Jane, this is your
health report

SAMPLE REPORT

Table of Contents

1. Introduction

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

As is common in our studies, on the first pages you will find a summary, with icons, of each of the values analysed, which we present in greater detail in the ensuing pages.

The report is organised into these sections.

1.1. Methodology

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 valuable for prevention and early diagnosis. While not a diagnostic tool, it helps you to see those areas where you need to be more careful.

Applying these studies to your genetic information, we obtain data on your predisposition relative to the rest of the population. At no time does it mean that you are going to suffer any particular disease. Rather, it only indicates that, statistically, and according to this study, you could have a greater propensity than the average person. We indicate that you have greater predisposition when it is greater than 90% of the population's, and smaller if your predisposition is less than 90% of the population's.

It is important to keep in mind that complex diseases are influenced by many factors. Genetics are only a part of it. Lifestyle and diet, food, for example, are in many cases the most important factors.

Genetic Health Risk: Mutations

In this section we analyse the mutations of the most important genes from an 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 analyse 700,000 of the 3.2 billion genetic links. In cases where no mutation is found, this does not mean that one is not a carrier, as it may be in genetic regions that we are not analysing. In this section we analyse a small percentage of the genes classified as pathogenic in the databases used, so there could be pathogenic mutations in a region that we cannot see in this test.

Carrier Status

Hereditary diseases are likely to be passed on to your offspring. In most cases one can be a carrier and never suffer the disease, but there is a risk that one's offspring will suffer it, under 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 the OMIM and ClinVar.

As in the previous section, we do not analyse all the genetic information related to each disease. Specifically, in this section we were able to analyse, on average, something less than half of the pathogenic markers reported in the databases consulted (ClinVar), so one 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 that analyse the entire gene or genes involved in a given disease, and they are valid for clinical use. If you have a family background related to a disease, we recommend that you see 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 towards 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 90% of the population's, and lower if your predisposition is lower than 90% of the population's. Due to the statistical distribution of this analysis, it is normal for several parameters to indicate high or low predispositions.

Pharmacogenomics

In this section we study your genetic predispositions with regards to certain medications. Depending on the drug, your genetics can affect their level of toxicity, effectiveness, or dose needed. This is something that a doctor must always supervise.

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

These reports, as well as the scientific research in the field of Genetics, may vary over time. New mutations are constantly being discovered, such that in the future we will better understand the ones we are analysing today. We make a great effort to periodically apply verified scientific discoveries to our reports.

We remind you should consult with a doctor before making any health-related changes. We encourage all our clients to contract a genetic counselling service to ensure a better understanding of this genetic report. This report is not valid for clinical or diagnostic use.

1.2. Frequently Asked 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 produce are based on statistics. You may have genetic predisposition to a particular disease and never develop it. Actually, this is what happens in most cases. Or, conversely, you may not have a predisposition to a disease, and suffer it in the future. Genetic analysis is just one more tool. Doctors and specialised health professionals should carry out any interpretations of the available set of health data.

Should I make drastic changes to my health management based on the data in this test?

Not at all. Any changes you make to your health management should be reviewed and approved by an expert geneticist or medical specialist. If you have any questions about the genetic test, consult with a healthcare expert in genetic diagnosis.

Does it all depend on my genes?

No at all. Your body responds to many different factors. Our genes are certainly an important parameter. Lifestyle, exercise, diet and many other circumstances also affect the body. Knowing yourself well will enable you to treat your body in the most appropriate way. And this is what these genetic reports are all about: more information.

Are all the genes analysed listed in the sections?

We include most of the genes we analyse; in some sections we analyse more genes than we can show, due to a lack of space.

What is this report based on?

This test is based on different genetic studies that have been internationally verified and accepted by the scientific community. There are scientific databases where studies are published when there exists a certain level of consensus. Our genetic tests are carried out by applying these studies to our clients' genotypes. In each section you will see some of the 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 for an inherited disease, does that mean that I will contract 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 whether we have detected it. This technology boasts reliability greater than 99%, but there is no 100% reliability with these types of genotyping technologies. If you have any questions, you should talk to your doctor or geneticist.

If the report reflects that I DO NOT have genetic mutations for an inherited disease, does that mean I will never contract it, for sure?

No. Our test does not analyse all the genetic zones where pathogenic mutations may exist, and we do not analyse deletions, duplications or intergenic zones. We analyse 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 that we do not see. There are diagnostic tests with greater coverage of certain pathologies that are valid for clinical use. If you have any questions, you should talk to your doctor or geneticist.st.

If I am a carrier of a mutation for 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 between 5 and 50 significant genetic mutations in a given person. However, the risk that your offspring will suffer the disease varies greatly depending on the type of inheritance: autosomal dominant, autosomal recessive, multifactorial ... Therefore, you should always see your doctor or geneticist for guidance in this regard.

2. Summary

Genetic Health Risks: Gwas

- Alopecia areata
- Rheumatoid arthritis
- Breast cancer
- Upper aerodigestive tract cancers
- Motion sickness
- Age-related macular degeneration
- Type 1 diabetes
- Type 2 diabetes
- Celiac disease
- Coronary heart disease
- Multiple sclerosis
- Schizophrenia
- Hypothyroidism
- Chronic lymphocytic leukemia
- Diffuse large B cell lymphoma
- Myasthenia gravis
- Neuroblastoma
- Psoriasis
- Wilms tumor
- Intracranial aneurysm
- Chronic bronchitis and chronic obstructive pulmonary disease
- Bladder cancer
- Basal cell carcinoma
- Primary biliary cirrhosis
- Conduct disorder
- Type 1 diabetes nephropathy
- Endometriosis
- 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
- 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: colorrectal and pancreatic cancer
- BARD1: breast cancer
- BRCA2: breast and ovarian cancer
- CDH1: breast and gastric cancer
- CHEK2: breast and colorrectal cancer
- MSH2: Lynch syndrome and colorrectal cancer
- MUTYH: MYH-associated polyposis and colorrectal cancer
- PMS2: Lynch syndrome and colorrectal cancer
- ATM: breast cancer
- BRCA1: breast and ovarian cancer
- BRIP1: breast cancer
- CDKN2A: pancreatic cancer
- MLH1: Lynch syndrome
- MSH6: Lynch syndrome and colorrectal cancer
- PALB2: breast and pancreatic cancer
- PTEN: breast, uterine and colorrectal cancer

- RAD51C: ovarian cancer
- RAD51D: ovarian cancer
- SDHB: gastric cancer
- SMAD4: juvenile polyposis syndrome and colorrectal cancer
- TP53: Li-Fraumeni syndrome, breast cancer and more
- VHL: Von Hippel-Lindau syndrome
- 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
- Achromatopsia 2
- Leukemia, Acute Myeloid
- Adrenoleukodystrophy
- Hypophosphatasia, Adult
- Allan-Herndon-Dudley Syndrome
- Alpha-1-Antitrypsin Deficiency
- Amyloidosis, Hereditary, Transthyretin-Related
- Anemia, Nonspherocytic Hemolytic, Due To G6Pd Deficiency
- Angelman Syndrome
- Antithrombin Iii Deficiency
- Arrhythmogenic Right Ventricular Dysplasia, Familial, 10
- Auriculocondylar Syndrome 1
- Hypophosphatemic Rickets, Autosomal Dominant
- Bardet-Biedl Syndrome 1
- Muscular Dystrophy, Becker Type
- Beta-Thalassemia
- Bloom Syndrome
- Brugada Syndrome 1
- Cardiofaciocutaneous Syndrome 1
- Cardiomyopathy, Dilated, 1S
- Cardiomyopathy, Familial Hypertrophic, 1
- Ceroid Lipofuscinosis, Neuronal, 1
- Ceroid Lipofuscinosis, Neuronal, 7
- Charcot-Marie-Tooth Disease, Type 4C
- Chondrodysplasia Punctata 1, X-Linked Recessive
- Granulomatous Disease, Chronic, X-Linked
- Adrenal Hypoplasia, Congenital
- Night Blindness, Congenital Stationary, Type 1C
- Cornelia De Lange Syndrome 1
- Costello Syndrome
- Cystic Fibrosis
- Danon Disease
- Deafness, Autosomal Recessive 1A
- Deafness, Autosomal Recessive 31
- Deafness, Autosomal Recessive 7
- Deafness, Autosomal Recessive 9
- Mannosidosis, Alpha B, Lysosomal
- Cardiomyopathy, Dilated, 1A
- Dubin-Johnson Syndrome
- Epileptic Encephalopathy, Early Infantile, 2
- Myoclonic Epilepsy Of Lafora
- Erythrocytosis, Familial, 2
- Fabry Disease
- Familial Adenomatous Polyposis 1

- Cardiomyopathy, Familial Hypertrophic, 2
- Thyroid Carcinoma, Familial Medullary
- Nephrotic Syndrome, Type 1
- Glut1 Deficiency Syndrome 1
- Multiple Acyl-Coa Dehydrogenase Deficiency
- Glycogen Storage Disease Ii
- Hermansky-Pudlak Syndrome 3
- Ectodermal Dysplasia 1, Hypohidrotic, X-Linked
- Joubert Syndrome 14
- Joubert Syndrome 3
- Joubert Syndrome 7
- Joubert Syndrome 9
- Leigh Syndrome
- Leukoencephalopathy With Vanishing White Matter
- Loeys-Dietz Syndrome 2
- Maple Syrup Urine Disease
- Maturity-Onset Diabetes Of The Young, Type 3
- Mental Retardation And Microcephaly With Pontine And Cerebellar Hypoplasia
- Methylmalonic Aciduria And Homocystinuria, Cblc Type
- Methylmalonic Aciduria, Cblb Type
- Mucopolysaccharidosis Type Vi
- Mucopolysaccharidosis, Type Iiia
- Mucopolysaccharidosis, Type Iva
- Myopathy, Myofibrillar, 1
- Myopathy Centronuclear
- Cystinosis, Nephropathic
- Niemann-Pick Disease, Type A
- Noonan Syndrome 1
- Familial Mediterranean Fever
- Fanconi Anemia, Complementation Group O
- Gaucher Disease, Type I
- Glutaric Acidemia I
- Glycogen Storage Disease Ia
- Hemophagocytic Lymphohistiocytosis, Familial, 2
- Histiocytosis-Lymphadenopathy Plus Syndrome
- Jervell And Lange-Nielsen Syndrome 1
- Joubert Syndrome 16
- Joubert Syndrome 5
- Joubert Syndrome 8
- Kabuki Syndrome 1
- Leopard Syndrome 1
- Lissencephaly 1
- Long Qt Syndrome 1
- Maturity-Onset Diabetes Of The Young, Type 2
- Meckel Syndrome, Type 3
- Metachromatic Leukodystrophy
- Methylmalonic Aciduria, Cbla Type
- Mitochondrial Complex Iii Deficiency, Nuclear Type 1
- Mucopolysaccharidosis, Type Vii
- Mucopolysaccharidosis, Type Iiib
- Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eye Anomalies), Type A, 1
- Myopathy, Centronuclear, X-Linked
- Nemaline Myopathy 2
- Niemann-Pick Disease, Type C1
- Niemann-Pick Disease, Type B
- Noonan Syndrome-Like Disorder With Or Without Juvenile Myelomonocytic Leukemia

- Noonan Syndrome 4
- Albinism, Oculocutaneous, Type Ib
- Diabetes Mellitus, Permanent Neonatal
- Polymicrogyria, Bilateral Frontoparietal
- Retinitis Pigmentosa
- Sotos Syndrome 1
- Tay-Sachs Disease
- Tuberous Sclerosis 2
- Tyrosinemia, Type I
- Usher Syndrome, Type Id
- Usher Syndrome, Type Iia
- Usher Syndrome, Type Iid
- Acyl-Coa Dehydrogenase, Very Long-Chain, Deficiency Of
- Wilson Disease
- Obesity Due To Melanocortin 4 Receptor Deficiency
- Osteogenesis Imperfecta, Type Iii
- Pitt-Hopkins Syndrome
- Microcephaly 5, Primary, Autosomal Recessive
- Rubinstein-Taybi Syndrome 1
- Supravalvular Aortic Stenosis
- Tuberous Sclerosis 1
- Albinism, Oculocutaneous, Type Ia
- Usher Syndrome, Type I
- Usher Syndrome, Type If
- Usher Syndrome, Type Iic
- Usher Syndrome, Type Iiia
- Weaver Syndrome
- Agammaglobulinemia, X-Linked

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
- Bilirubin levels
- Calcium levels
- Eosinophil counts
- Glycerophospholipid levels
- IgE levels
- Liver enzyme levels
- Monocyte count
- Phospholipid levels (plasma)
- Omega-6 levels
- Red blood cell count
- Serum total protein level
- Thyroid hormone levels
- White blood cell count
- Beta-2 microglubulin plasma levels
- C reactive protein
- Dehydroepiandrosterone sulphate levels
- Glycated hemoglobin levels
- Homocysteine levels
- Liver enzyme levels (gamma-glutamyl transferase)
- Magnesium levels
- Neutrophil count
- Phosphorus levels
- Platelet count
- Serum albumin level
- Sex hormone levels
- Uric acid levels

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.

Biometrics

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

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.

Traits

- Spirometric measure of pulmonary function (Forced vital capacity)
- Menopause (age at onset)
- Smoking behavior

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.

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

- Bupropion

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
- Vincristine
- Fluorouracil, capecitabine, pyrimidineanalogues, tegafur and Neoplasms

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

- Peginterferon Alpha-2b
- Ribavirin
- Tacrolimus

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
- Morphine
- Pentazocine
- Aspirin

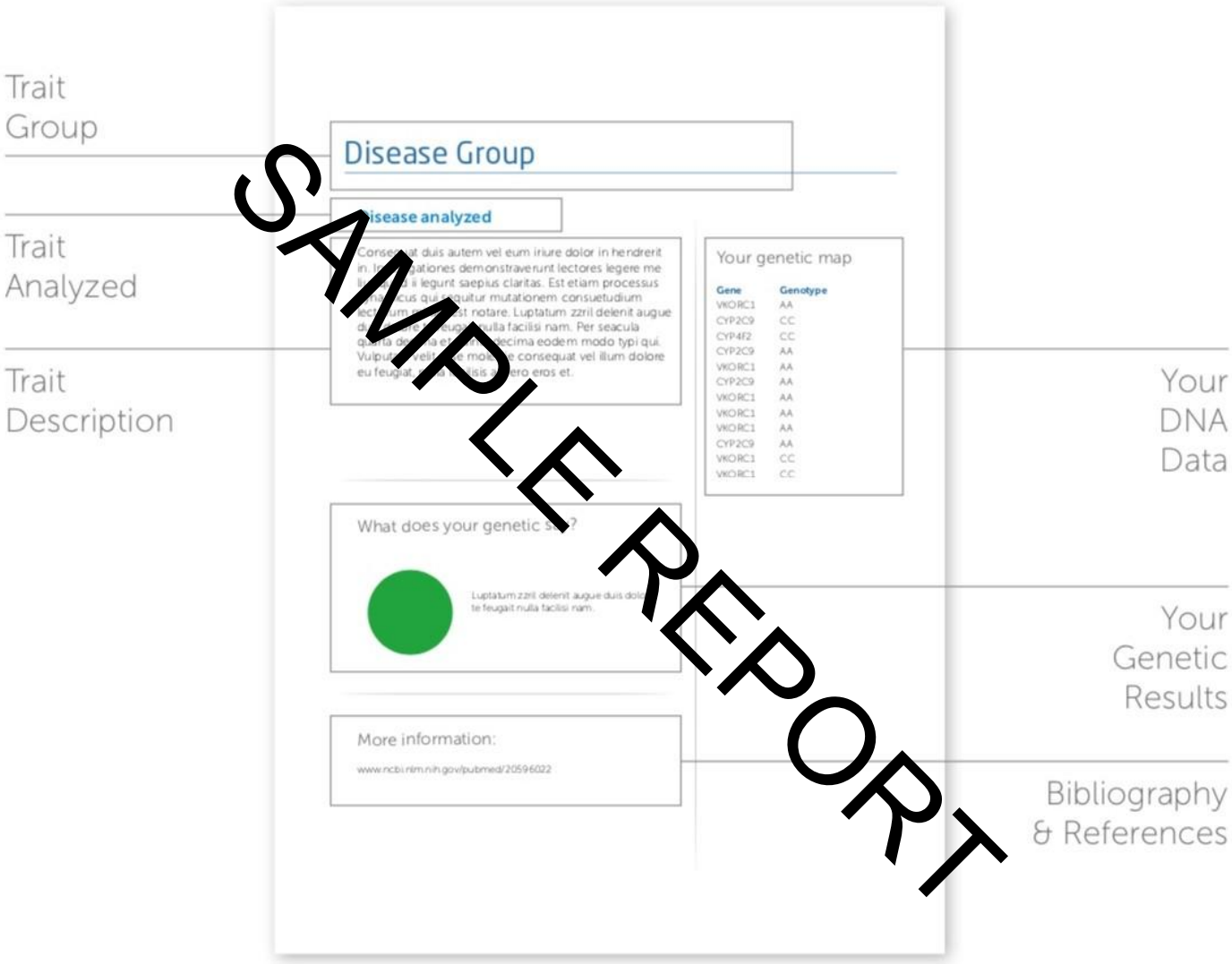
Caption:

- 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.


SAMPLE REPORT

3. Genetic Results

3.1. How to understand your report?



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.

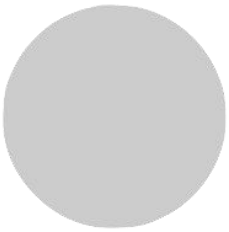
Alopecia areata is thought to be an autoimmune condition. This occurs when the immune system mistakenly attacks and destroys healthy body tissue.

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

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

Intracranial aneurysm

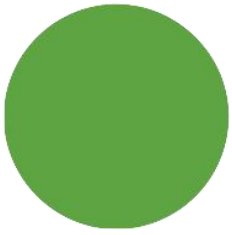
A brain aneurysm is an abnormal bulge or "ballooning" in the wall of an artery in the brain. They are sometimes called "berry aneurysms" because they are often the size of a small berry. Most brain aneurysms produce no symptoms until they become large, begin to leak blood, or burst.

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

Your genetic map

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

What do your genetics tell us?



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/20364137

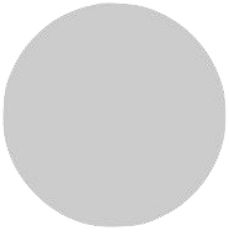
Genetic Health Risks: Gwas

Rheumatoid arthritis

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

More women than men suffer from 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 do your genetics tell us?



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

More information:

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

Your genetic map

Gene	SNP	Genotype
ACOXL	rs6732565	AG
AFF3	rs9653442	TT
ANKRD55	rs7731626	AA
ARID5B	rs71508903	CC
ATG5	rs9372120	TG
BLK	rs2736337	TT
C1QBP	rs72634030	CC
C4orf52	rs11933540	TC
C5orf30	rs2561477	GG
CCL19	rs11574914	AG
CD2	rs624988	CC
CD226	rs2469434	TT
CD28	rs1980422	TC
CD40	rs4239702	TC
CDK6	rs4272	AA
TYR	rs4409785	CC
CASP8	rs6715284	CC
CLNK	rs13142500	TT
CTLA4	rs3087243	AA
ABHD6	rs73081554	CC
EOMES	rs3806624	GG
ETS1	rs73013527	TC
FADS1	rs968567	CC
GRHL2	rs678347	AA
HLA	rs9268839	AG
IL20RB	rs9826828	GG
CSF2 IL3	rs657075	GG
IRAK1	rs5987194	GC
IRF8	rs13330176	TT
JAZF1	rs67250450	TC
LBH	rs10175798	GG

Genetic Health Risks: Gwas

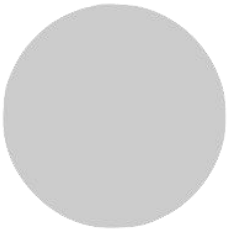
Chronic bronchitis and chronic obstructive pulmonary disease

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 timeMost people with COPD have a combination of both conditions. Smoking is the main cause of COPD. The more a person smokes, the more likely it is that he will develop COPD. However, some people smoke for years and never get COPD. In rare cases, non-smokers 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	TT
EFCAB4A	rs34391416	GG
HHIP AS1	rs13141641	TC
CHRNA3	rs12914385	TC
CYS1	rs12692398	AA

What do your genetics tell us?



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

More information:

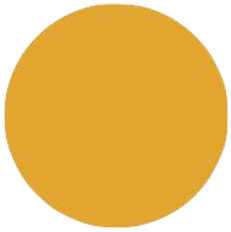
www.ncbi.nlm.nih.gov/pubmed/25241909

Genetic Health Risks: Gwas

Breast cancer

Breast cancer is the most common cancer among women. Common variants at 27 loci have been identified as associated with susceptibility to breast cancer, and these account for ~9% of the familial risk of the disease. We report here a meta-analysis of 9 genome-wide association studies, including 10,052 breast cancer cases and 12,575 controls of European ancestry, from which we selected 29,807 SNPs for further genotyping. These SNPs were genotyped in 45,290 cases and 41,880 controls of European ancestry in 41 studies by the Breast Cancer Association Consortium (BCAC).

What do your genetics tell us?



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/23535729

Your genetic map

Gene	SNP	Genotype
CDCA7	rs1550623	AG
PDE4D	rs1353747	TT
HNF4G	rs2943559	AG
DNAJC1	rs11814448	AA
CHST9	rs1436904	TG
Intergeni	rs11249433	AG
SLC4A7	rs4973768	TT
MAP3K1	rs889312	AC
Intergeni	rs17530068	TC
ESR1	rs3757318	GG
Intergeni	rs13281615	AA
CDKN2A	rs1011970	GG
Intergeni	rs865686	TT
ZNF365	rs10995190	GG
ZMIZ1	rs704010	TC
FGFR2	rs2981579	AG
LSP1	rs3817198	TT
[illegible]THLH	rs10771399	AA
[illegible]51D1	rs999737	CC
TOX3	rs3803662	AG
NRIP1	rs2823093	AG
PEX14	rs616488	AA
METAP1D	rs2016394	GG
DIRC3	rs16857609	TC
ITPR1	rs6762644	GG
TGFBR2	rs12493607	GC
TET2	rs9790517	TC
ADAM29	rs6828523	CC
RAB3C	rs10472076	TT
EBF1	rs1432679	TC
FOXQ1	rs11242675	TT

Genetic Health Risks: Gwas

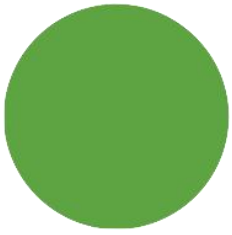
Bladder cancer

Bladder cancer is the fourth most frequently diagnosed in men. It is much more frequent in men than women, the ratio being 7-to-1. The incidence (new cases diagnosed in one year) in our country is the highest in the world: 11% of tumours 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 tumour, suggesting that there is an underlying genetic factor. In fact, large-scale association studies have found genes predisposing one to the disease.

Your genetic map

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

What do your genetics tell us?



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/24163127

Genetic Health Risks: Gwas

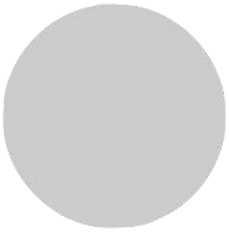
Upper aerodigestive tract cancers

Cancer of the upper aerodigestive tract includes tumours 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 the human papilloma virus infection and family history also play an important role. A large-scale genetic association study has found genetic variants that increase risk of the disease.

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

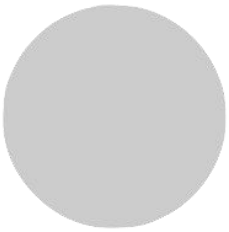
Basal cell carcinoma

Non-melanoma type tumours occur on the outermost layer of the epidermis, and account for some 95% of the cancers that appear on the skin. About 20% are squamous carcinomas, which come from the malignization of the skin's squamous cells. 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 predisposing one 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	TT
TERT	rs401681	CC
KRT5	rs11170164	CC
CDKN2A	rs2151280	AG
KLF14	rs157935	TG
TP53	rs78378222	TT
TGM3	rs214782	AG
RGS22	rs7006527	AA

What do your genetics tell us?



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

More information:

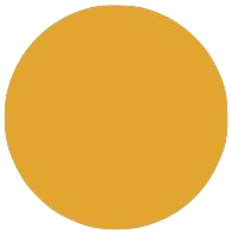
www.ncbi.nlm.nih.gov/pubmed/25855136

Genetic Health Risks: Gwas

Motion sickness

Motion sickness is a common problem in people traveling by car, train, airplanes and boats, especially. Anyone can suffer it, but it is more common in children, pregnant women, and people taking certain medicines. Motion sickness can start suddenly, causing a queasy feeling and cold sweats. It can then lead to dizziness, nausea and vomiting. Your brain senses movement by getting signals from your inner ears, eyes, muscles, and joints. When it receives signals that do not match, you can suffer from 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 its 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 do your genetics tell us?



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/25628336

Your genetic map

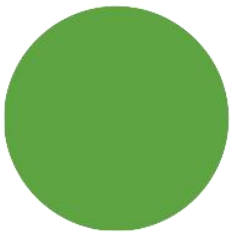
Gene	SNP	Genotype
PVRL3	rs66800491	AG
GPD2	rs56051278	AG
ACO1	rs10970305	AC
AUTS2	rs1195218	GG
GPR26	rs705145	AA
CBLN4	rs6069325	TT
MUTED	rs2153535	GC
LINGO2	rs2150864	AG
CPNE4	rs9834560	AA
RWDD3	rs1858111	AG
PRDM16	rs61759167	TT
NLGN1	rs11713169	AC
HOXD	rs2551802	GG
COPS8	rs2318131	AC
TLE4	rs149951341	AA
HOXB	rs9906289	CC
ST18	rs2360806	AA
SDK1	rs4343996	AG
MEF2	rs7170668	TC
CELF2	rs10752212	AG
CNTN1	rs7957589	AA
MCTP2	rs62018380	CC
ARAP2	rs6833641	CC
AUTS2	rs6946969	AG
RGS5	rs4076764	TT
MAP2K5	rs997295	TT
AGA	rs1378552	CC
POU6F2	rs60464047	AT
TUSC1	rs1782032	AG
GXYLT2	rs1847202	TT

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 facilitates digestion. All of the bile ducts together are called the biliary tract. When the bile ducts become swollen or inflamed, it 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 do your genetics tell us?



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/21399635

Your genetic map

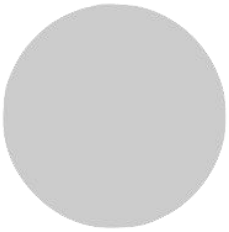
Gene	SNP	Genotype
DENND1	rs12134279	CC
STAT4	rs10931468	CC
CD80	rs2293370	AA
NFKB1	rs7665090	AG
IL7R	rs860413	AA
ELMO1	rs6974491	GG
CXCR5	rs6421571	CC
TNFRSF1	rs1800693	TT
RAD51L1	rs911263	TC
CLEC16A	rs12924729	GG
Intergeni	rs11117432	AG
MAP3K7I	rs968451	GG
IL12A	rs485499	TC
MHC	rs7774434	TC
IRF5	rs12531711	AA
ORMDL3	rs7208487	TG
SPIB	rs3745516	GG
[illegible]	rs1372072	AG
[illegible]	rs538147	GG
TNFAIP2	[illegible]8017161	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 perform tasks such as reading and driving. AMD affects the macula, the part of the eye that allows you to perceive details. 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. You gradually lose your central vision. A common early symptom is that straight lines appear crooked.

What do your genetics tell us?



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

More information:

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

Your genetic map

Gene	SNP	Genotype
ARMS2	rs10490924	GG
CFB C2	rs429608	AG
C3	rs2230199	CG
APOC1	rs4420638	AA
CETP	rs1864163	GG
VEGFA	rs943080	CC
TNFRSF1	rs13278062	TG
LIPC	rs920915	CC
CFI	rs4698775	TT
COL10A1	rs3812111	AT
FILIP1L	rs13081855	GG
IER3	rs3130783	AA
SLC16A8	rs8135665	TC
TGFBR1	rs334353	TT
RAD51B	rs8017304	AG
ADAMTS9	rs6795735	TT
B3GALTL	rs9542236	CC

Genetic Health Risks: Gwas

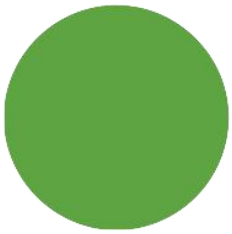
Conduct disorder

Behavioural 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, rule-breaking, the harassment of other children, robberies, violence, etc. This disorder is a risk factor for future addictive behaviour. Different genetic variants have been associated with the risk of onset of this disorder.

Your genetic map

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

What do your genetics tell us?



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/20585324

Genetic Health Risks: Gwas

Type 1 diabetes

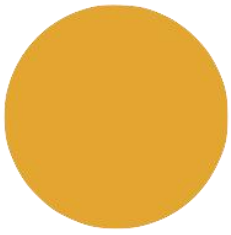
Diabetes means your blood glucose, or blood sugar, levels are too high. With type-1 diabetes, your pancreas does not make insulin. Insulin is a hormone that helps your cells get energy from glucose. Without insulin, too much glucose remains in your blood. Over time, high blood glucose can lead to serious 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	AA
PTPN22	rs6679677	CC
CTLA4	rs3087243	AA
IL2RA	rs12251307	CC
C12orf30	rs17696736	AA
ERBB3	rs2292239	GG
CLEC16A	rs12708716	AA
PTPN2	rs2542151	TT

What do your genetics tell us?



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/18978792

Genetic Health Risks: Gwas

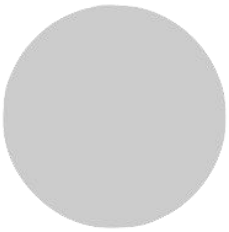
Type 1 diabetes nephropathy

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. 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.

Your genetic map

Gene	SNP	Genotype
MCTP2	rs12437854	TT
AFF3	rs7583877	TT
Intergeni	rs878889	GG
RP11	rs4871297	AA
RNF10	rs614226	CC
Intergeni	rs13045180	TC

What do your genetics tell us?



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

More information:

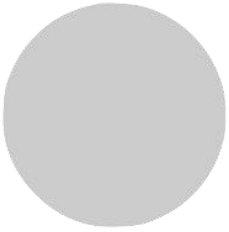
www.ncbi.nlm.nih.gov/pubmed/23028342

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 your cells get energy from glucose. Without insulin, too much glucose remains 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 pre-diabetes also increases your risk. Prediabetes means that your blood sugar is higher than normal, but not high enough to be called diabetes.

What do your genetics tell us?



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

More information:

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

Your genetic map

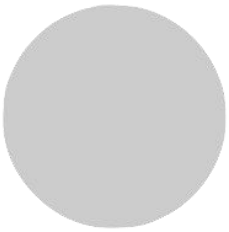
Gene	SNP	Genotype
RREB1	rs9502570	TT
FAF1	rs17106184	GG
TCF19	rs3132524	CC
LPP	rs6808574	TC
ARL15	rs702634	AA
MPHOSP	rs1727313	GG
PLEKHA1	rs10510110	TC
TMEM75	rs1561927	TC
VEGFA	rs9472138	CC
ETV1	rs7795991	AG
C6orf173	rs4273712	AA
TCF7L2	rs7903146	TT
CDKAL1	rs7756992	AG
GRB14	rs3923113	AA
TLE4	rs17791513	AA
CDC123	rs11257655	TC
CENTD2	rs1552224	AC
[illegible]CNQ1	rs163184	GG
[illegible]F1	rs849135	AG
KCNJ11	[illegible]5215	TT
ST64GAL	rs16861329	TC
MTNR1B	rs10830963	CC
HNF4A	rs4812829	AG
HMGA2	rs2261181	CC
SPRY2	rs1359790	AG
AP3S2	rs2028299	AC
FTO	rs9936385	TT
GLIS3	rs7041847	GG
IGF2BP2	rs4402960	TT
PPARG	rs1801282	CC
HNF1B	rs4430796	AG

Genetic Health Risks: Gwas

Endometriosis

The uterus, or womb, is the place where a baby grows when a woman is pregnant. Endometriosis is a disease in which the kind of tissue that normally grows inside the uterus grows outside it. It can grow on the ovaries, fallopian tubes, bowels, or bladder. Rarely, it grows in other parts of the body.

What do your genetics tell us?



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

More information:

www.ncbi.nlm.nih.gov/pubmed/23104006

Your genetic map

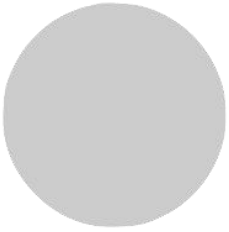
Gene	SNP	Genotype
GREB1	rs13394619	AA
NR	rs7739264	TC
Intergeni	rs12700667	GG
CDKN2B	rs1537377	CC
VEZT	rs10859871	AC

Genetic Health Risks: Gwas

Celiac disease

Celiac disease is an immune disease in which people cannot eat gluten because it damages 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 found 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 may be irritable or depressed. Irritability is one of the most common symptoms in children. Some people have no symptoms.

What do your genetics tell us?



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

More information:

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

Your genetic map

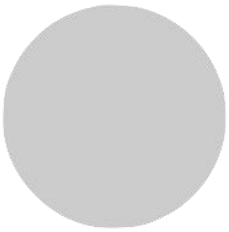
Gene	SNP	Genotype
RGS1	rs2816316	AA
AHSA2	rs13003464	AG
IL18R1	rs917997	CC
ITGA4	rs13010713	GG
ICOS	rs4675374	TC
CCRL2	rs13098911	CC
IL12A	rs17810546	AA
LPP	rs1464510	AC
IL2 IL21	rs13151961	AA
HLA	rs2187668	TT
TNFAIP3	rs2327832	AG
SH2B3	rs653178	CC
PTPN2	rs1893217	AA
MMEL1	rs3748816	AG
RUNX3	rs10903122	AG
Intergeni	rs296547	TC
PLEK	rs17035378	TC
CD80	rs11712165	TG
MAP3K	rs10806425	AC
THEMIS	rs802734	AA
Intergeni	rs9792269	AA
ZMIZ1	rs1250552	AG
ETS1	rs11221332	TC
CLEC16A	rs12928822	CC
ICOSLG	rs4819388	TT
CD247	rs864537	AA
TNFSF18	rs859637	CC
FRMD4B	rs6806528	CC
Intergeni	rs10936599	CC
ELMO1	rs6974491	GG
Intergeni	rs2074404	TT

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 analyses the predisposition to Late-Onset Alzheimer's.

What do your genetics tell us?



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

More information:

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

Your genetic map

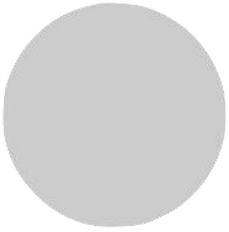
Gene	SNP	Genotype
CR1	rs6656401	GG
BIN1	rs6733839	TC
CD2AP	rs10948363	AG
EPHA1	rs11771145	AG
CLU	rs9331896	TT
MS4A6A	rs983392	AA
PICALM	rs10792832	GG
INPP5D	rs35349669	TC
MEF2C	rs190982	AG
NME8	rs2718058	AG
ZCWPW1	rs1476679	TT
CELF1	rs10838725	TC
FERMT2	rs17125944	TT
CASS4	rs7274581	TT
HLA	rs9271192	AC
PTK2B	rs28834970	TC
SORL1	rs11218343	TT
SLC24A4	rs10498633	TT
SCIMP	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 carry blood and oxygen to your heart.
This buildup causes the arteries to narrow. As a result, blood flow to the heart can slow down or stop.

What do your genetics tell us?



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

More information:

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

Your genetic map

Gene	SNP	Genotype
PCSK9	rs11206510	TC
CXCL12	rs1746048	CC
PPAP2B	rs17114036	AA
ANKS1A	rs17609940	GG
ZC3HC1	rs11556924	TT
ABO	rs579459	TC
CNNM2	rs12413409	GG
ZNF259	rs964184	GC
COL4A1	rs4773144	AA
HHIPL1	rs2895811	TC
ADAMTS7	rs3825807	AG
SMG6	rs216172	GG
RASD1	rs12936587	AG
SNF8 GIP	rs46522	TT
MIA3	rs17465637	AC
WDR12	rs6725887	TT
MRAS	rs2306374	TC
LPA	rs3798220	TT
CDKN2	rs4977574	AG
SH2B3	rs3184504	CC
LDLR	rs1122608	GG
SLC5A3	rs9982601	CC
Intergeni	rs10933436	AC
Intergeni	rs7651039	TC
Intergeni	rs7808424	TT
Intergeni	rs1231206	AG

Genetic Health Risks: Gwas

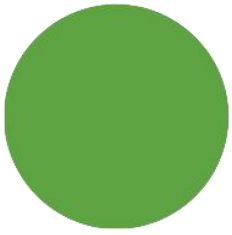
Parkinson's disease

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. Genetics is shedding new light on the disease, with the identification of several genes and markers associated with family forms; although these represent just 5 to 10% of cases, their study is key to the knowledge of the disease.

Your genetic map

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

What do your genetics tell us?



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

More information:

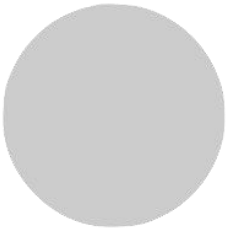
www.ncbi.nlm.nih.gov/pubmed/25064009

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. These can include: visual disturbances, muscle weakness, trouble with coordination and balance, sensations such as numbness, prickling, "pins and needles", and 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 its occurrence, which explains the higher frequency of the disease in the relatives of affected people.

What do your genetics tell us?



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

More information:

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

Your genetic map

Gene	SNP	Genotype
AGAP2	rs12368653	AG
AHI1	rs11154801	CC
BACH2	rs12212193	AG
BATF	rs2300603	TC
C1orf106	rs7522462	AA
CD80	rs2293370	AA
CD5 CD6	rs650258	TC
CD58	rs1335532	AA
CD86	rs9282641	GG
CHST12	rs6952809	TT
CLECL1	rs10466829	GG
CXCR5	rs630923	CC
CYP24A1	rs2248359	TT
DDAH1	rs233100	GG
DKKL1	rs2303759	TG
DLEU1	rs806321	TC
EOMES	rs11129295	TC
EVI5	rs11810217	TT
[illegible]M1	rs12048904	TT
FCRL3	rs3761959	CC
GPR65	rs2119704	CC
HHEX	rs7923837	GG
IL12A	rs2243123	TT
IL12B	rs2546890	AA
IL22RA2	rs17066096	AG
IL7R	rs6897932	CC
IRF8	rs13333054	CC
MALT1	rs7238078	TT
MAMSTR	rs281380	CC
MAPK1	rs2283792	TT
MERTK	rs17174870	CC

Genetic Health Risks: Gwas

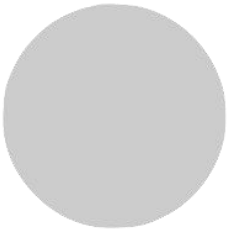
Systemic sclerosis

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 sclerosis; that is, it hardens. It can also affect other organs of the body such as the lungs, heart, kidneys, etc. although the part most often 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 worsen the 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	GG
HLA	rs6457617	CC
RHOB	rs13021401	CC
TNIP1	rs2233287	GG
CD247	rs2056626	TG
STAT4	rs7574865	GG
TNPO3	rs10488631	TC

What do your genetics tell us?



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

More information:

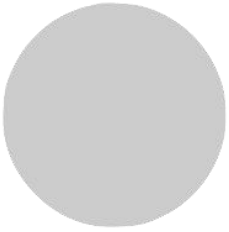
www.ncbi.nlm.nih.gov/pubmed/21750679

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 develop schizophrenia after age 45.

What do your genetics tell us?



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

More information:

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

Your genetic map

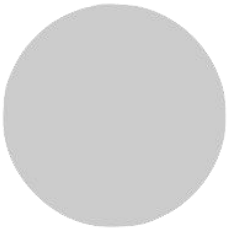
Gene	SNP	Genotype
PLCH2	rs4648845	CC
KDM4A	rs11210892	AA
LRRIQ3	rs12129573	CC
DPYD	rs1702294	CC
FAM5B	rs6670165	CC
C1orf132	rs7523273	AA
AKT3	rs77149735	GG
FANCL	rs11682175	TC
CYP26B1	rs3768644	GG
PCGEM1	rs59979824	CC
SATB2	rs6704641	AA
C2orf82	rs6704768	AA
CNTN4	rs17194490	GG
TRANK1	rs75968099	CC
ATXN7	rs832187	TT
MSL2	rs7432375	GG
C4orf27	rs10520163	TT
GPM6A	rs1106568	AA
LCN1	rs1501357	TC
ZSWIM6	rs4391122	AA
MEF2C	rs16867576	AG
MAN2A1	rs4388249	CC
CDC25C	rs3849046	TC
GALNT10	rs11740474	TT
RIMS1	rs1339227	CC
FUT9	rs117074560	CC
GRM3	rs12704290	GG
MLL5	rs6466055	AA
IMMP2L	rs13240464	TC
PODXL	rs7801375	GG
DGKI	rs3735025	TC

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.

What do your genetics tell us?



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

More information:

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

Your genetic map

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

Genetic Health Risks: Gwas

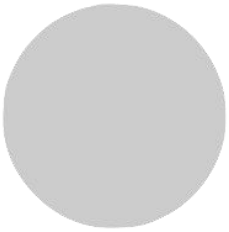
Hypothyroidism

Your thyroid is a butterfly-shaped gland in your neck, just above your collarbone. It is one of your endocrine glands, which produce 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 comprise your body's metabolism. If your thyroid gland is not active enough, it does not produce enough thyroid hormone to meet your body's needs. This condition is known as hypothyroidism. Hypothyroidism is more common in women, people with other thyroid problems, and those over age 60. 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.

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

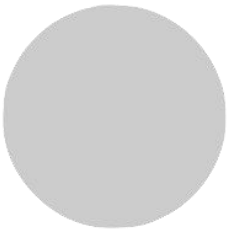
Myocardial infarction (early onset)

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, 5-10% occur in younger patients (men under 50 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 early onset myocardial infarction.

Your genetic map

Gene	SNP	Genotype
CDKN2A	rs4977574	AG
CELSR2	rs646776	TT
MIA3	rs17465637	AC
CXCL12	rs1746048	CC
SLC5A3	rs9982601	CC
WDR12	rs6725887	TT
LDLR	rs1122608	GG
PCSK9	rs11206510	TC

What do your genetics tell us?



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

More information:

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

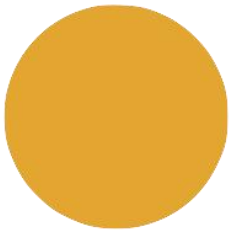
Genetic Health Risks: Gwas

Chronic lymphocytic leukemia

Leucemia is cancer of the white blood cells. White blood cells help your body fight infection. Your blood cells form in your bone marrow. In leucemia, 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 Leucemia (CLL), there are too many lymphocytes, a type of white blood cell.

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

What do your genetics tell us?



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/23770605

Your genetic map

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

Genetic Health Risks: Gwas

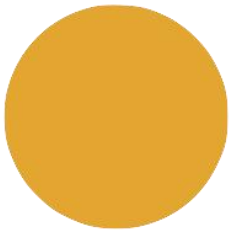
Hodgkin's lymphoma

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 features a bimodal distribution, affecting either the young, ages 15 to 35, or those well over 55. 60-70% of patients are asymptomatic, and cases are usually detected due to an increase in the volume of the lymph nodes. 45-60% of cases are associated with an Epstein-Barr virus infection.

Your genetic map

Gene	SNP	Genotype
EOMES	rs3806624	GG
HBS1L	rs7745098	TT
NR	rs1432295	GG
NR	rs501764	TG
PVT1	rs2019960	TT
NR	rs6903608	TT

What do your genetics tell us?



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/24149102

Genetic Health Risks: Gwas

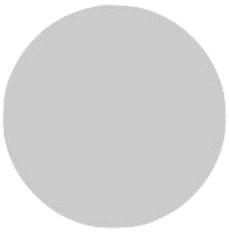
Diffuse large B cell lymphoma

Diffuse Large B-cell Lymphoma (DLBCL) is a clinically aggressive B-cell (immune system) cancer and is the most common non-Hodgkin lymphoma. In some European countries the incidence of non-Hodgkin lymphoma is estimated at 12.3 cases per 100,000/year in men, whereas in women it is 10.8 cases. It is a disease of the elderly, with an average diagnosis age of around 70. Diagnosis in the early stages may improve prognosis. Family history is a risk factor.

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

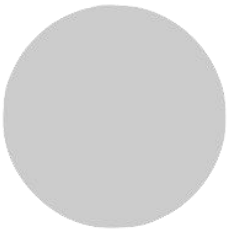
Follicular lymphoma

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

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

Myasthenia gravis

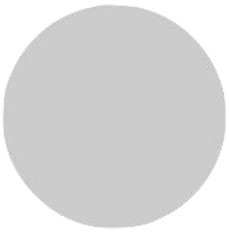
Myasthenia gravis is a disease that causes weakness in the voluntary muscles. These are the muscles that you control. For example, you may suffer weakness in the muscles used for eye movement, facial expressions, and swallowing. You can also have weakness in other muscles. This weakness gets worse with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

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

Your genetic map

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

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

Multiple myeloma

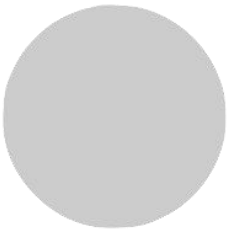
Multiple myeloma is a cancer that begins in plasma cells, a type of white blood cell. These cells are part of your immune system, which helps protect the body from germs and other harmful substances. Over time myeloma cells collect in the bone marrow and in the solid parts of bones.

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	CC
NR	rs11195062	CC
TNFRSF1	rs4273077	AA
CBX7	rs877529	AG

What do your genetics tell us?



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

More information:

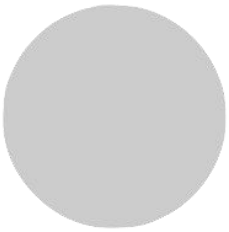
www.ncbi.nlm.nih.gov/pubmed/23955597

Genetic Health Risks: Gwas

Neuroblastoma

Neuroblastoma is a cancer that forms in your nerve tissue. It usually begins in the adrenal glands, located above your kidneys. It may also begin in the 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 do your genetics tell us?



According to this study, you have a propensity similar to that of 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	CC
LINC003	rs9295536	AC
LMO1	rs110419	AG
HSD17B1	rs11037575	TT

Genetic Health Risks: Gwas

Osteosarcoma

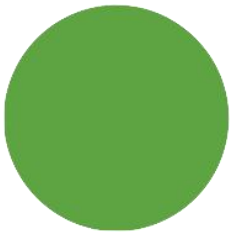
Osteosarcoma is a very rare type of cancerous bone tumour that usually develops in teenagers. It often occurs when a teen is growing rapidly. Osteosarcoma is the most common bone cancer in children. The average age at diagnosis is 15. Boys and girls are just as likely to develop this tumour, until the 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.

Your genetic map

Gene	SNP	Genotype
GRM4	rs1906953	CC
AJ412031	rs573666	CC
Intergeni	rs7591996	AA
ADAMTS6	rs17206779	TT

What do your genetics tell us?



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

More information:

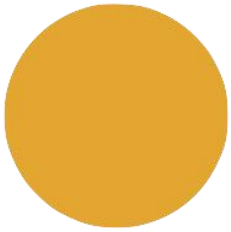
www.ncbi.nlm.nih.gov/pubmed/23727862

Genetic Health Risks: Gwas

Psoriasis

Psoriasis is a skin disease that causes itchy or sore patches of thick, red skin with silvery scales. Patients usually get the patches on their elbows, knees, scalp, back, face, palms and feet, but they can show up on other parts of the 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. This normally takes a month. In cases of psoriasis this happens in just days, because one's 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 do your genetics tell us?



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/25903422

Your genetic map

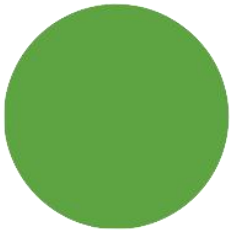
Gene	SNP	Genotype
TP63	rs28512356	AC
COG6	rs34394770	TC
LOC1448	rs9533962	TC
RUNX1	rs8128234	CC
CLIC6	rs9305556	GG
OSTN	rs11922372	TC
IL12B	rs7709212	TT
TNIP	rs17728338	GG
IL12B	rs4921493	TC
IFIH1	rs3747517	TT
LCE	rs4845459	AA
TNFAIP3	rs643177	TC
REL	rs842625	AG
IL12B	rs2853694	GG
PSMA6	rs8016947	TG
NOS2	rs4795067	AG
IL13	rs20541	GG
DDX58	rs11795343	TC
IL23RA	rs10794648	CC
ILF3	rs892085	AG
IL23R	rs12564022	TT
IL23A	rs2066807	GC
TRAF3IP2	rs240993	CC
ETS1	rs6590334	TC
TRAF3IP2	rs7769061	AA

Genetic Health Risks: Gwas

Allergic sensitization

Allergic sensitisation 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 sensitisation 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.

What do your genetics tell us?



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/23817571

Your genetic map

Gene	SNP	Genotype
LRRC32	rs2155219	TG
STAT6	rs1059513	TC
TSLP	rs10056340	TG
HLA	rs6906021	TC
IL18R1	rs3771175	TT
FAM114A	rs17616434	CC
LPP BCL6	rs9865818	AA
MYC	rs4410871	CC
IL2	rs17454584	GG
MICA	rs6932730	TC

Genetic Health Risks: Gwas

Wilms tumor

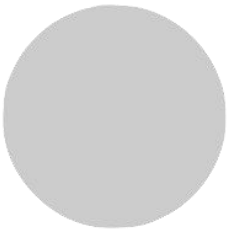
Wilms Tumour is a rare type of kidney cancer. It causes a tumor on one or both kidneys. It usually affects children, but can occur in adults. Having certain genetic conditions, or birth defects, can increase the risk of contracting it. Children that are at risk should be screened for Wilms tumor every 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	TC
DLG2	rs790356	AG
NR	rs2283873	GG
NR	rs5955543	AA
MYCN	rs807624	TG

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: Gwas

Vitiligo

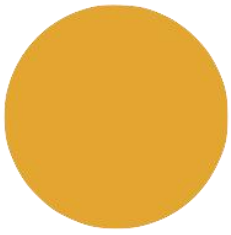
Vitiligo causes white patches on your skin. It can also affect your eyes, mouth, and nose. It occurs when the cells that give your skin its color are destroyed. No one knows what destroys them. It is more common in people with autoimmune diseases, and it might run in families. It usually starts before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

Your genetic map

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

What do your genetics tell us?



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/22561518

Genetic Health Risks: mutations

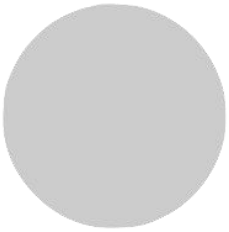
APC: colorrectal and pancreatic cancer

APC gene mutations may be related to diseases such colorrectal and pancreatic cancer. Some publications associate it, in some cases, with gastric cancer.

Your genetic map

Gene	SNP	Genotype
APC	rs387906230	TT
APC	rs121913327	CC
APC	rs398123116	GG
APC	rs587779786	AA

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: mutations

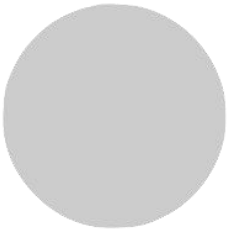
ATM: breast cancer

Mutations of the ATM gene may be related to diseases like breast cancer. Some publications have associated this gene, to a lesser extent, with other cancers, such as ovarian.

Your genetic map

Gene	SNP	Genotype
ATM	rs28904921	TT
ATM	rs55861249	CC
ATM	rs587776551	GG
ATM	rs587779866	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

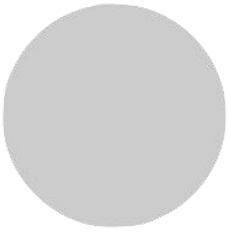
BARD1: breast cancer

BARD1 gene mutations may be related to diseases like breast cancer. Some publications have associated this gene, to a minor extent, with ovarian cancer.

Your genetic map

Gene	SNP	Genotype
BARD1	rs587780021	GG
BARD1	rs587780031	CC
BARD1	rs587781728	AA

What do your genetics tell us?



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More information:

https://www.ncbi.nlm.nih.gov/pubmed/21344236

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 are some studies that associated this gene, to a lesser extent, with other cancers, such as colon and pancreatic.

Your genetic map

Gene	SNP	Genotype
BRCA1	rs62625308	GG
BRCA1	rs28897686	CC
BRCA1	rs80357382	TT
BRCA1	rs80358061	AA

What do your genetics tell us?

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

More information:

Genetic Health Risks: mutations

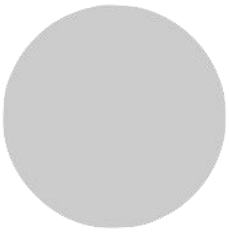
BRCA2: breast and ovarian cancer

Mutations of the BRCA2 gene may be related to diseases such as breast and ovarian cancer. Some studies have related this gene, to a lesser extent, with other cancers, such as pancreatic.

Your genetic map

Gene	SNP	Genotype
BRCA2	rs80359062	CC
BRCA2	rs81002897	GG
BRCA2	rs81002899	TT
BRCA2	rs81002853	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

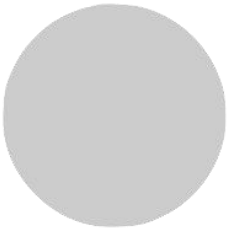
BRIP1: breast cancer

Mutations in the BRIP1 gene may be related to diseases like breast cancer. There are some studies that associated this gene, on a smaller scale, with ovarian cancer.

Your genetic map

Gene	SNP	Genotype
BRIP1	rs587780226	GG
BRIP1	rs587780228	CC
BRIP1	rs587782410	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

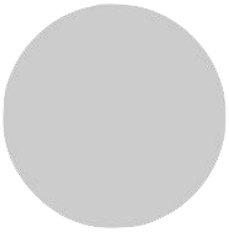
CDH1: breast and gastric cancer

Mutations of the CDH1 gene may be associated with diseases such as breast and gastric cancer. There are some studies linking this gene, to a lesser extent, with ovarian and colon cancer.

Your genetic map

Gene	SNP	Genotype
CDH1	rs587780784	CC
CDH1	rs587780787	GG

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

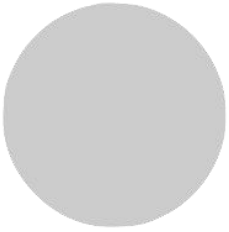
CDKN2A: pancreatic cancer

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

Your genetic map

Gene	SNP	Genotype
CDKN2A	rs104894097	CC

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

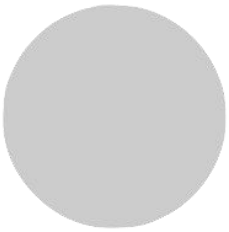
CHEK2: breast and colorrectal cancer

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

Your genetic map

Gene	SNP	Genotype
CHEK2	rs137853007	GG
CHEK2	rs121908698	CC
CHEK2	rs28909982	TT
CHEK2	rs587781705	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

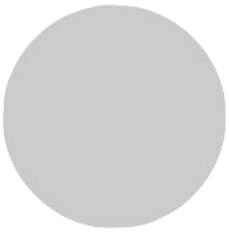
MLH1: Lynch syndrome

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

Your genetic map

Gene	SNP	Genotype
MLH1	rs63750198	CC
MLH1	rs63750710	AA
MLH1	rs63750206	GG
MLH1	rs63749906	TT

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

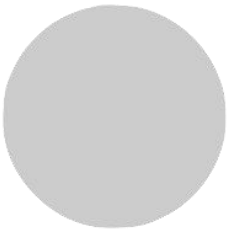
MSH2: Lynch syndrome and colorrectal cancer

MSH2 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Your genetic map

Gene	SNP	Genotype
MSH2	rs28929483	CC
MSH2	rs63750875	GG
MSH2	rs193922376	AA
MSH2	rs63751315	TT

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

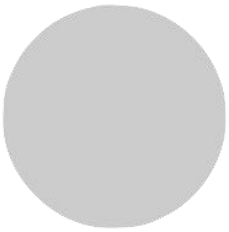
MSH6: Lynch syndrome and colorrectal cancer

MSH6 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Your genetic map

Gene	SNP	Genotype
MSH6	rs397515875	GG
MSH6	rs267608094	CC
MSH6	rs587779208	TT
MSH6	rs267608111	AA

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: mutations

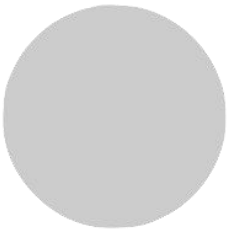
MUTYH: MYH-associated polyposis and colorrectal cancer

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

Your genetic map

Gene	SNP	Genotype
MUTYH	rs34612342	TT
MUTYH	rs36053993	CC
MUTYH	rs121908380	GG
MUTYH	rs730881832	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

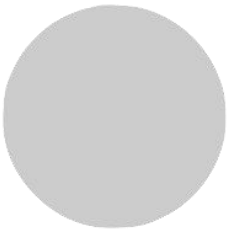
PALB2: breast and pancreatic cancer

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

Your genetic map

Gene	SNP	Genotype
PALB2	rs118203998	GG
PALB2	rs180177103	CC
PALB2	rs730881888	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

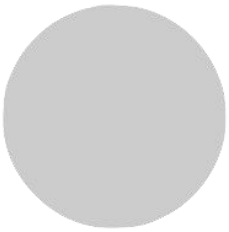
PMS2: Lynch syndrome and colorrectal cancer

PMS2 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Your genetic map

Gene	SNP	Genotype
PMS2	rs63750871	GG
PMS2	rs63750490	TT
PMS2	rs587780059	AA
PMS2	rs587780064	CC

What do your genetics tell us?



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

More information:

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

Genetic Health Risks: mutations

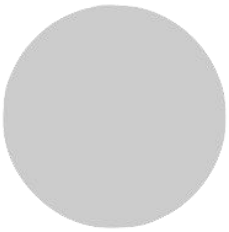
PTEN: breast, uterine and colorrectal cancer

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

Your genetic map

Gene	SNP	Genotype
PTEN	rs121909219	CC
PTEN	rs121909223	TT
PTEN	rs121909229	GG
PTEN	rs121909238	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

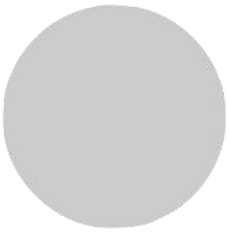
RAD51C: ovarian cancer

RAD51C gene mutations may be related to diseases such as ovarian cancer.

Your genetic map

Gene	SNP	Genotype
RAD51C	rs267606997	GG
RAD51C	rs587780259	AA
RAD51C	rs200293302	CC

What do your genetics tell us?



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More information:

https://www.ncbi.nlm.nih.gov/pubmed/20400964

Genetic Health Risks: mutations

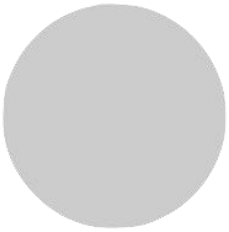
RAD51D: ovarian cancer

RAD51D gene mutations may be related to diseases such as ovarian cancer.

Your genetic map

Gene	SNP	Genotype
RAD51D	rs587780104	GG
RAD51D	rs561425038	TT

What do your genetics tell us?



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More information:

https://www.ncbi.nlm.nih.gov/pubmed/23372765

Genetic Health Risks: mutations

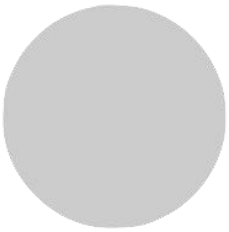
SDHB: gastric cancer

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

Your genetic map

Gene	SNP	Genotype
SDHB	rs74315366	GG
SDHB	rs74315368	CC
SDHB	rs587781270	AA

What do your genetics tell us?



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More information:

Genetic Health Risks: mutations

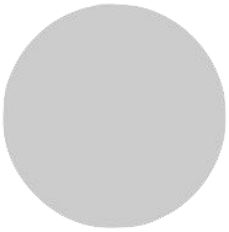
SMAD4: juvenile polyposis syndrome and colorrectal cancer

SMAD4 gene mutations may be related to diseases such as Juvenile Polyposis Syndrome and colorrectal cancer. Some studies have associated this gene, to a lesser extent, with pancreatic cancer.

Your genetic map

Gene	SNP	Genotype
SMAD4	rs80338963	CC
SMAD4	rs281875324	AA

What do your genetics tell us?



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More information:

Genetic Health Risks: mutations

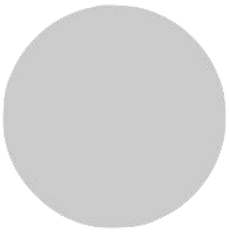
TP53: Li-Fraumeni syndrome, breast cancer and more

TP53 gene mutations may be related to diseases such Li-Fraumeni Syndrome; and breast, ovarian, uterine, colorrectal and pancreatic cancer. There are some studies that have associated this gene, to a lesser extent, with gastric cancer.

Your genetic map

Gene	SNP	Genotype
TP53	rs121912658	TT
TP53	rs121912651	GG
TP53	rs121912652	CC
TP53	rs121912653	AA

What do your genetics tell us?



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More information:

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

Genetic Health Risks: mutations

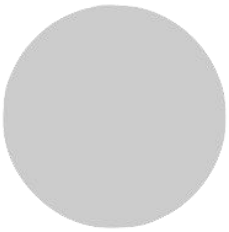
VHL: Von Hippel-Lindau syndrome

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

Your genetic map

Gene	SNP	Genotype
VHL	rs5030821	GG
VHL	rs5030818	CC
VHL	rs5030809	TT
VHL	rs5030804	AA

What do your genetics tell us?



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More information:

Genetic Health Risks: mutations

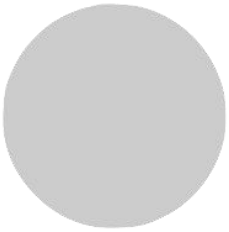
RET: thyroid carcinoma

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

Your genetic map

Gene	SNP	Genotype
RET	rs79781594	GG
RET	rs77316810	TT

What do your genetics tell us?



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More information:

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

Carrier Status

17-Beta Hydroxysteroid Dehydrogenase Iii Deficiency

17-beta-hydroxysteroid dehydrogenase isozyme 3 (17 beta HSD III) deficiency is a rare disorder leading to Male pseudohermaphroditism (MPH), a condition characterised by the incomplete differentiation of the male genitalia in 46, XY males. The estimated incidence of this disease is 1 in 147,000 in The Netherlands. The 17betaHSD III enzyme catalyses the conversion of androstenedione to testosterone in the testis. A 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, characterised by clitoromegaly, posterior labioscrotal fusion, and perineal blind vaginal pouch. Testes are inguinal or in the labioscrotal folds.

Your genetic map

Gene	SNP	Genotype
HSD17B3	rs119481077	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

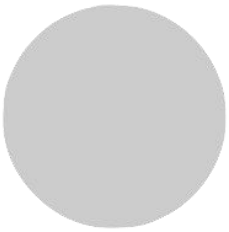
Aarskog-Scott Syndrome

Aarskog-Scott Syndrome (AAS) is a rare developmental disorder characterised by facial, limb and genital features, and a disproportionate acromelic, short stature. The prevalence of AAS is not known, but fewer than 100 cases have been reported in the literature since the first description in 1970. Prevalence estimates, however, are 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	rs398124162	DD

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Achromatopsia 2

Achromatopsia is characterised by reduced visual acuity, pendular nystagmus, increased sensitivity to light (photophobia), a small central scotoma, eccentric fixation, and reduced or complete loss of colour discrimination. All individuals with achromatopsia (achromats) have impaired color discrimination along all three axes of colour perception 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 across all three types of cones. In rare cases individuals may 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 less severe, generally. Hyperopia is common in achromatopsia.

Your genetic map

Gene	SNP	Genotype
CNGA3	rs104893613	CC
CNGA3	rs104893619	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

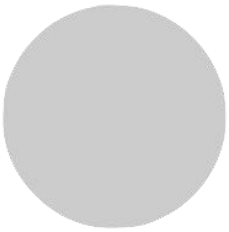
https://www.omim.org/entry/216900

Carrier Status

Leukemia, Acute Myeloid

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
HRAS	rs104894229	CC
TP53	rs28934576	CC
TP53	rs121912651	GG
TP53	rs760043106	AA
HRAS	rs121917759	GG
NRAS	rs121913250	CC
JAK2	rs77375493	GG
PTPN11	rs121918453	GG
IDH2	rs121913502	CC

Carrier Status

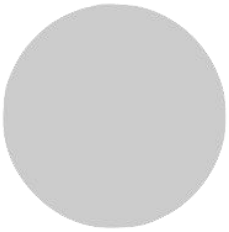
Adrenoleukodystrophy

X-linked Adrenoleukodystrophy (X-ALD) affects nervous system white matter and the adrenal cortex. Three main phenotypes are seen in affected males: the childhood cerebral form manifests most commonly between the ages of four and eight. It initially resembles Attention Deficit Disorder or hyperactivity; progressive impairment of cognition, behaviour, 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 in progressive paraparesis, sphincter disturbances, sexual dysfunction, and often impaired adrenocortical function; all the symptoms are progressive over decades. "Addison Disease only" presents with primary adrenocortical insufficiency between age two and adulthood, and most commonly by age 7.5, without evidence of neurologic abnormality. Approximately 20% of females who are carriers develop neurologic manifestations that resemble AMN, but have later onset (age ≥35) and a milder disease than affected males.

Your genetic map

Gene	SNP	Genotype
ABCD1	rs387906494	II
ABCD1	rs193922093	DD
ABCD1	rs128624218	GG
ABCD1	rs128624220	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

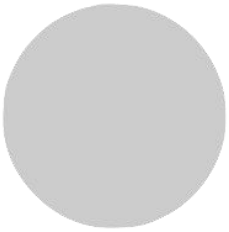
Hypophosphatasia, Adult

Hypophosphatasia (HPP) is a rare, heritable metabolic disorder characterised by the defective mineralisation of bone and/or teeth in the presence of reduced unfractionated serum alkaline phosphatase (ALP) activity. 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).

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

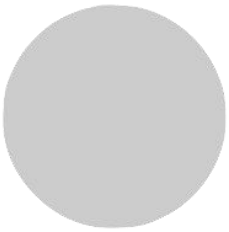
Allan-Herndon-Dudley Syndrome

Allan-Herndon-Dudley Syndrome (AHDS) is an X-linked intellectual disability syndrome with neuromuscular involvement characterised by infantile hypotonia, muscular hypoplasia, spastic paraparesis with dystonic/athetotic 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 aetiology. Only males are affected.

Your genetic map

Gene	SNP	Genotype
SLC16A2	rs387906501	II
SLC16A2	rs587784386	CC
SLC16A2	rs587784383	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

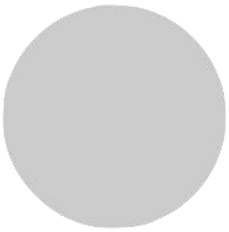
Alpha-1-Antitrypsin Deficiency

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

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

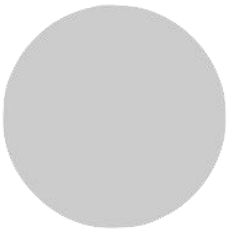
Amyloidosis, Hereditary, Transthyretin-Related

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. Its 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).

Your genetic map

Gene	SNP	Genotype
TTR	rs76992529	GG
TTR	rs386134269	AA
TTR	rs121918076	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

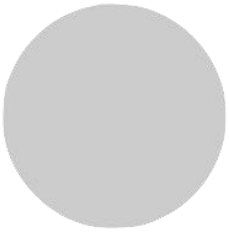
Anemia, Nonspherocytic Hemolytic, Due To G6Pd Deficiency

G6PD deficiency is the most common genetic cause of chronic and drug-, food-, or infection-induced hemolytic anemia. G6PD catalyses the first reaction in the pentose phosphate pathway, which is the only NADPH-generation process in mature red cells; therefore, defence 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 haemolysis is characterised 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 yielded evidence that G6PD deficiency confers resistance against malaria.

Your genetic map

Gene	SNP	Genotype
G6PD	rs5030868	GG
G6PD	rs137852331	TT
G6PD	rs72554665	CC
G6PD	rs76723693	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

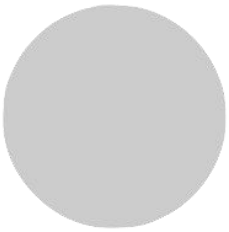
https://www.omim.org/entry/300908

Carrier Status

Angelman Syndrome

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
UBE3A	rs587780570	II
UBE3A	rs587781204	DD
UBE3A	rs111033595	CC
UBE3A	rs587780577	AA
UBE3A	rs587781241	GG
UBE3A	rs587782919	TT
MECP2	rs28935468	GG

Carrier Status

Antithrombin Iii Deficiency

Antithrombin III deficiency 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. Most 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 further classified. Type-1 (low functional and immunologic antithrombin) has been subdivided into subtype 1a (reduced levels of normal antithrombin), and type 1b (reduced levels of antithrombin and the presence of low levels of a variant).

Your genetic map

Gene	SNP	Genotype
SERPINC1	rs28929469	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

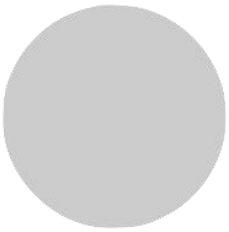
Arrhythmogenic Right Ventricular Dysplasia, Familial, 10

Familial Isolated Arrhythmogenic Right Ventricular Dysplasia (ARVC) is the familial autosomal dominant form of ARVC, a heart muscle disease characterised by life-threatening ventricular arrhythmias with Left Bundle Branch Block Configuration (LBBBC), which may manifest with palpitations, ventricular tachycardia, syncope and sudden, fatal attacks. It is due to dystrophy and fibro-fatty replacement of the right ventricular myocardium, which may lead to right ventricular aneurysms.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

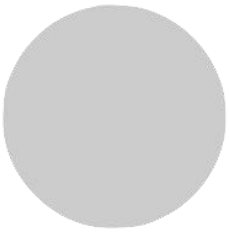
https://www.omim.org/entry/610193

Carrier Status

Auriculocondylar Syndrome 1

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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

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

Carrier Status

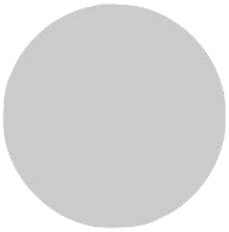
Hypophosphatemic Rickets, Autosomal Dominant

Autosomal Dominant Hypophosphatemic Rickets (ADHR) is a hereditary renal phosphate-wasting disorder characterised 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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Bardet-Biedl Syndrome 1

Bardet-Biedl Syndrome (BBS) is a ciliopathy with multi-system involvement. Its prevalence in Europe is estimated at between 1/125,000 and 1/175,000. This disorder is characterised 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.

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

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

Carrier Status

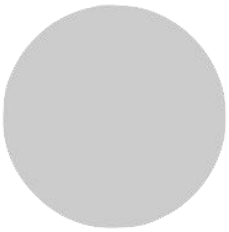
Muscular Dystrophy, Becker Type

Becker Muscular Dystrophy (BMD) is a neuromuscular disease characterised by progressive muscle wasting and weakness due to the 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 Duchenne and Becker Muscular Dystrophy in female carriers; see this term).

Your genetic map

Gene	SNP	Genotype
DMD	rs398123837	II
DMD	rs398123854	DD
DMD	rs104894787	GG
DMD	rs398123828	CC
DMD	rs72468700	TT
DMD	rs398123993	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

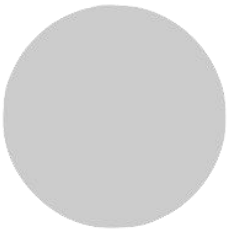
Beta-Thalassemia

Beta-thalassemia (BT) is characterised by deficiency (Beta+) or absence (Beta0) of synthesis of the beta globin chains of haemoglobin (Hb).Its 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 led to global distribution of the disease.

Your genetic map

Gene	SNP	Genotype
HBB	rs35497102	II
HBB	rs33994806	GG
HBB	rs34305195	TT
HBB	rs35703285	AA
HBB	rs33960103	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

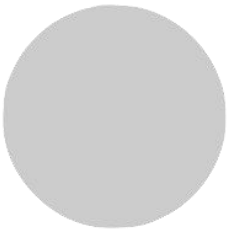
Bloom Syndrome

Bloom Syndrome (BSyn) is a rare chromosomal breakage syndrome characterised 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. Its 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	rs148969222	GG
BLM	rs200389141	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Brugada Syndrome 1

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 East countries indicate a prevalence of 1/700-1/800. Its prevalence in Europe and the United States is lower: 1/3,300 to 1/10,000. An analysis of worldwide literature suggests a prevalence of the Type 1 (diagnostic ECG pattern of 1/1000.

Your genetic map

Gene	SNP	Genotype
SCN5A	rs137854604	GG
SCN5A	rs28937318	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

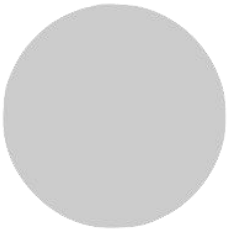
Cardiofaciocutaneous Syndrome 1

Cardiofaciocutaneous (CFC) Syndrome is an RASopathy characterised 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. Its prevalence has been estimated at 1/810,000 people in Japan.

Your genetic map

Gene	SNP	Genotype
BRAF	rs180177039	TT
BRAF	rs180177036	CC
MAP2K2	rs730880517	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

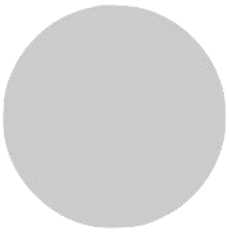
Cardiomyopathy, Dilated, 1S

Familial isolated Dilated Cardiomyopathy (DCM) is a rare, genetically heterogeneous cardiac disease characterised 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
TTN	rs761807131	CC
MYH7	rs121913642	AA
MYH7	rs727503253	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Cardiomyopathy, Familial Hypertrophic, 1

Hypertrophic Cardiomyopathy (HCM) is typically defined by the presence of Left Ventricular Hypertrophy (LVH). SUCH LVH occurs in a non-dilated ventricle in the absence of other cardiac or systemic diseases 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
MYBPC3	rs730880649	DD
MYH7	rs397516155	II
MYBPC3	rs121909374	CC
MYH7	rs121913627	CC
MYH7	rs121913631	GG
MYH7	rs397516161	TT
MYH7	rs727505202	AA
MYBPC3	rs190228518	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

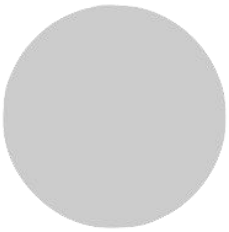
Ceroid Lipofuscinosis, Neuronal, 1

Neuronal Ceroid Lipofuscinoses (NCLs) are a group of inherited progressive degenerative brain diseases characterised clinically by a decline in mental and other capacities, epilepsy, 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.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Ceroid Lipofuscinosis, Neuronal, 7

Neuronal Ceroid-lipofuscinoses (NCLs) are a group of inherited, neurodegenerative, lysosomal storage disorders characterised by progressive intellectual and motor deterioration, seizures, and early death. Visual loss is a feature of most forms. Clinical phenotypes have traditionally been characterised according to the age of onset and the 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, infantile-onset CLN1 disease, and juvenile-onset CLN1 disease are both caused by pathogenic variants in PPT1, but with differing ages of onset. The most prevalent NCLs are classic juvenile CLN3 disease and classic late infantile CLN2 disease (although prevalence varies by ethnicity and country of family origin). The first symptoms typically appear between age two and four.

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed 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

Type 4C Charcot-Marie-Tooth Disease (CMT4C) is a subtype of Type-4 Charcot-Marie-Tooth Disease characterised by childhood or adolescent-onset of a relatively mild, demyelinating sensorimotor neuropathy that contrasts with 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.

Your genetic map

Gene	SNP	Genotype
SH3TC2	rs80338931	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Chondrodysplasia Punctata 1, X-Linked Recessive

Brachytelephalangic Chondrodysplasia Punctata (BCDP) is a form of nonrhizomelic chondrodysplasia punctata, a primary bone dysplasia characterised 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.

Your genetic map

Gene	SNP	Genotype
ARSE	rs145946864	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

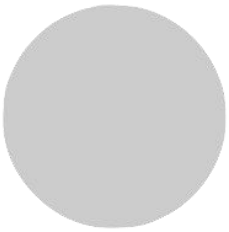
Granulomatous Disease, Chronic, X-Linked

Chronic Granulomatous Disease (CGD) is a rare primary immunodeficiency, mainly affecting phagocytes and characterised 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.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Adrenal Hypoplasia, Congenital

X-linked Adrenal Hypoplasia Congenita (X-linked AHC) is characterised by infantile-onset, acute primary adrenal insufficiency at an average age of three weeks in approximately 60% of affected individuals. Onset in approximately 40% of cases occurs 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 hyperkalaemia, acidosis, hypoglycaemia and shock. Affected males typically have delayed puberty (onset age >14 years) or arrested puberty caused by Hypogonadotropic Hypogonadism (HH).

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
NR0B1	rs386134262	AA
NR0B1	rs386134263	GG

Carrier Status

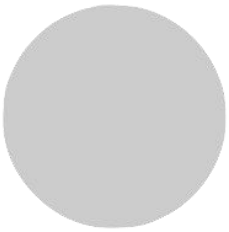
Night Blindness, Congenital Stationary, Type 1C

Congenital Stationary Night Blindness (CSNB) refers to a non-progressive group of retinal disorders characterised by night-time or dim light vision disturbance, delayed adaptation to the dark, poor visual acuity, nystagmus, strabismus, normal colour vision and fundus abnormalities. Two forms of CSNB are recognised: complete and incomplete CSNB (CSNB1 and CSNB2, respectively).

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

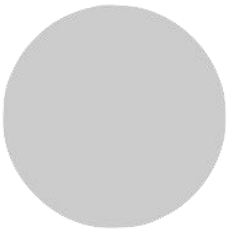
https://www.omim.org/entry/613216

Carrier Status

Cornelia De Lange Syndrome 1

Cornelia de Lange Syndrome (CdLS) is a multi-system 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
NIPBL	rs80358382	II
NIPBL	rs80358371	DD
NIPBL	rs121918267	CC
NIPBL	rs398124470	TT
NIPBL	rs80358380	GG
NIPBL	rs80358373	AA

Carrier Status

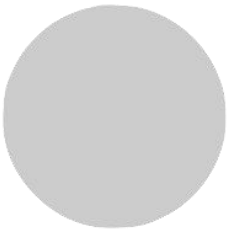
Costello Syndrome

Costello Syndrome (CS) is a rare multi-systemic disorder characterised 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 tumours. 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	rs104894227	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

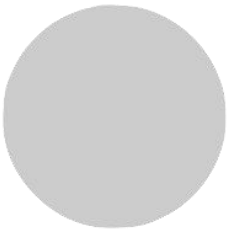
Cystic Fibrosis

Cystic Fibrosis (CF) is a genetic disorder characterised by the production of sweat with 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 variations within each country. Its exact prevalence in Europe is unknown but estimates range between 1/8,000 and 1/10,000 individuals.

Your genetic map

Gene	SNP	Genotype
CFTR	rs121908788	DD
CFTR	rs121908811	II
CFTR	rs75541969	GG
CFTR	rs77101217	CC
CFTR	rs387906362	AA
CFTR	rs193922500	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

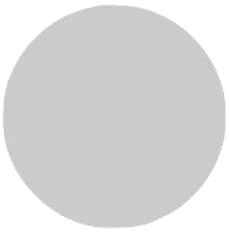
Danon Disease

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 thus far.

Your genetic map

Gene	SNP	Genotype
LAMP2	rs727504557	II
LAMP2	rs397516743	TT
LAMP2	rs727504742	CC
LAMP2	rs727503118	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

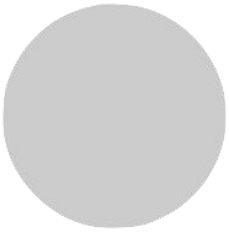
Deafness, Autosomal Recessive 1A

(DFNB1) is characterised by congenital, non-progressive, mild-to-profound sensorineural hearing impairment. No other associated medical findings are present.

Your genetic map

Gene	SNP	Genotype
GJB2	rs80338943	II
GJB2	rs104894413	CC
GJB2	rs111033296	GG
GJB2	rs772264564	AA
GJB2	rs111033294	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Deafness, Autosomal Recessive 31

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 siblings 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.

Your genetic map

Gene	SNP	Genotype
WHRN	rs779760634	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Deafness, Autosomal Recessive 7

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 characterised 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 non-progressive and impedes oral language acquisition.

Your genetic map

Gene	SNP	Genotype
TMC1	rs121908073	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Deafness, Autosomal Recessive 9

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 characterised 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. Initially, language development is not significantly delayed.

Your genetic map

Gene	SNP	Genotype
OTOF	rs80356591	II
OTOF	rs80356590	GG
OTOF	rs111033373	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Mannosidosis, Alpha B, Lysosomal

Alpha-mannosidosis is an inherited lysosomal storage disorder characterised 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 do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

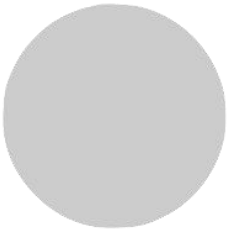
Cardiomyopathy, Dilated, 1A

Non-syndromic isolated Dilated Cardiomyopathy (DCM) is characterised 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	rs28933093	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

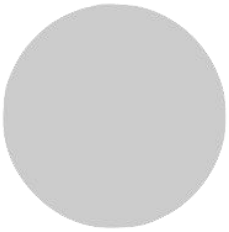
Dubin-Johnson Syndrome

Dubin-Johnson Syndrome (DJS) is a benign, inherited liver disorder characterised clinically by chronic, predominantly conjugated, hyperbilirubinemia; and, histopathologically, by black-brown pigment deposition in parenchymal liver cells. Its 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	rs146405172	GG
ABCC2	rs17222547	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

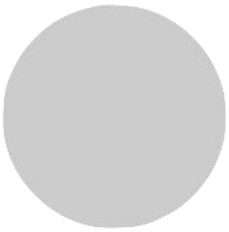
Epileptic Encephalopathy, Early Infantile, 2

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

Your genetic map

Gene	SNP	Genotype
CDKL5	rs61753251	II
CDKL5	rs267608420	DD
CDKL5	rs62653623	CC
CDKL5	rs267608500	AA
CDKL5	rs587783399	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

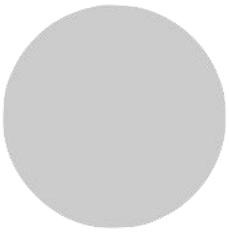
Myoclonic Epilepsy Of Lafora

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

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Erythrocytosis, Familial, 2

Familial erythrocytosis-2 is an autosomal recessive disorder characterised 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.

Your genetic map

Gene	SNP	Genotype
VHL	rs104893826	GG
VHL	rs5030818	CC
VHL	rs5030809	TT

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

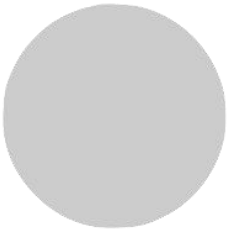
Fabry Disease

Fabry Disease (FD) is a progressive, inherited, multi-systemic lysosomal storage disease characterised 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.

Your genetic map

Gene	SNP	Genotype
GLA	rs398123214	II
GLA	rs104894828	CC
GLA	rs727503950	AA
GLA	rs104894827	GG
GLA	rs104894835	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

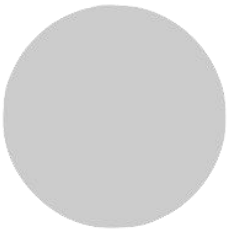
Familial Adenomatous Polyposis 1

Familial Adenomatous Polyposis (FAP) is characterised 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.

Your genetic map

Gene	SNP	Genotype
APC	rs397515732	II
APC	rs137854568	CC
APC	rs387906230	TT
APC	rs559510809	GG
APC	rs587779786	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Cardiomyopathy, Familial Hypertrophic, 2

Hypertrophic Cardiomyopathy (HCM) is typically defined by the presence of Left Ventricular Hypertrophy (LVH). SUCH LVH occurs in a non-dilated ventricle in the absence of other cardiac or systemic diseases 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	rs397516470	II
TNNT2	rs397516463	GG
TNNT2	rs111377893	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

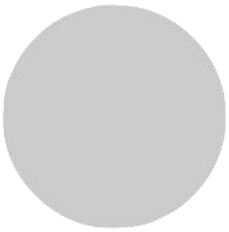
Familial Mediterranean Fever

Familial Mediterranean Fever (FMF) is an autoinflammatory disorder characterised 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.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

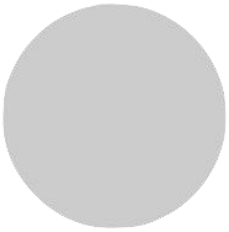
Thyroid Carcinoma, Familial Medullary

Type-2 Multiple Endocrine Neoplasia (MEN2) is a multiple endocrine neoplasia, a polyglandular cancer syndrome characterised by the occurrence of Medullary Thyroid Carcinoma (MTC), Pheochromocytoma (PCC; see these terms) and, 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%.

Your genetic map

Gene	SNP	Genotype
RET	rs75234356	TT
RET	rs77503355	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

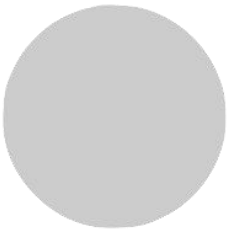
Fanconi Anemia, Complementation Group O

Fanconi Anemia (FA) is a hereditary DNA repair disorder characterised by progressive pancytopenia with bone marrow failure, variable congenital malformations, and a predisposition to develop haematological or solid tumours.

Your genetic map

Gene	SNP	Genotype
RAD51C	rs779582317	AA
RAD51C	rs587782036	GG
RAD51C	rs587782818	CC
RAD51C	rs730881931	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Nephrotic Syndrome, Type 1

Finnish-type Congenital Nephrotic Syndrome 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.

Your genetic map

Gene	SNP	Genotype
NPHS1	rs386833895	CC
NPHS1	rs386833909	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Gaucher Disease, Type I

Gaucher Disease Type 1 is the chronic, non-neurological form of Gaucher Disease (GD; see this term) characterised 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.

Your genetic map

Gene	SNP	Genotype
GBA	rs80356772	CC
GBA	rs364897	TT

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

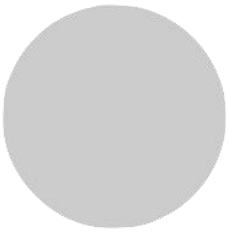
Glut1 Deficiency Syndrome 1

Glucose Transporter (GLUT1) Type-1 deficiency syndrome is characterised by an encephalopathy marked by childhood epilepsy that is refractory to treatment; the 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 gestation and birth.

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs587784391	II
SLC2A1	rs587784397	GG
SLC2A1	rs587784390	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

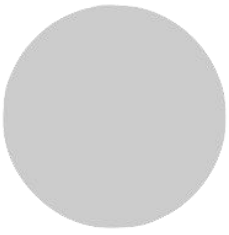
Glutaric Acidemia I

Glutaryl-CoA Dehydrogenase (GCDH) deficiency (GDD) is an autosomal recessive neurometabolic disorder clinically characterised 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 Old Order Amish communities, Canadian Oji-Cree natives, Irish travellers, and Lumbee Native Americans.

Your genetic map

Gene	SNP	Genotype
GCDH	rs121434369	CC
GCDH	rs121434366	TT
GCDH	rs199999619	AA
GCDH	rs121434371	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Multiple Acyl-Coa Dehydrogenase Deficiency

Multiple acyl-CoA Dehydrogenation Deficiency (MADD) is a disorder of fatty acid and amino acid oxidation, and 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	rs398124153	II
ETFDH	rs377686388	TT
ETFDH	rs398124152	CC
ETFDH	rs398124151	GG
ETFA	rs727503918	AA

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Glycogen Storage Disease Ia

Glycogen Storage Disease (GSDI) Type 1 is characterised by the 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 hypoglycaemia; more commonly, however, untreated infants present at age three to four months with hepatomegaly, lactic acidosis, hyperuricemia, hyperlipidemia, hypertriglyceridemia, and/or hypoglycaemic seizures. Affected children typically have doll-like faces with fat cheeks, relatively thin extremities, short stature, and a protuberant abdomen. Xanthoma and diarrhoea may also be present. Impaired platelet function can lead to a bleeding tendency, with frequent epistaxis. Normal growth and puberty is expected in treated children. Most individuals affected live into adulthood.

Your genetic map

Gene	SNP	Genotype
G6PC	rs104894566	TT
G6PC	rs80356484	GG
G6PC	rs104894563	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

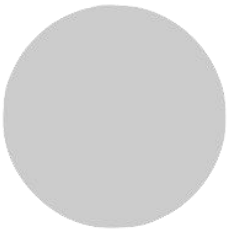
Glycogen Storage Disease Ii

Glycogen Storage Disease due to Acid Maltase Deficiency (AMD) is an autosomal recessive trait leading to metabolic myopathy, affecting 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.

Your genetic map

Gene	SNP	Genotype
GAA	rs28937909	GG
GAA	rs121907938	CC
GAA	rs386834236	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Hemophagocytic Lymphohistiocytosis, Familial, 2

Familial Hemophagocytic Lymphohistiocytosis (FHL) is characterised 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.

Your genetic map

Gene	SNP	Genotype
PRF1	rs28933973	GG
PRF1	rs751161742	TT

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Hermansky-Pudlak Syndrome 3

Hermansky-Pudlak Syndrome (HPS) is a multi-system disorder characterised by tyrosinase-positive oculocutaneous albinism; a bleeding diathesis, resulting from a platelet storage pool deficiency; and, in some cases, pulmonary fibrosis, granulomatous colitis, and immunodeficiency. The albinism is characterised by hypopigmentation of the skin and hair; ocular findings of reduced iris pigment, with iris transillumination; reduced retinal pigment, foveal hypoplasia, with a significant reduction in visual acuity (usually in the range of 20/50 to 20/400); nystagmus; and increased crossing of the optic nerve fibres. Hair colour ranges from white to brown; skin colour 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 haemorrhage, colonic bleeding, and prolonged bleeding with menses, or after tooth extraction, circumcision, and other surgeries.

Your genetic map

Gene	SNP	Genotype
HPS3	rs201227603	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Histiocytosis-Lymphadenopathy Plus Syndrome

Rosaï-Dorfman Disease is a rare benign non-Langerhans cell histiocytosis characterised by the development of large, painless histiocytic masses in the lymph nodes, predominantly in 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.

Your genetic map

Gene	SNP	Genotype
SLC29A3	rs121912583	GG
SLC29A3	rs587780462	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

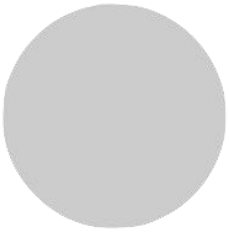
Ectodermal Dysplasia 1, Hypohidrotic, X-Linked

Hypohidrotic Ectodermal Dysplasia (HED) is characterised by hypotrichosis (sparseness of scalp and body hair), 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 characterised by mild manifestations of any or all the characteristic features.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

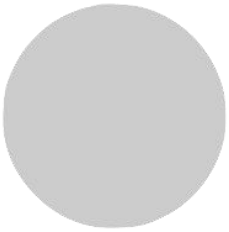
Jervell And Lange-Nielsen Syndrome 1

Jervell and Lange-Nielsen Syndrome (JLNS) is an autosomal recessive variant of familial long QT syndrome (see this term) characterised by congenital, profound, bilateral, sensorineural hearing loss, a long QT interval on electrocardiogram, and ventricular tachyarrhythmias. The disease is very rare. Its 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
KCNQ1	rs397508117	II
KCNE1	rs74315445	CC
KCNQ1	rs120074190	GG
KCNQ1	rs120074189	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

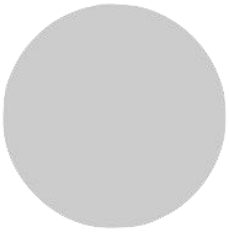
Joubert Syndrome 14

Autosomal recessive development disorder is characterised by severe mental retardation, cerebellar vermis hypoplasia, hypotonia, abnormal breathing patterns 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

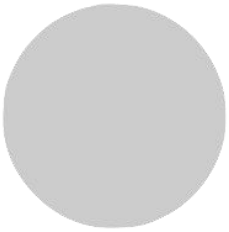
Joubert Syndrome 16

Autosomal recessive development disorder characterised by the Molar Tooth Sign in cerebral images, oculomotor apraxia, variable coloboma, and rare renal involvement.

Your genetic map

Gene	SNP	Genotype
TMEM138	rs387907133	CC
TMEM138	rs387907132	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

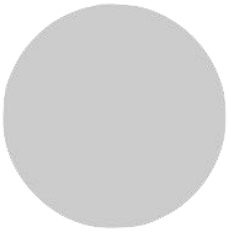
Joubert Syndrome 3

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	rs777668842	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Joubert Syndrome 5

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

Your genetic map

Gene	SNP	Genotype
CEP290	rs727503853	II
CEP290	rs137852834	TT
CEP290	rs370119681	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

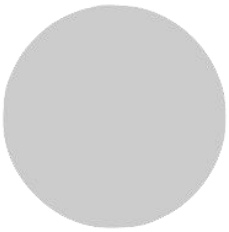
Joubert Syndrome 7

Joubert Syndrome is a clinical and genetically heterogeneous group of disorders characterised by cerebellar vermis hypoplasia, with the characteristic neuroradiological Molar Tooth Sign 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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

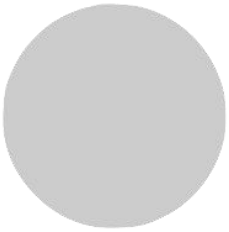
Joubert Syndrome 8

It is characterised by congenital malformation of the brain stem and agenesis or hypoplasia of the cerebellar vermis, which 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

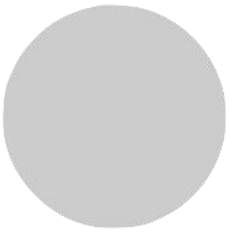
Joubert Syndrome 9

Joubert Syndrome is a clinical and genetically heterogeneous group of disorders characterised by cerebellar vermis hypoplasia, with the characteristic neuroradiological Molar Tooth Sign 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	rs200407856	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Kabuki Syndrome 1

Kabuki Syndrome (KS) is a multiple congenital anomaly syndrome characterised 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. Its prevalence estimation is approximately 1:32,000.

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
KMT2D	rs587783704	II
KMT2D	rs398123720	DD
KMT2D	rs267607237	CC
KMT2D	rs587783700	TT
KMT2D	rs587783699	GG

Carrier Status

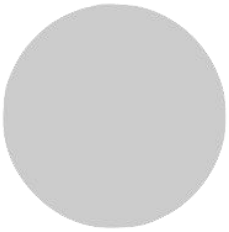
Leigh Syndrome

Leigh Syndrome or subacute necrotizing encephalomyelopathy is a progressive neurological disease defined by specific neuropathological features associated with 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

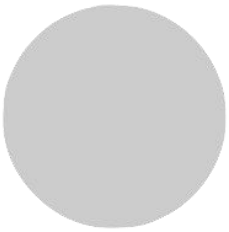
Leopard Syndrome 1

Noonan Syndrome with Multiple Lentigines (NSML), previously known as LEOPARD Syndrome, is a rare, multi-system genetic disorder characterised by lentigines, hypertrophic cardiomyopathy, short stature, pectus deformity, and dysmorphic facial features.

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918457	CC
PTPN11	rs121918468	GG
PTPN11	rs397507548	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

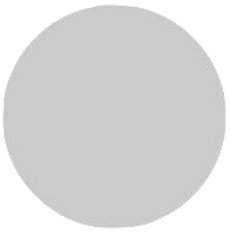
Leukoencephalopathy With Vanishing White Matter

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.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Lissencephaly 1

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 grey matter located just beneath the cortex and separated from it by a thin zone of normal white matter. MDS is characterised by lissencephaly, typical facial features, and severe neurological abnormalities. ILS is characterised by lissencephaly and its direct sequelae: developmental delay, intellectual disability, and seizures.

Your genetic map

Gene	SNP	Genotype
PAFAH1B	rs587784253	II
PAFAH1B	rs587784284	DD
PAFAH1B	rs121434487	GG
PAFAH1B	rs587784260	CC
PAFAH1B	rs587784272	TT
PAFAH1B	rs587784263	AA

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Loeys-Dietz Syndrome 2

Loeys-Dietz Syndrome (LDS) is characterised 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 Type-1 LDS, with craniofacial manifestations (widely spaced eyes, bifid uvula/cleft palate, craniosynostosis); approximately 25% have Type-1 LDS, with systemic manifestations of LDSI, but minimal or absent craniofacial features. LDSI and LDSII form a clinical continuum. The natural history of LDS is characterised by aggressive arterial aneurysms (mean age at death of 26.1) and a high incidence of pregnancy-related complications, including death and uterine rupture

Your genetic map

Gene	SNP	Genotype
TGFBR2	rs104893809	CC
TGFBR2	rs104893816	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

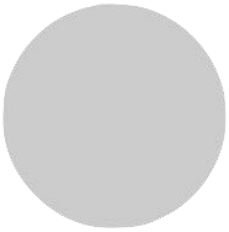
Long Qt Syndrome 1

Congenital Long QT Syndrome (LQTS) is a hereditary cardiac disease characterised by a prolongation of the QT interval at basal ECG and by a high risk of life-threatening arrhythmias. The disease's prevalence is estimated at close to 1 in 2,500 live births.

Your genetic map

Gene	SNP	Genotype
KCNQ1	rs199473457	CC
KCNQ1	rs120074181	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

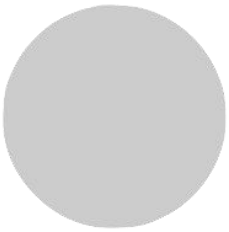
https://www.omim.org/entry/192500

Carrier Status

Maple Syrup Urine Disease

Maple Syrup Urine Disease (MSUD) is a rare inherited disorder of branched-chain amino acid metabolism, classically characterised by poor feeding, lethargy, vomiting and a maple syrup odour 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
BCKDHA	rs398123492	II
DBT	rs398123667	II
BCKDHB	rs398124572	II
BCKDHA	rs137852871	GG
BCKDHA	rs137852875	CC
DBT	rs121964999	AA
BCKDHB	rs386834234	GG
BCKDHA	rs398123509	AA
DBT	rs398123665	CC
DBT	rs398123674	TT
DBT	rs398123675	GG
BCKDHB	rs398124561	CC
BCKDHB	rs398124573	TT
BCKDHB	rs398124577	AA

Carrier Status

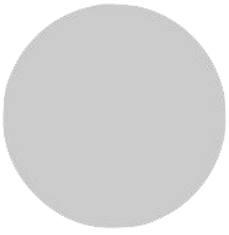
Maturity-Onset Diabetes Of The Young, Type 2

MODY is a form of NIDDM (125853) characterised by monogenic autosomal dominant transmission and early age of onset. For a general phenotypic description and a discussion of the 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 hyperglycaemia 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.

Your genetic map

Gene	SNP	Genotype
GCK	rs193922253	DD
GCK	rs193922295	II
GCK	rs193922331	AA
GCK	rs193922259	TT
GCK	rs193922262	CC
GCK	rs193922263	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

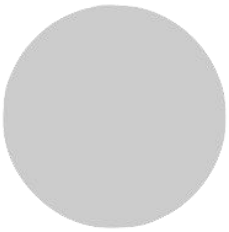
Maturity-Onset Diabetes Of The Young, Type 3

A form of diabetes that is characterised 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.

Your genetic map

Gene	SNP	Genotype
HNF1A	rs386134267	II
HNF1A	rs193922577	TT
HNF1A	rs193922580	CC
HNF1A	rs193922589	AA
HNF1A	rs193922602	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Meckel Syndrome, Type 3

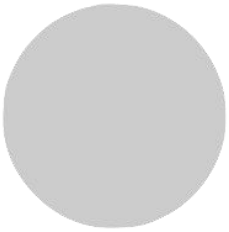
Meckel Syndrome is an autosomal, recessive, pre- or perinatal lethal malformation syndrome characterised by renal cystic dysplasia and variably associated features, including developmental anomalies of the central nervous system (typically occipital encephalocele), hepatic ductal dysplasia and cysts,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

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

Your genetic map

Gene	SNP	Genotype
TMEM67	rs386834182	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Mental Retardation And Microcephaly With Pontine And Cerebellar Hypoplasia

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 has 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.

Your genetic map

Gene	SNP	Genotype
CASK	rs587783362	II
CASK	rs387906705	GG
CASK	rs587783366	TT
CASK	rs587783368	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

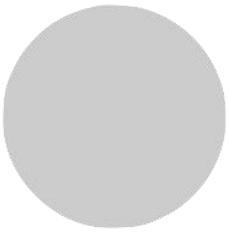
Metachromatic Leukodystrophy

Metachromatic Leukodystrophy (MLD) is a rare lysosomal storage disorder characterised by the intralysosomal accumulation of sulfatides in various tissues, leading to the progressive deterioration of motor and neurocognitive function.

Your genetic map

Gene	SNP	Genotype
ARSA	rs398123414	II
ARSA	rs28940893	GG
ARSA	rs398123419	CC
ARSA	rs74315457	AA
ARSA	rs398123411	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

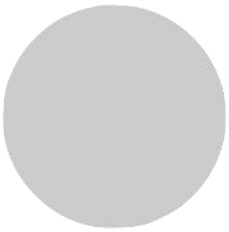
Methylmalonic Aciduria And Homocystinuria, Cblc Type

Methylmalonic acidemia with homocystinuria is an inborn error of Vitamin B12 (cobalamin) metabolism characterised 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	rs398124295	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

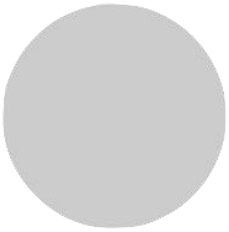
Methylmalonic Aciduria, Cbla Type

Vitamin B12-responsive Methylmalonic Acidemia (MA) is an inborn error of vitamin B12 (cobalamin) metabolism characterised 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. A prevalence of 1/48,000 -1/61,000 has been reported for MA of all causes in North America, and 1/26,000 in China.

Your genetic map

Gene	SNP	Genotype
MMAA	rs104893851	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

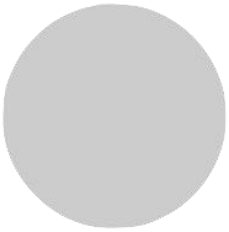
Methylmalonic Aciduria, Cblb Type

Vitamin B12-responsive Methylmalonic Acidemia (MA) is an inborn error of vitamin B12 (cobalamin) metabolism characterised 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. A prevalence of 1/48,000-1/61,000 has 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	rs756414548	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Mitochondrial Complex Iii Deficiency, Nuclear Type 1

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

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

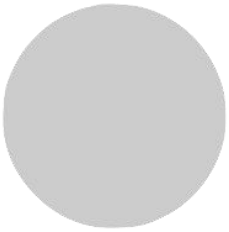
Mucopolysaccharidosis Type Vi

Mucopolysaccharidosis Type-6 (MPS 6) is a lysosomal storage disease with progressive multi-system 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.

Your genetic map

Gene	SNP	Genotype
ARSB	rs201101343	TT
ARSB	rs118203941	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

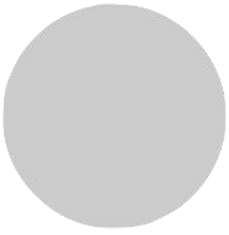
Mucopolysaccharidosis, Type Vii

Type-VII Mucopolysaccharidosis (MPS VII) is a very rare lysosomal storage disease belonging to the group of mucopolysaccharidoses. Fewer 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.

Your genetic map

Gene	SNP	Genotype
GUSB	rs121918173	GG
GUSB	rs398123234	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Mucopolysaccharidosis, Type Iiia

Type-III mucopolysaccharidosis (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 its generally very mild dysmorphism). It is the most frequent MPS in the Netherlands and Australia, with respective prevalences of 1/53,000 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	rs778700037	DD
SGSH	rs104894636	GG
SGSH	rs104894641	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

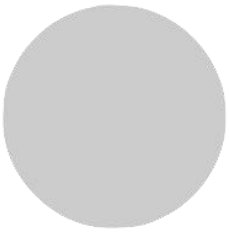
Mucopolysaccharidosis, Type Iiib

Type-III mucopolysaccharidosis (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). 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	rs104894597	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

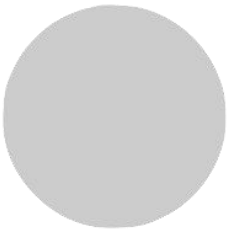
Mucopolysaccharidosis, Type Iva

Type-IV mucopolysaccharidosis (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. Its prevalence is approximately 1/250,000 for type IVA, but its incidence varies widely between countries. MPS IVB is even rarer.

Your genetic map

Gene	SNP	Genotype
GALNS	rs118204438	TT
GALNS	rs746756997	AA
GALNS	rs118204437	GG
GALNS	rs372893383	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

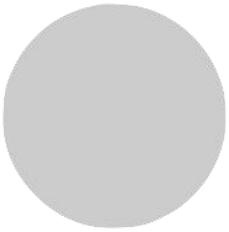
Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eye Anomalies), Type A, 1

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 little muscle tone and poor spontaneous movements. Affected children may present with the delay or arrest of gross motor development, together with joint and/or spinal rigidity. Muscle weakness may improve, worsen, or stabilise in the short term. However, over time progressive weakness and joint contracture, spinal deformities, and compromised breathing may affect quality of life and life span.

Your genetic map

Gene	SNP	Genotype
POMT1	rs398124245	II
POMT1	rs119462982	GG
POMT1	rs149682171	CC
POMT1	rs398124244	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

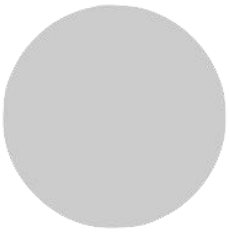
Myopathy, Myofibrillar, 1

Myofibrillar myopathy is characterised by slow, 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	rs267607482	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

Carrier Status

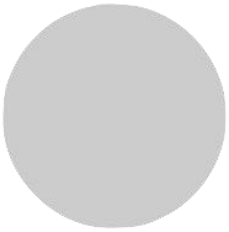
Myopathy, Centronuclear, X-Linked

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

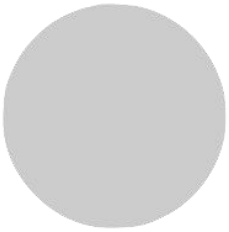
Myopathy Centronuclear

Autosomal dominant centronuclear myopathy is a congenital myopathy characterized by slowly progressive muscle 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 start using wheelschairs in their fifties. Ptosis and limitation of eye movements occur frequently. The most prominent histopathologic features include high frequency of centrally located nuclei in a large number of extrafusal muscle fibers (which is the basis of the name of the disorder), radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. Genetic Heterogeneity of Centronuclear Myopathy Centronuclear myopathy is a genetically heterogeneous disorder.

Your genetic map

Gene	SNP	Genotype
MTM1	rs587783803	II
DNM2	rs121909095	CC
MTM1	rs132630302	AA
MTM1	rs132630305	CC
MTM1	rs587783817	TT
MTM1	rs587783823	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Nemaline Myopathy 2

Nemaline Myopathy (referred to in this entry as NM) is characterised 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 the 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 (AMC) 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]

Your genetic map

Gene	SNP	Genotype
NEB	rs398124167	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

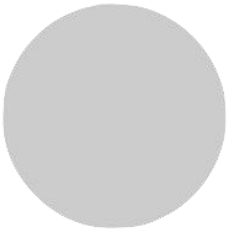
Cystinosis, Nephropathic

Cystinosis is a metabolic disease characterised by an accumulation of cystine inside the lysosomes, causing damage in different organs and tissues, particularly 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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

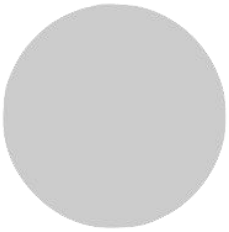
Niemann-Pick Disease, Type C1

Niemann-Pick Disease, Type C (NP-C), is a lysosomal lipid storage disease characterised 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.

Your genetic map

Gene	SNP	Genotype
NPC1	rs398123284	DD
NPC1	rs80358257	GG
NPC1	rs80358252	CC
NPC1	rs372030650	TT
NPC1	rs80358259	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

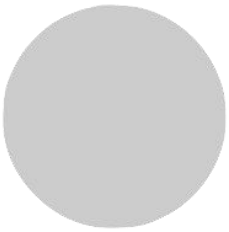
Niemann-Pick Disease, Type A

Type-A Niemann-Pick Disease is a very severe subtype of Niemann-Pick Disease, an autosomal recessive lysosomal disease, and is characterised 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	rs281860677	DD
SMPD1	rs120074122	GG
SMPD1	rs727504166	TT
SMPD1	rs120074128	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

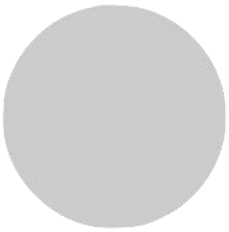
Niemann-Pick Disease, Type B

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

Your genetic map

Gene	SNP	Genotype
SMPD1	rs769904764	CC
SMPD1	rs398123475	TT
SMPD1	rs120074117	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

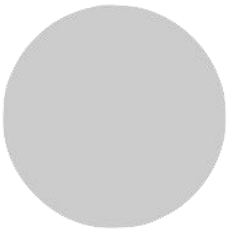
Noonan Syndrome 1

Noonan Syndrome (NS) is characterised by short stature, typical facial dysmorphism, and congenital heart defects. The incidence of NS is estimated to be between 1:1,000 and 1:2,500 live births.

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918463	TT
PTPN11	rs397507509	GG
PTPN11	rs397507529	AA
NRAS	rs267606921	GG
BRAF	rs387906660	GG
PTPN11	rs121918454	CC
NRAS	rs267606920	CC
BRAF	rs606231228	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

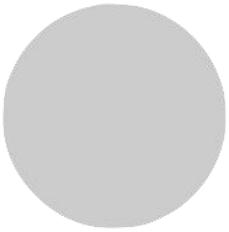
Noonan Syndrome-Like Disorder With Or Without Juvenile Myelomonocytic Leukemia

A syndrome characterised 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 a triangular face, with hypertelorism; large, low-set ears; ptosis, and a flat nasal bridge. Some patients manifest cardiac defects. Some are at increased risk for certain malignancies, particularly juvenile myelomonocytic leucemia.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

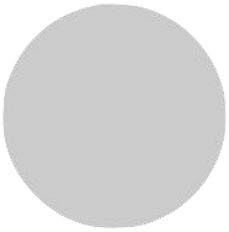
Noonan Syndrome 4

Noonan Syndrome (NS) is characterised by short stature, typical facial dysmorphism, and congenital heart defects. The incidence of NS is estimated to be between 1:1,000 and 1:2,500 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 a webbed neck, chest deformity, mild intellectual deficit, cryptorchidism, poor feeding in infancy, bleeding tendencies, and lymphatic dysplasia.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed 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 most common form of monogenic obesity identified to date. MC4R deficiency is characterised by severe obesity, a decrease 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 2,000. 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	rs52804924	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Albinism, Oculocutaneous, Type Ib

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	rs28940881	AA
TYR	rs61754381	TT

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Osteogenesis Imperfecta, Type Iii

Type-III Osteogenesis Imperfecta is a severe type of osteogenesis imperfecta, a genetic disorder characterised 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, greyish 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	rs768171831	CC
COL1A1	rs72645357	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

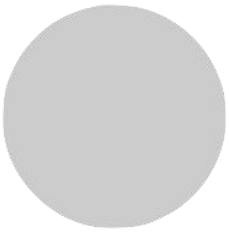
Diabetes Mellitus, Permanent Neonatal

Permanent Neonatal Diabetes Mellitus (PNDM) is a monogenic form of neonatal diabetes characterised by persistent hyperglycaemia 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.

Your genetic map

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

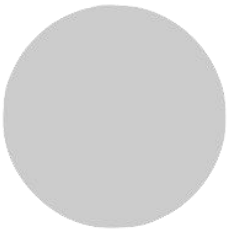
Pitt-Hopkins Syndrome

Pitt-Hopkins Syndrome (PHS) is characterised 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.

Your genetic map

Gene	SNP	Genotype
TCF4	rs587784468	II
TCF4	rs121909123	CC
TCF4	rs727504175	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Polymicrogyria, Bilateral Frontoparietal

Bilateral Frontoparietal Polymicrogyria (BFPP) is a subtype of polymicrogyria, a cerebral cortical malformation characterised by excessive cortical folding and abnormal cortical layering, involving the frontoparietal region of the brain and presenting 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	rs587783660	GG
ADGRG1	rs587783653	TT

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

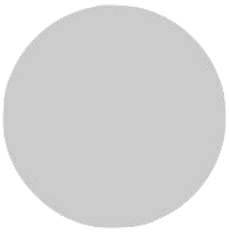
Microcephaly 5, Primary, Autosomal Recessive

Autosomal Recessive Primary Microcephaly (MCPH) is a rare, genetically heterogeneous neurogenic brain development disorder characterised by reduced head circumference at birth, with no gross brain architecture anomalies, and variable degrees of intellectual impairment. The 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.

Your genetic map

Gene	SNP	Genotype
ASPM	rs587783220	II
ASPM	rs759632528	DD
ASPM	rs137852997	AA
ASPM	rs140602858	GG
ASPM	rs587783238	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

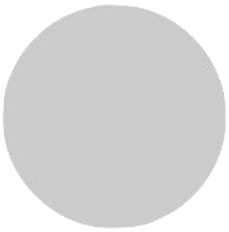
Retinitis Pigmentosa

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. The 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 do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

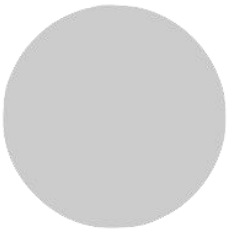
Rubinstein-Taybi Syndrome 1

Rubinstein-Taybi Syndrome is a rare malformation syndrome characterised 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.

Your genetic map

Gene	SNP	Genotype
CREBBP	rs587783508	II
CREBBP	rs587783510	GG
CREBBP	rs587783503	AA
CREBBP	rs587783497	TT
CREBBP	rs587783491	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

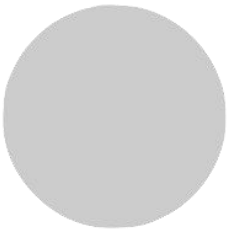
Sotos Syndrome 1

Sotos Syndrome is a rare, multi-systemic genetic disorder characterised by an atypical facial appearance, overgrowth of the body in early life with macrocephaly, and mild to severe intellectual disability.

Your genetic map

Gene	SNP	Genotype
NSD1	rs587784068	II
NSD1	rs587784071	GG
NSD1	rs587784084	CC
NSD1	rs587784111	TT
NSD1	rs587784120	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

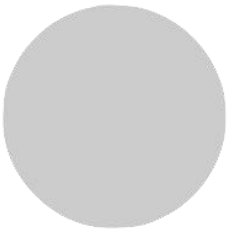
Supravalvular Aortic Stenosis

SupraValvar Aortic Stenosis (SVAS) is characterised 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 cases 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.

Your genetic map

Gene	SNP	Genotype
ELN	rs727503782	II
ELN	rs727503022	DD
ELN	rs727503027	AA
ELN	rs727503029	GG
ELN	rs727503033	TT
ELN	rs137854452	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

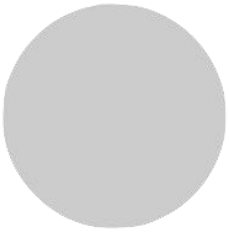
Tay-Sachs Disease

GM2 gangliosidosis, variant B, or Tay-Sachs Disease, is characterised by an 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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

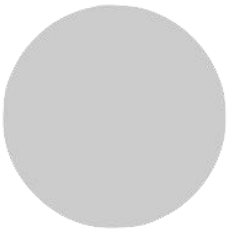
Tuberous Sclerosis 1

Tuberous Sclerosis Complex (TSC) is a neurocutaneous disorder characterised by multi-system hamartomas and associated with neuropsychiatric features. Its prevalence is estimated to be 1/25,000-1/11,300 in Europe.

Your genetic map

Gene	SNP	Genotype
TSC1	rs118203506	II
TSC1	rs118203682	GG
TSC1	rs118203352	TT
TSC1	rs118203423	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

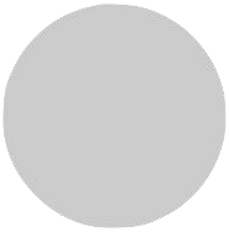
Tuberous Sclerosis 2

Tuberous Sclerosis Complex (TSC) is a neurocutaneous disorder characterised by multi-system hamartomas and associated with neuropsychiatric features. Its prevalence is estimated to be 1/25,000-1/11,300 in Europe. TSC is characterised by multi-system 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 appearing at age 3-4 as erythematous and papulonodular lesions; ungual fibromas; cephalic and lumbar (shagreen patch) fibrous plaques; and "confetti" skin lesions appearing in childhood to early adolescence. The brain is involved in almost all cases of TSC, with the presence of different neuropathological lesions, such as cortico/subcortical tubers, radial migration lines, and subependymal nodules, SEGA. SEGA can cause hydrocephalus (growth risk higher in the first 3 decades).

Your genetic map

Gene	SNP	Genotype
TSC2	rs137854250	II
TSC2	rs45517182	GG
TSC2	rs45451497	CC
TSC2	rs45517096	AA
TSC2	rs137854298	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Albinism, Oculocutaneous, Type Ia

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	rs758115945	GG
TYR	rs151206295	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

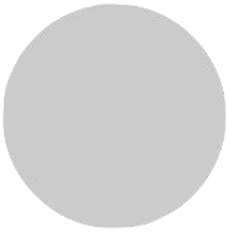
Tyrosinemia, Type I

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

Your genetic map

Gene	SNP	Genotype
FAH	rs11555096	CC
FAH	rs80338901	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

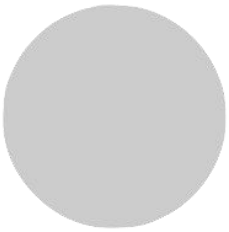
https://www.omim.org/entry/276700

Carrier Status

Usher Syndrome, Type I

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

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Your genetic map

Gene	SNP	Genotype
MYO7A	rs111033510	DD
MYO7A	rs397516294	II
PCDH15	rs397517451	II
MYO7A	rs397516281	TT
MYO7A	rs397516283	GG
MYO7A	rs111033180	CC
MYO7A	rs111033482	AA
USH1C	rs151045328	CC

Carrier Status

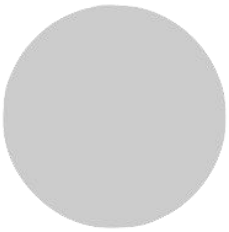
Usher Syndrome, Type Id

USH is a genetically heterogeneous condition characterised 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 characterised by profound congenital sensorineural deafness, absent vestibular function, and prepubertal onset of progressive retinitis pigmentosa, leading to blindness.

Your genetic map

Gene	SNP	Genotype
CDH23	rs397517313	II
CDH23	rs111033270	GG
PCDH15	rs111033260	GG
CDH23	rs397517323	CC

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Usher Syndrome, Type If

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

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Usher Syndrome, Type Iia

Usher Syndrome Type II is characterised 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
USH2A	rs397518008	II
USH2A	rs397517988	DD
USH2A	rs146733615	GG
USH2A	rs397517978	TT
USH2A	rs111033264	AA
USH2A	rs111033265	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Usher Syndrome, Type IIc

Usher Syndrome Type II is characterised 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	rs397517426	II
ADGRV1	rs397517429	DD
ADGRV1	rs376689763	CC
ADGRV1	rs371981035	AA
ADGRV1	rs397517436	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Usher Syndrome, Type IId

Usher Syndrome Type II is characterised 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	rs397517258	II
WHRN	rs397517255	GG

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

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

Carrier Status

Usher Syndrome, Type Iiia

Usher Syndrome Type III is characterised 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 the phenotypic heterogeneity of Usher Syndrome, see USH1 (276900). The genetic heterogeneity of Usher Syndrome Type III and 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	rs397517932	II
CLRN1	rs121908140	AA
CLRN1	rs111033267	GG
CLRN1	rs111033434	CC

What do your genetics tell us?

We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

https://www.omim.org/entry/276902

Carrier Status

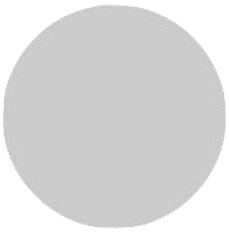
Acyl-Coa Dehydrogenase, Very Long-Chain, Deficiency Of

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 hypoglycaemia, liver disease, exercise intolerance and rhabdomyolysis. Over 400 cases have been reported worldwide. Its prevalence in Germany is of 1/50, 000.

Your genetic map

Gene	SNP	Genotype
ACADVL	rs753108198	II
ACADVL	rs751995154	GG
ACADVL	rs113994170	CC
ACADVL	rs113994167	TT
ACADVL	rs398123092	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

https://www.omim.org/entry/201475

Carrier Status

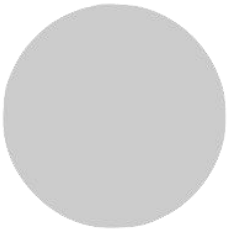
Weaver Syndrome

Weaver Syndrome (WVS) is a rare, multisystem disorder characterized by tall stature, an atypical 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. Its precise prevalence and incidence rates are not available.

Your genetic map

Gene	SNP	Genotype
EZH2	rs587783627	TT
EZH2	rs587783626	GG
EZH2	rs587783625	CC
EZH2	rs775407864	AA

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

https://www.omim.org/entry/277590

Carrier Status

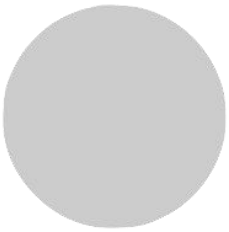
Wilson Disease

Wilson Disease is a very rare inherited multi-systemic disease presenting non-specific neurological, hepatic, psychiatric or osseo-muscular manifestations due to excessive copper deposition in the body.

Your genetic map

Gene	SNP	Genotype
ATP7B	rs193922111	II
ATP7B	rs768729972	DD
ATP7B	rs121907992	CC
ATP7B	rs121907998	AA
ATP7B	rs372436901	TT
ATP7B	rs76151636	GG

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

https://www.omim.org/entry/277900

Carrier Status

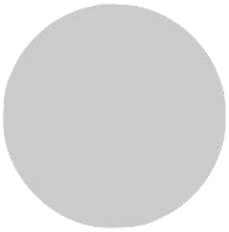
Agammaglobulinemia, X-Linked

X-linked Agammaglobulinemia (XLA) is a clinically variable form of isolated agammaglobulinemia, an inherited immunodeficiency disorder (see this term), and is characterised in affected males by recurrent bacterial infections during infancy. Its estimated prevalence is 1/350,000 to 1/700,000. Its annual incidence is not known. The disorder has been reported in various ethnic groups worldwide. Only males are affected, and females are asymptomatic carriers.

Your genetic map

Gene	SNP	Genotype
BTK	rs193922126	II
BTK	rs128620183	CC
BTK	rs128620187	GG
BTK	rs193922125	TT

What do your genetics tell us?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

More information:

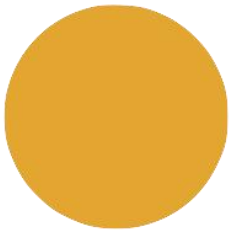
https://www.omim.org/entry/300755

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 do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22479202

Your genetic map

Gene	SNP	Genotype
LOC1027	rs3001032	TC
LOC6467	rs1515110	TG
GNL3	rs1108842	CC
ADIPOQ	rs182052	GG
ARL15	rs6450176	AG
VEGFA	rs998584	AC
LOC6454	rs668459	TT
TRIB1	rs2980879	TA
ADRB1	rs10885531	CC
PDE3B	rs11023332	GG
LOC1053	rs7955516	AC
ATP6V0A	rs6488898	AA
CDH13	rs12051272	GG
PEPD	rs731839	AG
PBRM1	rs2590838	AG
LOC1027	rs6810075	TT
LOC6454	rs592423	CC
LNTC1	rs601339	AA
GNP	rs2925979	TC
PEPD	rs4805885	TC

Biomarkers

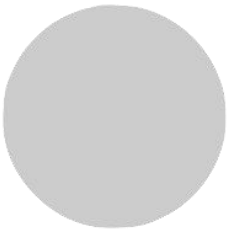
Beta-2 microglubulin plasma levels

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	CC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23417110

Biomarkers

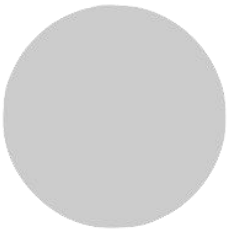
Bilirubin levels

Variation in serum bilirubin is associated with altered cardiovascular disease risk and drug metabolism.

Your genetic map

Gene	SNP	Genotype
UGT1A8	rs6742078	GG
HIST1H1T	rs12206204	CC
ARHGEF7	rs4773330	GG
SLCO1B1	rs4149056	TT

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19414484

Biomarkers

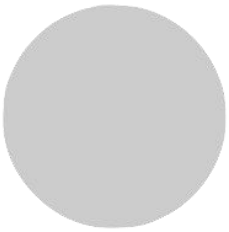
C-reactive protein

C-reactive Protein (CRP) have been used as critical markers contributing to acute and chronic inflammation.

Your genetic map

Gene	SNP	Genotype
FLJ20021	rs6846071	TG
DOCK4	rs10255299	GG
LOC1053	rs6904416	TT
KCNE4	rs960246	GG
HNF1A	rs2393791	TT
LOC1053	rs7600502	AA
PSMD3	rs8078723	TC
LOC1005	rs16993221	AA

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/22788528

Biomarkers

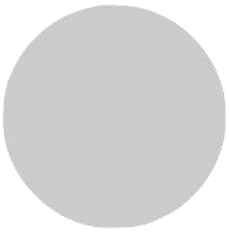
Calcium levels

Calcium is vital to the normal functioning of multiple organ systems, and its serum concentration is tightly regulated.

Your genetic map

Gene	SNP	Genotype
CASR	rs1801725	GG
DGKD	rs1550532	GG
GCKR	rs780094	TC
LOC1019	rs10491003	TC
CARS	rs7481584	AG
LOC1053	rs7336933	AG
CYP24A1	rs1570669	AA
WDR81	rs12150338	CC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/24068962

Biomarkers

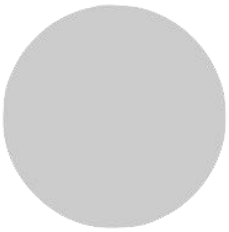
Dehydroepiandrosterone sulphate levels

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	GG
SRP14	rs7181230	AA
HHEX	rs2497306	CC
LOC1079	rs2185570	TT
TRIM4	rs17277546	GG
BCL2L11	rs6738028	CG
ARPC1A	rs740160	CC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/21533175

Biomarkers

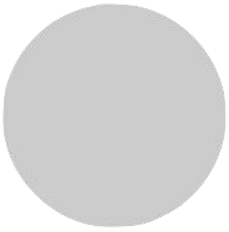
Eosinophil counts

Eosinophils are involved in the initiation and propagation of inflammatory responses. As such, they play important roles in the pathogenesis of inflammatory diseases

Your genetic map

Gene	SNP	Genotype
IL1RL1	rs1420101	TC
LOC1027	rs12619285	AG
TMED10P	rs4857855	CC
SH2B3	rs3184504	CC
IRF1 IL5	rs4143832	GG
WDR36	rs2416257	TC
TNXB	rs2269426	AA

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19198610

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	TC
FADS2	rs174570	CC
PIEZO1	rs9933309	CC
MYO9B	rs11667918	CC
ANK1	rs4737009	GG
FN3KRP	rs1046875	GG
ABCB11	rs3755157	CC
CDKAL1	rs7772603	TT
GCK	rs1799884	CC
LOC1053	rs13266634	CC

What do your genetics tell us?

According to this study, your propensity is to have normal levels, in line with the average person.

More information:

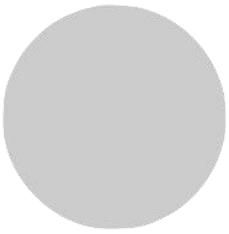
www.ncbi.nlm.nih.gov/pubmed/24647736

Biomarkers

Glycerophospholipid levels

Metabolites are small molecules involved in cellular metabolism, which can be detected in biological samples using metabolomic techniques

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/26068415

Your genetic map

Gene	SNP	Genotype
PKD2L1	rs603424	GG
MYRF	rs174536	AA
MYRF	rs174537	GG
TMEM25	rs102275	TC
FADS1	rs174546	TC
FADS1	rs174547	TT
FADS2	rs968567	CC
FADS2	rs1535	AG
FADS2	rs174578	TA
SGPP1	rs7157785	GG
TMEM22	rs1077989	AC
NTAN1	rs7200543	AG
NTAN1	rs6498540	AA
SPTLC3	rs680379	GG

Biomarkers

Homocysteine levels

Homocysteine (HC) is a sulfur amino acid important in the transfer of methyl groups in cell metabolism. It has been considered an influential factor in the development of cardiovascular and cerebrovascular diseases.

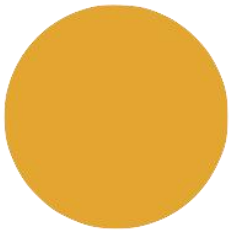
Recent studies have focused on the analysis of the relationship between hyperhomocysteinemia (increased plasma homocysteine concentration) and damage to neuronal cells by neurotoxic mechanisms, such as an increase in oxidative stress, the generation of homocysteine derivatives, as well as an increase in the toxicity of ß-amyloid protein, among others.

Homocysteine is synthesised as an intermediate product of the metabolism of methionine through the action of the methionine adenosyl transferase enzyme.

Your genetic map

Gene	SNP	Genotype
MTHFR	rs1801133	AG
MTR	rs2275565	GG
EEF1A1P4	rs9369898	AA
NOX4	rs7130284	CC
DPEP1	rs154657	AG
CBS	rs234709	CC
PRDX1	rs4660306	TC
SLC17A3	rs548987	CG
LOC1079	rs42648	AG
RPL12P33	rs2251468	CC
FGF21	rs838133	GG
TRDMT1	rs12780845	AA
NOX4	rs957140	GG
CBS	rs2851391	TC

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23824729

Biomarkers

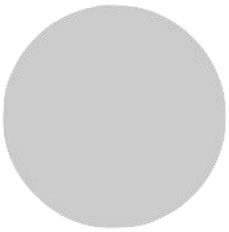
IgE levels

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 research

Your genetic map

Gene	SNP	Genotype
FCER1A	rs2251746	TC
STAT6	rs1059513	TC
IL13	rs20541	GG
LOC1053	rs2523809	TG
HLA W	rs2571391	AC
ACKR1	rs13962	GG
MTCO3P	rs2858331	AA
OR10J7P	rs4656784	AA
LPP	rs9290877	TC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

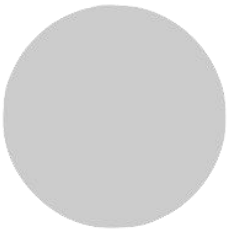
www.ncbi.nlm.nih.gov/pubmed/22075330

Biomarkers

Liver enzyme levels (gamma-glutamyl transferase)

Concentrations of liver enzymes in plasma are widely used as indicators of liver disease.

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/22001757

Your genetic map

Gene	SNP	Genotype
PNPLA3	rs738409	CC
RNU6	rs6984305	TT
LOC1053	rs10819937	GG
ABO	rs579459	TC
JMJD1C	rs7923609	GG
FADS2	rs174601	TC
ST3GAL4	rs2236653	TT
ASGR1	rs314253	TT
ABHD12	rs7267979	GG
LOC1019	rs1497406	AG
CEPT1	rs1335645	AA
EFHD1	rs2140773	AA
SLC2A2	rs10513686	GG
HPRT1P2	rs6888304	AA
MLXIPL	rs17145750	TC
DLG5	rs754466	AA
EXOC3L4	rs944002	AG
[illegible]ORA	rs339969	AC
[illegible]376	rs8038465	CC
LOC1027	rs4581712	AA
SOX9 AS1	rs9913711	CC
FUT2	rs516246	TC
MICAL3	rs1076540	TC
GGT1	rs2073398	CC

Biomarkers

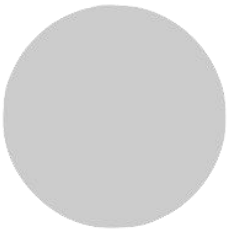
Liver enzyme levels

Plasma liver-enzyme tests are widely used at the clinic for the diagnosis of liver diseases and to monitor responses to drug treatment. There is considerable evidence that human genetic variation influences the plasma levels of liver enzymes

Your genetic map

Gene	SNP	Genotype
JMJD1C	rs12355784	CC
JMJD1C	rs10761779	AA
LINC0136	rs9803659	TC
ADAMTS1	rs4962153	GG
PNPLA3	rs2281135	AG
NBPF3	rs1780324	AA
	rs657152	AC
GPLD1	rs9467160	AG
GGT1	rs4820599	AA

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/18940312

Biomarkers

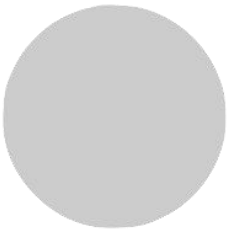
Magnesium levels

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	GG
MECOM	rs448378	AG

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/20700443

Biomarkers

Monocyte count

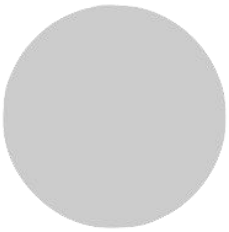
Monocytes are a type of agranulocyte white blood cells. It is the largest leukocyte.

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 investigate further. Circulating monocytes play a critical role in vascular diseases, such as in the formation of atherosclerotic plaque.

Your genetic map

Gene	SNP	Genotype
ITGA4	rs2124440	AG
LINC0156	rs2712381	AC
ACKR2	rs2228467	TT
PTGR1	rs2273788	CC
IRF8	rs424971	TT

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23314186

Biomarkers

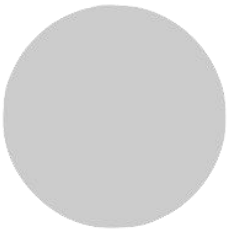
Neutrophil count

Neutrophils are leukocytes (white blood cells) of the granulocyte type, also called polymorphonuclear (PMN). White Blood Cell (WBC) count is a common clinical measurement used as a predictor of certain aspects of human health, including immunity and infection status. WBC count is also a complex trait that varies among individuals and ancestry groups.

Your genetic map

Gene	SNP	Genotype
CDK6	rs445	CC
MED24	rs8078723	TC
PSMD3	rs4794822	CC
AK12388	rs6936204	TC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

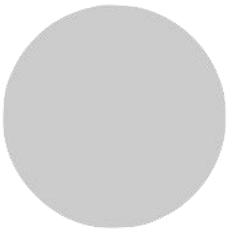
www.ncbi.nlm.nih.gov/pubmed/25096241

Biomarkers

Phospholipid levels (plasma)

Long-chain n-3 polyunsaturated fatty acids (PUFAs) can be the result of diet, or of α-linolenic acid (ALA), through elongation and desaturation

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/21829377

Your genetic map

Gene	SNP	Genotype
TMEM25	rs102275	TC
MYRF	rs174536	AA
RPLP0P2	rs1692120	AG
FADS1	rs174547	TT
FADS2	rs1535	AG
FADS2	rs174448	AG
FEN1	rs4246215	GG
UBXN4	rs16832011	AA
TMEM25	rs174538	AG
MYRF	rs174535	TC
FADS1	rs174550	TC
FADS2	rs174574	AC
ELOVL2	rs3798713	GC
BEST1	rs1109748	AC
LOC1019	rs1514178	TT
ELOVL2	rs3734398	CC
CYCP2L	rs4713103	TT
RAB3IL1	rs2521572	GG
PAGLA	rs198426	TT
GCNR	rs780094	TC
LOC105	rs9586179	TT
RPS2P37	rs4963452	TT
STIM2	rs6844153	TC
ELOVL2	rs4711171	CC

Biomarkers

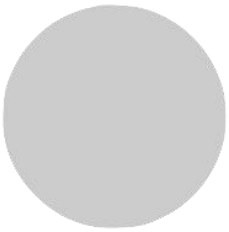
Phosphorus levels

Phosphorus is an essential mineral that sustains cellular energy and mineralizes the skeleton. Because the complex actions of ion transporters and regulatory hormones regulate serum phosphorus concentrations, genetic variation may determine inter-individual variations in phosphorus metabolism.

Your genetic map

Gene	SNP	Genotype
NBPF3	rs1697421	TT
CSTA	rs17265703	AA
IP6K3	rs9469578	CC
PDE7B	rs947583	TT
C12orf4	rs2970818	TT

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/20558539

Biomarkers

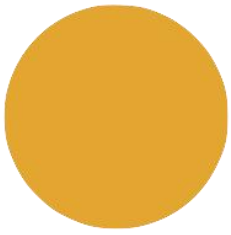
Omega-6 levels

Omega-6 are essential fatty acids that are crucial for certain bodily functions, but the body does not generate them, meaning it must obtain them through diet. They play a crucial role in brain function and normal growth and development. They also help to stimulate hair and skin growth, maintain bone health, regulate metabolism and maintain the reproductive system. They are found mainly in nuts, cereals, vegetable oils, avocados and eggs. Excess omega-6 in the blood can contribute to the onset of inflammatory diseases, while low levels can cause dermal disorders, such as eczema or hair loss, liver dysfunctions or kidney disorders.
Large-scale studies have shown that certain variants of the ELOVL2 gene cause people who carry that variant to have abnormal levels of omega-6.

Your genetic map

Gene	SNP	Genotype
PDXDC1	rs2280018	AA
TMEM25	rs102275	TC
IL23R	rs7517847	TT
C10orf12	rs17009617	GG
FADS1	rs174550	TC
FADS2	rs2727270	CC
PDXDC1	rs1136001	GG
FTLP19	rs2069036	CC
FADS1	rs174547	TT
PDXDC1	rs4985155	AG
TMEM39	rs16829840	CC
PDXDC1	rs1741	GC
ELOVL2	rs2236212	CC

What do your genetics tell us?



Based on this study, your predisposition to have abnormal levels is above average. Other genetic and clinical factors may be relevant.

More information:

www.ncbi.nlm.nih.gov/pubmed/24823311

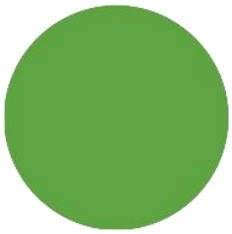
Biomarkers

Platelet count

Platelets are small fragments of blood cells. Their function is to form blood clots, which help to heal wounds and prevent bleeding. 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 do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22139419

Your genetic map

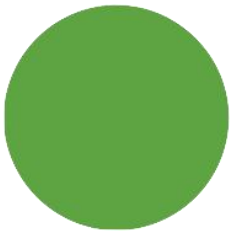
Gene	SNP	Genotype
MFN2	rs2336384	TT
DNM3	rs10914144	TC
TMCC2	rs1668871	TT
GCSAML	rs7550918	TT
TRIM58	rs3811444	TT
EHD3	rs625132	AG
THADA	rs17030845	TT
LOC3398	rs7641175	AA
ARHGEF3	rs1354034	TC
PDIA5	rs3792366	AG
KLHL8	rs7694379	GG
F2R	rs17568628	TT
MEF2C	rs700585	TC
IRF1	rs2070729	AC
LRRC16A	rs441460	AA
HLA B	rs3819299	TT
HLA DOA	rs399604	TT
RN7SL26	rs210134	GG
LOC1070	rs9399137	TC
LOC1027	rs342275	TC
HYAL4	rs4731120	AA
PLEC	rs6995402	TC
AK3	rs409801	TC
RCL1	rs13300663	GG
CDKN2A	rs3731211	TA
PSMD13	rs505404	TT
FEN1	rs4246215	GG
CBL	rs4938642	GG
LOC1053	rs7342306	GG
BAZ2A	rs941207	CC
SH2B3	rs3184504	CC

Biomarkers

Red blood cell count

Haemoglobin is a protein present in red blood cells that carries oxygen to the body's organs and tissues, and transports carbon dioxide from organs and tissues back to the lungs. If the level of haemoglobin is lower than normal, it means that one has a low red blood cell count (anemia).

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20139978

Your genetic map

Gene	SNP	Genotype
PRKCE	rs10168349	GG
ABO	rs495828	TG
LOC1053	rs7173947	TT
ALPL	rs2242420	CC
GPLD1	rs6911965	TT
PNPLA3	rs2896019	TT
BRAP	rs3782886	TT
MRC1	rs2477664	TT
LOC1053	rs9820070	CC
SLC14A2	rs4890568	AA
LOC1053	rs11709625	CC
CD163	rs7136716	AG
ALDH2	rs671	GG
TMPRSS6	rs5756504	TC
PRKCE	rs10495928	AG
LIPC	rs1077834	TT
LOC1019	rs7350481	CC
HERPUD1	rs3764261	CC
[illegible]	rs12678919	AG
LOC1079	rs7775698	TC
TMPRSS6	rs2413450	CC
WDR72	rs10518733	AC
TNFRSF1	rs4273077	AA
RPS11	rs2280401	AA
HBA2	rs2858942	AC
RCL1	rs2236496	TT
LINC008	rs4916483	TT
TMPRSS6	rs855791	AA
LOC6454	rs632057	GG
DENND4	rs6494537	CC
TYMP	rs470119	CC

Biomarkers

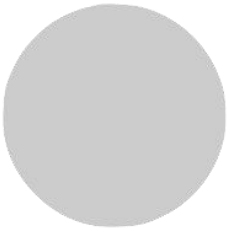
Serum albumin level

Many disorders are associated with altered serum protein concentrations, including malnutrition, cancer, and cardiovascular, kidney, and inflammatory diseases.

Your genetic map

Gene	SNP	Genotype
MIR22HG	rs11078597	TT
ACTBP9	rs694419	CC
RPS11	rs2280401	AA
FRMD5	rs16948098	GG
TNFRSF1	rs4561508	CC
FKBPL	rs204999	AG
LOC1079	rs2675609	CC
HPN AS1	rs11671010	TC
CHRNA3	rs12914385	TC
ELL2	rs3777200	CC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23022100

Biomarkers

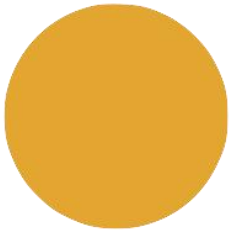
Serum total protein level

Many disorders are associated with altered serum protein concentrations, including malnutrition, cancer, and cardiovascular, kidney, and inflammatory diseases.

Your genetic map

Gene	SNP	Genotype
TNFRSF1	rs4561508	CC
intergeni	rs204999	AG
FNDC4	rs1260326	TC
ARID5B	rs2675609	CC
FCGRT	rs2280401	AA
ELL2	rs3777200	CC

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23022100

Biomarkers

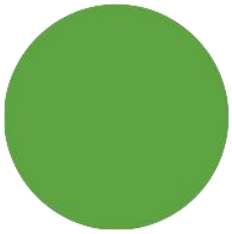
Sex hormone levels

Genetic factors contribute strongly to sex hormone levels, yet knowledge of the regulatory mechanisms remains incomplete.

Your genetic map

Gene	SNP	Genotype
ZNF789	rs148982377	CC
LOC1462	rs117145500	AA
LOC1053	rs11031002	TT
ANO2	rs117585797	CC
ZKSCAN5	rs34670419	GG
SLC22A2	rs112295236	CC
SULT2A1	rs2637125	GG
LOC1027	rs12294104	CC

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

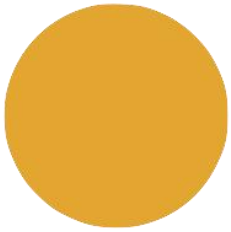
www.ncbi.nlm.nih.gov/pubmed/26014426

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 spans. In addition, even mild alterations in thyroid function are associated with weight changes, atrial fibrillation, osteoporosis, and psychiatric disorders.

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23408906

Your genetic map

Gene	SNP	Genotype
PDE8B	rs6885099	AG
PDE10A	rs753760	GC
LOC1053	rs10799824	GG
LOC1053	rs3813582	TT
LOC1079	rs9472138	CC
LINC0151	rs11755845	CC
LOC1079	rs10032216	TT
LOC1019	rs13015993	AA
SOX9	rs9915657	TT
NFIA	rs334699	GG
FAM227B	rs10519227	TT
PRDM11	rs17723470	TC
DET1	rs17776563	GG
INSR	rs4804416	TG
	rs657152	AC
ITPK1	rs11624776	AA
IRG1	rs7825175	GG
LINC006	rs1537424	TC
SASH1	rs9497965	CC
GLI3	rs1571583	GG
DIO1	rs2235544	AC
LHX3	rs7860634	AA
KRT18P13	rs7045138	TC
LOC1053	rs11726248	GG
LPCAT2	rs6499766	AA
LOC1005	rs7240777	GG

Biomarkers

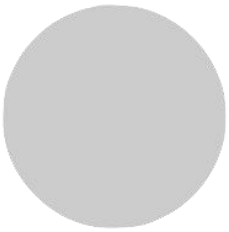
Uric acid levels

Elevated serum uric acid levels cause gout and are a risk factor for cardiovascular disease and diabetes.

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	AA
SLC22A11	rs17300741	AA
SLC22A11	rs505802	TT

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19503597

Biomarkers

White blood cell count

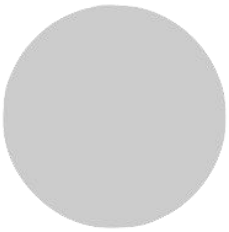
White blood cells are a type of blood cell that is produced in the bone marrow and found in blood and lymphatic tissues. White blood cells are part of the body's immune system. These help the body fight infections and other diseases. The types of white blood cells are granulocytes (neutrophils, eosinophils, and basophils), monocytes, and lymphocytes (T cells and B cells).

White blood cell count is a common clinical measurement of whole blood count tests, and varies widely among healthy individuals.

Your genetic map

Gene	SNP	Genotype
LINC0156	rs4328821	AA
EPS15L1	rs10411936	AG
LOC1019	rs1449263	TC
LINC0156	rs9880192	GC
CCDC26	rs10098310	AG
LOC1053	rs10980800	TT
PSMD3	rs8078723	TC
HCG22	rs2517510	TG
PSMD3	rs4794822	CC

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/21738480

Biometrics

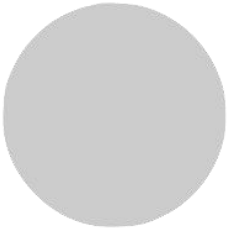
Aortic root size

Echocardiographic measures of Left Ventricular (LV) structure and function are heritable phenotypes of cardiovascular disease.

Your genetic map

Gene	SNP	Genotype
SLC35F1	rs89107	GG
TMEM23	rs17132261	CC
SMG6	rs10852932	TG
PRDM6	rs17470137	AG
HMGA2	rs4026608	TT
LOC1005	rs10770612	AA
LOXL1	rs893817	AG

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

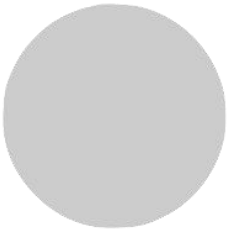
www.ncbi.nlm.nih.gov/pubmed/19584346

Biometrics

Bone mineral density

Bone Mineral Density (BMD) is the most widely used predictor of fracture risk.

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/22504420

Your genetic map

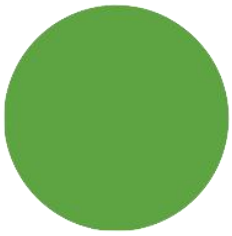
Gene	SNP	Genotype
FABP3P2	rs9533090	CC
ARHGAP1	rs7932354	TC
AXIN1	rs9921222	TC
TMEM26	rs1053051	TC
RPS3AP2	rs13336428	AG
C17orf53	rs227584	AC
FAM210A	rs4796995	AG
CCDC170	rs4869742	TC
CPED1	rs13245690	AA
LOC1001	rs4817775	CC
CPN1	rs7084921	CC
LOC1053	rs430727	TC
LOC1079	rs1564981	AG
DCDC5	rs163879	TC
RHEBL1	rs12821008	CC
DNM3	rs479336	GG
LOC1079	rs2887571	AA
FOXL1	rs10048146	AA
FOXP3	rs7851693	CC
CSDNP3	rs1346004	GG
GPATCH1	rs10416218	TC
HOXC6	rs736825	CG
IDUA	rs3755955	AG
LOC1053	rs1878526	GG
JAG1	rs3790160	CC
KCNMA1	rs7071206	TT
KIAA2018	rs1026364	TG
LOC1053	rs7953528	TT
LEKR1	rs344081	TT
RPL37AP	rs10835187	TC
LRP5	rs3736228	CC

Biometrics

Heart rate

An elevated resting heart rate is associated with a greater risk of cardiovascular disease.

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23583979

Your genetic map

Gene	SNP	Genotype
TFPI	rs4140885	GG
LOC1053	rs180242	AA
RNU3P3	rs17796783	TC
SYT10	rs7980799	CC
LOC1053	rs17287293	AG
CD46	rs11118555	TT
MYH6	rs365990	AA
LOC1053	rs1015451	TT
ACHE	rs13245899	AA
FADS1	rs174549	GG
SLC35F1	rs11153730	TC
KIAA1755	rs6127471	TC
CCDC141	rs17362588	GG
GNB4	rs7612445	GG
CHRM2	rs2350782	TT
NKX2 5	rs6882776	GG
LOC1053	rs13030174	AC
FNDC3B	rs9647379	CG
[illegible]4	rs2067615	AT
CPNE8	rs826838	TT
RBFOX	rs11645781	GG
SLC10A7	rs10213084	GG
RNU4	rs11154027	TC
LOC1079	rs11578508	AA
HMGN2P	rs17083533	GG
LOC1019	rs7722600	AA

Biometrics

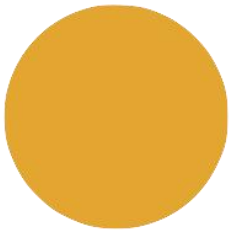
Resting heart rate

A high 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	GG
LOC1053	rs17287293	AG
SLC35F1	rs281868	GG
SLC12A9	rs314370	TT
UFSP1	rs12666989	GG
FADS1	rs174547	TT

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/20639392

Traits

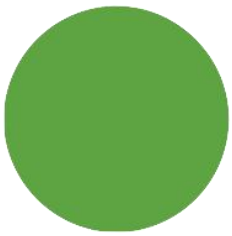
Spirometric measure of pulmonary function (Forced vitalcapacity)

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	TT
BMP6	rs6923462	CC
MIR129 2	rs4237643	TT
PRDM11	rs2863171	AA
WWOX	rs1079572	AG

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

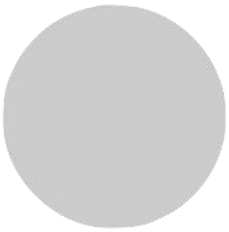
www.ncbi.nlm.nih.gov/pubmed/24929828

Traits

Menopause (age at onset)

Menopause is the cessation of the reproductive function of the human ovaries. This life stage is associated with one of the major hormonal changes in women, characterised by a decline in the secretion of estrogen, progesterone and, to a lesser degree, testosterone. It influences a woman's well-being and is associated with several major age-related diseases, including cardiovascular disease, breast cancer, osteoarthritis, and osteoporosis.

What do your genetics tell us?



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

More information:

www.ncbi.nlm.nih.gov/pubmed/22267201

Your genetic map

Gene	SNP	Genotype
EXO1	rs1635501	TC
FNDC4	rs2303369	TC
TLK1	rs10183486	TC
UIMC1	rs365132	TG
SYCP2L	rs2153157	AG
ASH2L	rs2517388	TT
LOC1027	rs12294104	CC
PRIM1	rs2277339	TT
TDRD3	rs4886238	GG
POLG	rs2307449	TG
GSPT1	rs10852344	TT
TMEM150	rs11668344	AA
NLRP11	rs12461110	GG
MCM8	rs16991615	GG

Traits

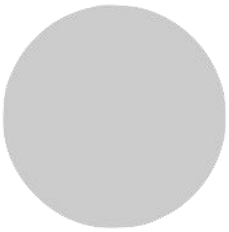
Smoking behavior

Consistent but indirect evidence has implicated genetic factors in smoking as a behaviour.

Your genetic map

Gene	SNP	Genotype
HECTD2	rs1329650	TG
RAB4B	rs3733829	GG
BDNF	rs6265	CC
FAM163B	rs3025343	GG

What do your genetics tell us?



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

More information:

www.ncbi.nlm.nih.gov/pubmed/20418890

Pharmacogenomics: Cardiology

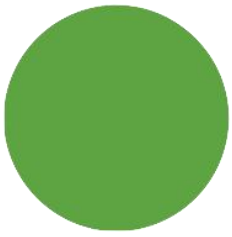
Pravastatin

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 do your genetics tell us?



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 the breakdown of LDL cholesterol.

Your genetic map

Gene	SNP	Genotype
SLCO1B1	rs4149056	TT

What do your genetics tell us?

Patients with the TT genotype may be at a lower risk of simvastatin-related myopathy as compared to patients with the CT or CC genotype. Other genetic and clinical factors may also affect a patient's risk for toxicity.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/28482130

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.

Your genetic map

Gene	SNP	Genotype
VKORC1	rs9923231	CC

What do your genetics tell us?

Patients with the CC genotype may require an increased dose of warfarin as compared to patients with the TC or TT genotype. Other genetic and clinical factors may also influence a patient's warfarin dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/gtr/conditions/CN078029

Pharmacogenomics: Neurology

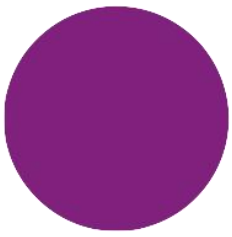
Bupropion

A unicyclic, aminoketone antidepressant. The mechanism of its therapeutic actions is not well understood, but it does appear to block dopamine uptake. Hydrochloride is available as an aid to smoking cessation treatments.

Your genetic map

Gene	SNP	Genotype
ANKK1	rs1800497	AA

What do your genetics tell us?



Patients with the AA genotype who are treated with bupropion may be less likely to quit smoking as compared to patients with the GG genotype, although this has been contradicted in one study. Other genetic and clinical factors may also influence a patient's capacity to quit smoking.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15492764

Pharmacogenomics: Oncology

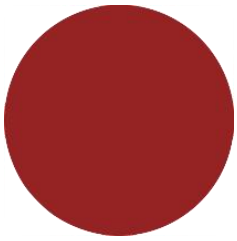
Methotrexate

An antineoplastic antimetabolite with immunosuppressive 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	AG

What do your genetics tell us?



Patients with AG genotype and leucemia or lymphoma who are treated with methotrexate: 1) may have a poorer response 2) may be at an increased risk of toxicity 3) may require a lower dose of methotrexate, and 4) may be at a greater risk of folate deficiency as compared to patients with GG genotype. When comparing with AA genotype, the opposite is true. This association has been contradicted in other studies. Other factors may also have an effect.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/22143415

Pharmacogenomics: Oncology

Vincristine

Vincristine is an anti-tumour 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 leucemia, malignant lymphoma, Hodgkin's disease, acute erythraemia, and acute panmyelosis. Vincristine sulfate is often chosen as part of polychemotherapy because of its lack of significant bone–marrow suppression (at recommended doses) and unique clinical toxicity (neuropathy).

Your genetic map

Gene	SNP	Genotype
LOC1009	rs924607	TC

What do your genetics tell us?

Patients with the TC genotype may have decreased, but not absent, risk of peripheral nervous system diseases when treated with vincristine as compared to patients with the TT genotype. Other genetic and clinical factors may also affect 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, converting 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 S phase of the cell phase cycle. 5-Fluorouracil is involved in the synthesis of DNA and inhibits, to a small degree, the formation of RNA. The two 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, which, preferentially, take advantage of the uracil molecule for nucleic acid biosynthesis.

Your genetic map

Gene	SNP	Genotype
DPYD	rs67376798	TT

What do your genetics tell us?

TT-genotype patients treated with fluoropyrimidine-based chemotherapy may exhibit 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 have an influence.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/17700593

Pharmacogenomics: Other

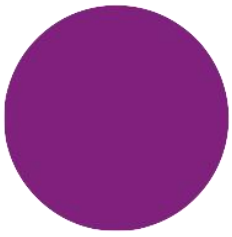
Peginterferon Alpha-2b

Peginterferon alfa-2b is a form of recombinant interferon used as part of combination therapy to treat chronic Hepatitis C, an infectious liver disease caused by infection with the Hepatitis C Virus (HCV). HCV is a single-stranded RNA virus that is categorised into nine distinct genotypes, with genotype 1 being the most common in the United States, and affecting 72% of all chronic HCV patients. Treatment options for chronic Hepatitis C have advanced significantly since 2011, with the development of Direct Acting Antivirals (DAAs) resulting in less use of Peginterferon alfa-2b. Peginterferon alfa-2b is derived from the alfa-2b moiety of recombinant human interferon, and acts by binding to human type-1 interferon receptors. The activation and dimerization of this receptor induces the body's innate antiviral response by activating the Janus kinase/signal transducer and activator of transcription (JAK/STAT) pathway.

Your genetic map

Gene	SNP	Genotype
IFNL3	rs12979860	TC

What do your genetics tell us?



Patients with the TC genotype and Hepatitis C genotype 1 may exhibit a decreased response (sustained virological response, SVR) when administered peg interferon alpha (2a, 2b) and ribavirin as compared to patients with the CC genotype. Patients with the TC genotype may also have lower spontaneous clearance in acute HCV infections than patients with the CC genotype. Other genetic and clinical factors may also affect a patient's response to peg interferon and ribavirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21145807

Pharmacogenomics: Other

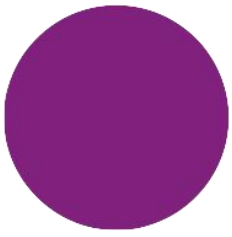
Ribavirin

Producing broad-spectrum activity against several RNA and DNA viruses, Ribavirin is a synthetic guanosine nucleoside and antiviral agent that interferes with the synthesis of viral mRNA. It is primarily indicated for use in treating hepatitis C and viral hemorrhagic fevers. HCV is a single-stranded RNA virus that is categorised into nine distinct genotypes, with genotype 1 being the most common in the United States, and affecting 72% of all chronic HCV patients. It is reported that ribavirin might be effective only in the early stages of viral hemorrhagic fevers, including Lasser fever, Crimean-Congo hemorrhagic fever, Venezuelan hemorrhagic fever, and Hantavirus infection. Ribavirin is a prodrug that is metabolised into nucleoside analogs, blocking viral RNA synthesis and viral mRNA capping. Before the development of newer drugs, ribavirin and dual therapy was considered the first-generation and standard antiviral treatment. Newer drugs developed as hepatitis C viral infection treatments can be used to reduce or eliminate the use of ribavirin, which is associated with serious adverse effects.

Your genetic map

Gene	SNP	Genotype
IFNL3IFN	rs12979860	TC

What do your genetics tell us?



Patients with the TC genotype and Hepatitis C genotype 1 may exhibit a decreased response (sustained virological response, SVR) when administered peg interferon alpha (2a, 2b) and ribavirin. They may also exhibit lower spontaneous clearance in acute HCV infections than patients with the CC genotype. Other genetic and clinical factors may also affect a patient's response to peg interferon and ribavirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21145807

Pharmacogenomics: Other

Tacrolimus

Tacrolimus (also FK-506 or Fujimycin) is an immunosuppressive drug mainly used after an organ transplant, to reduce the activity of the patient's immune system and, thereby, the risk of organ rejection. It is also used in a topical preparation for 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 containing 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 do your genetics tell us?

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 factors, such as CYP3A5 (rs776746), may also influence a patient's dose requirements.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23778326

Pharmacogenomics: Pain

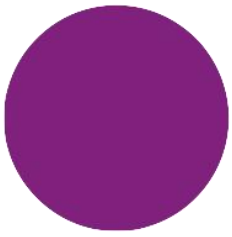
Meperidine

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 labour. Prolonged use may lead to dependence on 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 do your genetics tell us?



Patients with the TC genotype may have decreased opioid analgesic requirements after surgery. Other genetic and clinical factors may also have an effect.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenomics: Pain

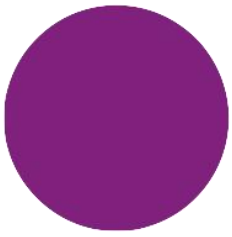
Morphine

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 do your genetics tell us?



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 affect a patient's opioid dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenomics: Pain

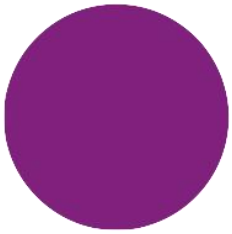
Pentazocine

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 do your genetics tell us?



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

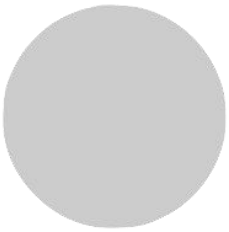
Aspirin

Aspirin, also known as acetylsalicylic acid (ASA), is a medication used to treat pain, fever, and inflammation. Specific inflammatory conditions for which aspirin is used include Kawasaki disease, pericarditis, and rheumatic fever. Aspirin is a non-steroidal 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 do your genetics tell us?



Patients with the AA genotype who are treated with aspirin may be at a decreased, though 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

SAMPLE REPORT