2	rs1692120	AA
FADS1	rs174547	CC
FADS2	rs1535	GG
FADS2 -	rs174448	GG
FEN1	rs4246215	TT
UBXN4 -	rs16832011	AA
TMEM25	rs174538	AA
MYRF	rs174535	CC
FADS1	rs174550	CC
FADS2	rs174574	AA
FEN1	rs4246215	TT
FADS2 -	rs174448	GG
ELOVL2	rs3798713	GG
BEST1	rs1109748	CC
LOC1019	rs1514178	TT
FADS1	rs174547	CC
TMEM25	rs102275	CC
FADS2	rs1535	GG
MYRF	rs174535	CC
FEN1	rs4246215	TT
FADS2 -	rs174448	GG
ELOVL2	rs3734398	TT
SYCP2L	rs4713103	TG
RAB3IL1	rs2521572	GG
DAGLA	rs198426	CC
GCKR	rs780094	TC
LOC1053	rs9586179	CC
RPS2P37	rs4963452	
STIM2	rs6844153	TC

Biomarkers

Phosphorus levels

Phosphorus is an essential mineral that maintains cellular energy and mineralizes the skeleton. Because complex actions of ion transporters and regulatory hormones regulate serum phosphorus concentrations, genetic variation may determine interindividual variation in phosphorus metabolism.

Your genetic map

Gene	SNP	Genotype
NBPF3 -	rs1697421	TT
CSTA	rs17265703	AG
IP6K3	rs9469578	CC
PDE7B	rs947583	TC
C12orf4	rs2970818	TT

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20558539

Biomarkers

Plasma omega-6 polyunsaturated fatty acid levels (dihomo-gamma-linolenic acid)

Omega6 (n6) polyunsaturated fatty acids (PUFAs) and their metabolites are involved in cell signaling, inflammation, clot formation, and other crucial biological processes. Genetic components, such as variants of fatty acid desaturase (FADS) genes, determine the composition of n6 PUFAs.

What does your genetics say?

●

According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/24823311

Your genetic map

Gene	SNP	Genotype
PDXDC1	rs2280018	AA
PDXDC1	rs2280018	AA
TMEM25	rs102275	CC
IL23R	rs7517847	TT
C10orf12	rs17009617	GG
FADS1	rs174550	CC
FADS2	rs2727270	CC
PDXDC1,	rs1136001	GG
FTLP19 -	rs2069036	TC
FADS1	rs174547	CC
PDXDC1	rs4985155	AA
TMEM39	rs16829840	CC
FADS1	rs174547	CC
PDXDC1	rs1741	GG
ELOVL2	rs2236212	GG
FADS1	rs174550	CC
FADS1	rs174555	CC
PDXDC1	rs4985155	AA

Biomarkers

Platelet count

Platelets are small fragments of blood cells. Its function is to form blood clots that help to heal wounds and prevent bleeding. The bone marrow produces platelets. Problems can arise when you have too few or too many platelets or they do not perform their function correctly.

If the blood has few platelets, it is called thrombocytopenia and there is a risk of moderate to severe bleeding. If the blood contains too many platelets, there is a risk of blood clots.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22139419

Your genetic map

Gene	SNP	Genotype
MFN2	rs2336384	GG
DNM3	rs10914144	
TMCC2	rs1668871	CC
GCSAML	rs7550918	TT
TRIM58	rs3811444	TC
EHD3	rs625132	GG
THADA	rs17030845	TC
LOC3398	rs7641175	AA
ARHGEF3	rs1354034	TT
PDIA5	rs3792366	AG
KLHL8 -	rs7694379	AG
F2R -	rs17568628	TC
MEF2C	rs700585	TT
IRF1	rs2070729	AC
LRRC16A	rs441460	AG
HLA-B	rs3819299	TT
HLA-	rs399604	TC
RN7SL26	rs210134	GG
LOC1079	rs9399137	TT
LOC1027	rs342275	CC
HYAL4	rs4731120	AA
PLEC	rs6995402	TC
AK3 -	rs409801	TC
RCL1	rs13300663	CG
CDKN2A	rs3731211	TA
BRD3	rs11789898	
PSMD13,	rs505404	TG
FEN1	rs4246215	TT
CBL	rs4938642	GG
LOC1053	rs7342306	GG
BAZ2A	rs941207	GG

Biomarkers

Red blood cell count

Hemoglobin is a protein present in red blood cells that carries oxygen to your body's organs and tissues and transports carbon dioxide from organs and tissues back to the lungs. If the level of hemoglobin is lower than normal, it means that we have a low red blood cell count (anemia).

What does your genetics say?

🟢

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20139978

Your genetic map

Gene	SNP	Genotype
PRKCE	rs10168349	CC
ABO -	rs495828	TG
LOC1053	rs7173947	TC
ALPL	rs2242420	TC
GPLD1	rs6911965	TT
PNPLA3	rs2896019	TT
BRAP	rs3782886	TT
MRC1	rs2477664	
PNPLA3	rs2896019	TT
LOC1053	rs9820070	AC
SLC14A2,	rs4890568	AA
LOC1053	rs11709625	CC
CD163 -	rs7136716	AA
GGT1	rs5751902	CC
ALDH2	rs671	GG
TMPRSS6	rs5756504	CC
ABO -	rs495828	TG
PRKCE	rs10495928	GG
LIPC,	rs1077834	TC
LOC1019	rs7350481	CC
HERPUD1	rs3764261	CC
LPL -	rs12678919	AA
LOC1079	rs7775698	CC
ABO -	rs495828	TG
TMPRSS6	rs2413450	TC
WDR72	rs10518733	AC
TNFRSF1	rs4273077	AA
RPS11	rs2280401	GG
HBA2 -	rs2858942	CC
LOC1079	rs7775698	CC
RCL1	rs2236496	TC

Biomarkers

Serum albumin level

Many disorders are associated with altered serum protein concentrations, including malnutrition, cancer, and cardiovascular, kidney, and inflammatory diseases.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23022100

Your genetic map

Gene	SNP	Genotype
MIR22HG	rs11078597	TT
ACTBP9 -	rs694419	CC
RPS11	rs2280401	GG
FRMD5	rs16948098	GG
TNFRSF1	rs4561508	CC
FKBPL -	rs204999	AA
TNFRSF1	rs4561508	CC
LOC1079	rs2675609	CC
RPS11	rs2280401	GG
HPN-AS1	rs11671010	TT
MIR22HG	rs11078597	TT
ACTBP9 -	rs694419	CC
RPS11	rs2280401	GG
CHRNA3	rs12914385	CC
TNFRSF1	rs4561508	CC
FKBPL -	rs204999	AA
ELL2	rs3777200	CC
RPS11	rs2280401	GG

Biomarkers

Sex hormone levels

Genetic factors contribute strongly to sex hormone levels, yet knowledge of the regulatory mechanisms remains incomplete.

What does your genetics say?

●

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/26014426

Your genetic map

Gene	SNP	Genotype
ZNF789	rs148982377	CC
LOC1462	rs117145500	AA
LOC1053	rs11031005	TT
LOC1053	rs11031002	TT
ANO2,	rs117585797	CC
ZKSCAN5	rs34670419	GG
SLC22A2	rs112295236	CC
ZNF789	rs148982377	CC
ZNF789	rs148982377	CC
ZKSCAN5	rs34670419	GG
ZKSCAN5	rs34670419	GG
SULT2A1	rs2637125	AG
LOC1027	rs12294104	CC
LOC1027	rs12294104	CC

Biomarkers

Thyroid hormone levels

Thyroid hormone is essential for normal metabolism and development, and overt abnormalities in thyroid function lead to common endocrine disorders affecting approximately 10% of individuals over their life span. In addition, even mild alterations in thyroid function are associated with weight changes, atrial fibrillation, osteoporosis, and psychiatric disorders.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23408906

Your genetic map

Gene	SNP	Genotype
PDE8B	rs6885099	AG
PDE10A	rs753760	CC
LOC1053	rs10799824	GG
LOC1053	rs3813582	TC
LOC1079	rs9472138	TC
LINC0151	rs11755845	CC
LOC1079	rs10032216	TT
LOC1019	rs13015993	AA
SOX9 -	rs9915657	CC
NFIA	rs334699	GG
FAM227B,	rs10519227	TT
PRDM11	rs17723470	CC
DET1 -	rs17776563	AG
INSR	rs4804416	TT
	rs657152	AC
ITPK1 -	rs11624776	AA
NRG1	rs7825175	AG
LINC006	rs1537424	TC
SASH1	rs9497965	CC
GLIS3	rs1571583	AG
DIO1	rs2235544	CC
LHX3	rs7860634	AG
KRT18P13	rs7045138	TT
LOC1053	rs11726248	GG
LPCAT2	rs6499766	TT
PDE8B	rs6885099	AG
PDE8B	rs6885099	AG
PDE10A	rs753760	CC
PDE10A	rs753760	CC
LOC1053	rs10799824	GG
LOC1053	rs10799824	GG

Biomarkers

Uric acid levels

Elevated serum uric acid levels cause gout and are a risk factor for cardiovascular disease and diabetes.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/19503597

Your genetic map

Gene	SNP	Genotype
PDZK1 -	rs12129861	AG
GCKR	rs780094	TC
SLC2A9	rs734553	TT
ABCG2	rs2231142	GG
LRRC16A	rs742132	AG
SLC17A1	rs1183201	AT
SLC16A9	rs12356193	AG
SLC22A11	rs17300741	AG
SLC22A11	rs505802	TT
SLC2A9	rs734553	TT
ABCG2	rs2231142	GG

Biomarkers

Urinary uromodulin levels

Uromodulin is expressed exclusively in the thick ascending limb and is the most abundant protein excreted in normal urine. Variants in UMOD, which encodes uromodulin, are associated with renal function, and urinary uromodulin levels may be a biomarker for kidney disease.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/24578125

Your genetic map

Gene	SNP	Genotype
PDILT	rs12446492	AT
UMOD -	rs12917707	TG
MARCH1	rs4533720	AA
PDILT	rs4494548	GG

Biomarkers

Vitamin B levels in ischemic stroke

B vitamins play an important role in homocysteine metabolism, with vitamin deficiencies resulting in increased levels of homocysteine and increased risk for stroke.

Your genetic map

Gene	SNP	Genotype
NBPF3 -	rs1697421	TT
TCN1	rs34324219	CC
RASIP1	rs2287921	TC
FUT2,	rs492602	AG

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/25147783

Biomarkers

White blood cell count

White blood cells are a type of blood cell that is produced in the bone marrow and found in blood and lymphatic tissues. White blood cells are part of the body's immune system. These help the body fight infections and other diseases. The types of white blood cells are granulocytes (neutrophils, eosinophils, and basophils), monocytes, and lymphocytes (T cells and B cells).

The white blood cell count is a common clinical measure from the whole blood count assays, and varies widely among healthy individuals.

What does your genetics say?

●

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/21738480

Your genetic map

Gene	SNP	Genotype
LINC0156	rs4328821	AA
EPS15L1	rs10411936	GG
LOC1019	rs1449263	CC
LINC0156	rs9880192	GG
CCDC26	rs10098310	AA
LOC1053	rs10980800	TC
PSMD3 -	rs8078723	TT
HCG22 -	rs2517510	TG
PSMD3 -	rs4794822	CC

Biometrics

Aortic root size

Echocardiographic measures of left ventricular (LV) structure and function are heritable phenotypes of cardiovascular disease.

Your genetic map

Gene	SNP	Genotype
SLC35F1	rs89107	AG
TMEM23	rs17132261	CC
SMG6	rs10852932	GG
PRDM6 -	rs17470137	GG
HMGA2 -	rs4026608	TC
LOC1005	rs10770612	AA
LOXL1	rs893817	AG

What does your genetics say?

●

According to this study, you have a better predisposition than the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/19584346

This report is not valid for clinical or diagnostic use.

Biometrics

Bone mineral density

Bone mineral density (BMD) is the most widely used predictor of fracture risk.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22504420

Your genetic map

Gene	SNP	Genotype
ABCF2	rs7812088	GG
FABP3P2	rs9533090	TC
ARHGAP1	rs7932354	TC
AXIN1	rs9921222	TC
TMEM26	rs1053051	TT
RPS3AP2	rs13336428	AG
C17orf53	rs227584	AA
FAM210A	rs4796995	AG
CCDC170	rs4869742	CC
CPED1	rs13245690	AA
LOC1001	rs4817775	AA
CPN1	rs7084921	CC
LOC1053	rs430727	TC
LOC1079	rs1564981	AG
DCDC5	rs163879	CC
RHEBL1 -	rs12821008	CC
DNM3	rs479336	TG
LOC1079	rs2887571	AA
FOXL1 -	rs10048146	AA
FUBP3	rs7851693	GC
CSRNP3	rs1346004	AG
GPATCH1	rs10416218	TC
HOXC6,	rs736825	CC
IDUA	rs3755955	GG
LOC1053	rs1878526	AA
JAG1	rs3790160	CC
KCNMA1	rs7071206	TC
KIAA2018	rs1026364	TG
LOC1053	rs7953528	TT
LEKR1	rs344081	TC
RPL37AP	rs10835187	

Biometrics

Heart rate

Elevated resting heart rate is associated with greater risk of cardiovascular disease.

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23583979

Your genetic map

Gene	SNP	Genotype
TFPI,	rs4140885	AG
LOC1053	rs180242	AA
RNU3P3	rs17796783	TC
SYT10	rs7980799	AC
LOC1053	rs17287293	AG
CD46	rs11118555	TT
MYH6	rs365990	AA
LOC1053	rs1015451	TT
ACHE -	rs13245899	AA
FADS1	rs174549	AA
SLC35F1	rs11153730	TT
KIAA1755	rs6127471	TT
CCDC141	rs17362588	GG
GNB4 -	rs7612445	TG
CHRM2,	rs2350782	TC
NKX2-5 -	rs6882776	GG
LOC1053	rs13030174	AA
FNDC3B	rs9647379	CC
RFX4,	rs2067615	TT
CPNE8	rs826838	TC
RBFOX1	rs11645781	AG
SLC10A7	rs10213084	TG
RNU4	rs11154027	TC
LOC1079	rs11578508	AG
HMGN2P	rs17083533	GG
LOC1019	rs7722600	AA

Biometrics

Resting heart rate

Higher resting heart rate is associated with increased cardiovascular disease and mortality risk

Your genetic map

Gene	SNP	Genotype
LOC1053	rs9398652	CC
MYH6	rs452036	GG
NGDN -	rs223116	AG
LOC1053	rs17287293	AG
SLC35F1	rs281868	AG
SLC12A9	rs314370	TT
UFSP1	rs12666989	GG
FADS1	rs174547	CC

What does your genetics say?

According to this study, you have a similar predisposition to the majority of the population to have normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20639392

Traits

Alcoholism (alcohol dependence factor score)

Given moderately strong genetic contributions to variation in alcoholism and heaviness of drinking (50% to 60% heritability) with high correlation of genetic influences

Your genetic map

Gene	SNP	Genotype
LOC1079	rs2827312	TG
KRT18P5	rs2548145	AA
MBNL2	rs9556711	GG
DCC	rs768048	CC
LOC1053	rs10253361	TC
MBNL2	rs9556711	GG
LINC009	rs933769	TT
COL6A1 -	rs4293630	AA
HIP1	rs237238	AA

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21529783

Traits

Spirometric measure of pulmonary function (Forced vital capacity)

Forced vital capacity (FVC), a spirometric measure of pulmonary function, reflects lung volume and is used to diagnose and monitor lung diseases.

Your genetic map

Gene	SNP	Genotype
EFEMP1	rs1430193	AA
BMP6	rs6923462	TC
MIR129-2	rs4237643	GG
PRDM11	rs2863171	AC
WWOX	rs1079572	GG

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24929828

Traits

Smoking behavior

Consistent but indirect evidence has implicated genetic factors in smoking behavior.

Your genetic map

Gene	SNP	Genotype
HECTD2-	rs1329650	TG
RAB4B-	rs3733829	AA
BDNF,	rs6265	CC
FAM163B	rs3025343	GG

What does your genetics say?

According to this study, you have a predisposition similar to most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20418890

Pharmacogenomics: Cardiology

Pravastatin

Pravastatin is a cholesterol-lowering agent that belongs to a class of medications known as statins. It was derived from microbial transformation of mevastatin, the first statin discovered. It is a ring-opened dihydroxyacid with a 6′-hydroxyl group that does not require in vivo activation. Pravastatin is one of the lower potency statins; however, its increased hydrophilicity is thought to confer advantages such as minimal penetration through lipophilic membranes of peripheral cells, increased selectivity for hepatic tissues, and a reduction in side effects compared with lovastatin and simvastatin.

Your genetic map

Gene	SNP	Genotype
HMGCR	rs17244841	AA

What does your genetics say?

🟢

Patients with the AA genotype who are treated with statins may be more likely to respond as compared to patients with the AT or TT genotype. Other genetic and clinical factors may also influence a patient's response when treated with statins.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15199031

Pharmacogenomics: Cardiology

Simvastatin

Simvastatin is a lipid-lowering agent that is derived synthetically from the fermentation of Aspergillus terreus. It is a potent competitive inhibitor of 3-hydroxy-3-methylglutaryl coenzyme A reductase (hydroxymethylglutaryl COA reductases), which is the rate-limiting enzyme in cholesterol biosynthesis. It may also interfere with steroid hormone production. Due to the induction of hepatic LDL receptors, it increases breakdown of LDL cholesterol.

Your genetic map

Gene	SNP	Genotype
SLCO1B1	rs4149056	TT

What does your genetics say?

Patients with the TT genotype may have a lower risk of simvastatin-related myopathy as compared to patients with the CT or CC genotype. Other genetic and clinical factors may also influence a patient's risk for toxicity.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/28482130

This report is not valid for clinical or diagnostic use.

Pharmacogenomics: Cardiology

Warfarin

Warfarin is an anticoagulant drug normally used to prevent blood clot formation as well as migration. Although originally marketed as a pesticide (d-Con, Rodex, among others), Warfarin has since become the most frequently prescribed oral anticoagulant in North America. Warfarin has several properties that should be noted when used medicinally, including its ability to cross the placental barrier during pregnancy which can result in fetal bleeding, spontaneous abortion, preterm birth, stillbirth, and neonatal death. Additional adverse effects such as necrosis, purple toe syndrome, osteoporosis, valve and artery calcification, and drug interactions have also been documented with warfarin use. Warfarin does not actually affect blood viscosity, rather, it inhibits vitamin-k dependent synthesis of biologically active forms of various clotting factors in addition to several regulatory factors.

What does your genetics say?

●

Patients with the TT genotype may require a lower dose of warfarin as compared to patients with the CC or TC genotype. Other genetic and clinical faTCors may also influence a patient's warfarin dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/gtr/conditions/CN078029

Your genetic map

Gene	SNP	Genotype
VKORC1	rs9923231	TT

Pharmacogenomics: Neurology

Amitriptyline

Amitriptyline hydrochloride is a dibenzocycloheptene-derivative tricyclic antidepressant (TCA). TCAs are structurally similar to phenothiazines. They contain a tricyclic ring system with an alkyl amine substituent on the central ring. In non-depressed individuals, amitriptyline does not affect mood or arousal, but may cause sedation. In depressed individuals, amitriptyline exerts a positive effect on mood. TCAs are potent inhibitors of serotonin and norepinephrine reuptake. Tertiary amine TCAs, such as amitriptyline, are more potent inhibitors of serotonin reuptake than secondary amine TCAs, such as nortriptyline. TCAs also down-regulate cerebral cortical β-adrenergic receptors and sensitize post-synaptic serotonergic receptors with chronic use. The antidepressant effects of TCAs are thought to be due to an overall increase in serotonergic neurotransmission. TCAs also block histamine-H1 receptors, α1-adrenergic receptors and muscarinic receptors, which accounts for their sedative, hypotensive and anticholinergic effects (e.g. blurred vision, dry mouth, constipation, urinary retention), respectively.

What does your genetics say?

●

Patients with the GG genotype who are treated with amitriptyline may have increased metabolism of amitriptyline (decreased amitriptyline plasma concentrations and increased nortriptyline plasma concentrations) as compared to patients with the AA or AG genotype. Other genetic factors, including other CYP2C19 alleles *17 rs12248560 and *3 rs4986893, along with clinical factors, may also influence a patient's required dose and should be taken into consideration.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/16024198

Your genetic map

Gene	SNP	Genotype
CYP2C19	rs4244285	GG

Pharmacogenomics: Neurology

Antidepressants

It's thought that antidepressants work by increasing levels of a group of chemicals in the brain called neurotransmitters. Certain neurotransmitters, such as serotonin and noradrenaline, can improve mood and emotion, although this process isn't fully understood. Increasing levels of neurotransmitters can also disrupt pain signals sent by nerves, which may explain why some antidepressants can help relieve long-term pain

Your genetic map

Gene	SNP	Genotype
GRIK4	rs1954787	TC

What does your genetics say?

●

Patients with the TC genotype and Depressive Disorder or Depression may be less likely to respond to antidepressant treatment as compared to patients with the CC genotype. Other genetic and clinical factors may also influence a patient's response to anti-depressants.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25303296

Pharmacogenomics: Neurology

Bupropion

A unicyclic, aminoketone antidepressant. The mechanism of its therapeutic actions is not well understood, but it does appear to block dopamine uptake. The hydrochloride is available as an aid to smoking cessation treatment.

What does your genetics say?

🟢

Patients with the GG genotype who are treated with bupropion may be more likely to quit smoking as compared to patients with the AA or AG genotypes, although this has been contradicted in one study. Other genetic and clinical factors may also influence a patient's chance for quitting smoking.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15492764

Your genetic map

Gene	SNP	Genotype
ANKK1	rs1800497	GG

Pharmacogenomics: Oncology

Methotrexate

An antineoplastic antimetabolite with immunosuppressant properties. It is an inhibitor of tetrahydrofolate dehydrogenase and prevents the formation of tetrahydrofolate, necessary for synthesis of thymidylate, an essential component of DNA.

Your genetic map

Gene	SNP	Genotype
MTHFR	rs1801133	GG

What does your genetics say?

●

Patients with the GG genotype and leukemia or lymphoma who are treated with methotrexate: 1) may have better response to treatment 2) may be at decreased risk of toxicity 3) may require a higher dose of methotrexate, and 4) may be at lower risk of folate deficiency as compared to patients with the AA or AG genotype. This association has been contradicted or not found in multiple studies. Other genetic and clinical factors may also influence a patient's risk for toxicity and response with methotrexate treatment.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/22143415

Pharmacogenomics: Oncology

Vincristine

Vincristine is an antitumor vinca alkaloid isolated from Vinca Rosea. It is marketed under several brand names, many of which have different formulations such as Marqibo (liposomal injection) and Vincasar. Vincristine is indicated for the treatment of acute leukaemia, malignant lymphoma, Hodgkin's disease, acute erythraemia, and acute panmyelosis. vincristine sulfate is often chosen as part of polychemotherapy because of lack of significant bone–marrow suppression (at recommended doses) and of unique clinical toxicity (neuropathy).

Your genetic map

Gene	SNP	Genotype
LOC1009	rs924607	TT

What does your genetics say?

●

Patients with the TT genotype may have increased risk of peripheral nervous system diseases when treated with vincristine may have as compared to patients with the CC or TC genotype. Other genetic and clinical faTCors may also influence a patient's response to vincristine.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25710658

Pharmacogenomics: Oncology

Fluorouracil, capecitabine, pyrimidine analogues, tegafur and Neoplasms

Fluorouracil (5-FU), sold under the brand name Adrucil among others, is a medication used to treat cancer. By injection into a vein it is used for colon cancer, esophageal cancer, stomach cancer, pancreatic cancer, breast cancer, and cervical cancer. As a cream it is used for actinic keratosis and basal cell carcinoma.

It is a potent antimetabolite used in the treatment of cancer. It is a drug that blocks the methylation reaction of deoxyuridic acid to convert it into thymidylic acid by inhibiting an enzyme that is important for the synthesis of thymidine, which being part of the DNA molecule stops its formation. The drug is specific to the cell phase cycle, S phase. 5-Fluorouracil intervenes in the synthesis of DNA and inhibits to a small degree the formation of RNA. Both actions combine to promote a metabolic imbalance that results in cell death. The inhibitory activity of the drug, by its analogy with uracil, has an effect on the rapid growth of the neoplastic cells that preferentially take advantage of the uracil molecule for nucleic acid biosynthesis. The effects of a deprivation of DNA and RNA attack more cells that grow and

Your genetic map

Gene	SNP	Genotype
DPYD	rs67376798	TT

What does your genetics say?

Patients TT genotype treated with fluoropyrimidine-based chemotherapy may have 1) increased clearance of the drug and 2) decreased, but not absent, risk and reduced severity of drug toxicity as compared to patients with the AT genotype. The combination (FOLFOX, FOLFIRI or FEC) and delivery of the drug may influence risk for toxicity. Other genetic and clinical factors may also influence.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/17700593

Pharmacogenomics: Other

Tacrolimus

Tacrolimus (also FK-506 or Fujimycin) is an immunosuppressive drug whose main use is after organ transplant to reduce the activity of the patient's immune system and so the risk of organ rejection. It is also used in a topical preparation in the treatment of severe atopic dermatitis, severe refractory uveitis after bone marrow transplants, and the skin condition vitiligo. It was discovered in 1984 from the fermentation broth of a Japanese soil sample that contained the bacteria Streptomyces tsukubaensis. Tacrolimus is chemically known as a macrolide. It reduces peptidyl-prolyl isomerase activity by binding to the immunophilin FKBP-12 (FK506 binding protein) creating a new complex. This FKBP12-FK506 complex interacts with and inhibits calcineurin thus inhibiting both T-lymphocyte signal transduction and IL-2 transcription.

Your genetic map

Gene	SNP	Genotype
CYP3A4	rs2740574	TT

What does your genetics say?

●

Transplant recipients with the TT (CYP3A4 genotype may require a decreased dose of tacrolimus as compared to patients with the TC or CC genotype. Other genetic and clinical faTCors, such as CYP3A5 (rs776746), may also influence a patient's dose requirements.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23778326

Pharmacogenomics: Other

Sildenfail (Viagra)

Sildenfail is a vasoactive agent used to treat erectile dysfunction and reduce symptoms in patients with pulmonary arterial hypertension (PAH). Sildenafil elevates levels of the second messenger, cGMP, by inhibiting its breakdown via phosphodiesterase type 5 (PDE5). PDE5 is found in particularly high concentrations in the corpus cavernosum, erectile tissue of the penis. It is also found in the retina and vascular endothelium. Increased cGMP results in vasodilation which facilitates generation and maintenance of an erection.

Your genetic map

Gene	SNP	Genotype
GNB3	rs5443	CC

What does your genetics say?

●

Patients with the CC genotype and erectile dysfunction who are treated with sildenafil may be less likely to have positive erectile response as compared to patients with the TT genotype. Other genetic and clinical factors may also influence a patient's response to sildenafil.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/12576843

Pharmacogenomics: Pain

Meperidine

A narcotic analgesic that can be used for the relief of most types of moderate to severe pain, including postoperative pain and the pain of labor. Prolonged use may lead to dependence of the morphine type; withdrawal symptoms appear more rapidly than with morphine and are of shorter duration.

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

What does your genetics say?

●

Patients with the TC genotype may have decreased opioid analgesic requirements after surgery. Other genetic and clinical factors may influence.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenomics: Pain

Morphine

The principal alkaloid in opium and the prototype opiate analgesic and narcotic. Morphine has widespread effects in the central nervous system and on smooth muscle. In January, 2017, morphine was approved for the treatment of chronic pain.

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

What does your genetics say?

●

Patients with the TC genotype may have decreased opioid analgesic requirements after surgery as compared to patients with the CC genotype. Other genetic and clinical factors may influence a patient's opioid dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenomics: Pain

Pentazocine

The first mixed agonist-antagonist analgesic to be marketed. It is an agonist at the kappa and sigma opioid receptors and has a weak antagonist action at the mu receptor

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

What does your genetics say?

●

Patients with the TC genotype may have decreased opioid analgesic requirements after surgery as compared to patients with the CC genotype. Other genetic and clinical faTCors may influence a patient's opioid dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenomics: Pain

Aspirin

Aspirin, also known as acetylsalicylic acid (ASA), is a medication used to treat pain, fever, or inflammation. Specific inflammatory conditions in which aspirin is used include Kawasaki disease, pericarditis, and rheumatic fever. Aspirin is a nonsteroidal anti-inflammatory drug (NSAID) and works similar to other NSAIDs but also suppresses the normal functioning of platelets.

Your genetic map

Gene	SNP	Genotype
PTGS1	rs10306114	AA

What does your genetics say?

Patients with the AA genotype who are treated with aspirin may have a decreased, but not absent, risk for non-response to aspirin as compared to patients with the AG or GG genotype. Other genetic and clinical factors may also influence a patient's response to aspirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/16493486

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