GENETIC HEALTH RISK: **WHAT YOU SHOULD KNOW**

Genetic Health Risk reports tell you about genetic variants associated with increased risk for certain health conditions. They do not diagnose cancer or any other health conditions or determine medical action.

Factors like lifestyle and environment can also affect whether a person develops most health conditions. Our reports cannot tell you about your overall risk for these conditions, and they cannot determine if you will or will not develop a condition.

Having a risk variant does not mean you will definitely develop a health condition. Similarly, you could still develop the condition even if you do not have a variant detected. It is possible to have other genetic risk variants not included in these reports.

These reports do not replace visits to a healthcare professional. Consult with a healthcare professional for help in interpreting genetic results. Results should not be used to make medical decisions.

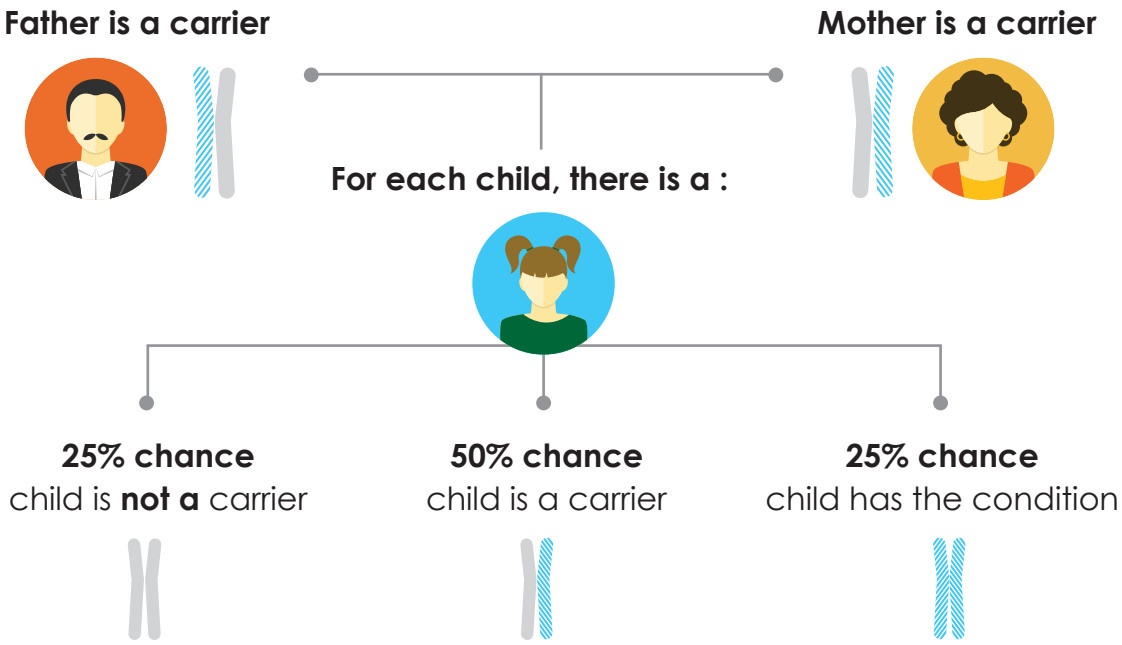
CARRIER STATUS TESTS: WHAT YOU SHOULD KNOW

Carrier status tests detect genetic variants that can cause inherited conditions. These variants are often found primarily in certain ethnicities.

Being a carrier means you have one variant for the condition. Carriers typically do not have the condition but can pass the variant to their children.

Knowing your carrier status is important when having children. If you and your partner are both carriers, you may have a child with the condition.

Genetic counseling can help you understand your results and options. It is recommended before and after testing, and also if you are a carrier.



If both parents are carriers, their child may inherit two variants and have the condition.

DRUG RESPONSE: **WHAT YOU SHOULD KNOW**

Introduction

Many drugs have well understood and narrowly defined targets such as a particular cellular receptor or intracellular enzyme. They may also have known specific interactions with enzymes responsible for activation, breakdown and clearance of the drugs. Genetic variants can determine how these drug-processing enzymes work.

Our body has more than thousands of genes that we inherited from our parents. Some genes are responsible for how your body processes medications. Drug response tests look for changes or variants in these genes that may determine whether a medication could be an effective treatment for you or whether you could have side effects to a specific medication.

Understanding Your Results

We have grouped the drugs we tested into several responses to a particular drug. Your DNA results provide information about how genes affect your response to different drugs.

Ultra Good Response
Based on your DNA profile, your metabolism of these drugs is too rapid and it may result in suboptimal therapeutic response to these drugs. You may consider to increase the starting dosage of these drugs.

Good Response
Based on your DNA profile, your metabolism of these drugs is at optimum rate (better than normal rate) and it may result in better therapeutic response to these drugs. You may use it as what is directed.

Normal Response
Based on your DNA profile, your metabolism of these drugs is at normal rate and it may result in normal therapeutic response to these drugs. You may use it as what is directed.

Intermediate Response
Based on your DNA profile, your metabolism of these drugs is at slightly slower rate and it may result in suboptimal therapeutic response to these drugs. Furthermore, the risk for developing side effects is slightly higher. You may consider to decrease the starting dosage of these drugs.

Low Response
Based on your DNA profile, your metabolism of these drugs is at a lower rate and it may result in suboptimal therapeutic response to these drugs. Furthermore, the risk for developing side effects is higher. You may consider to decrease the starting dosage of these drugs and use them with caution.

Disclaimer
These results are independent of medical diagnosis. For medical diagnosis and treatment decisions, consultation with a doctor or healthcare profession is necessary. One single DNA test cannot be used to determine how you will respond to all medications. You may need more than one test if you are taking more than one medication. This test is not available for all medications. It is available only for certain medications.

GENETIC HEALTH RISKS: **SUMMARY**

Disease Name	Variant Detected	Page
VISION		
Exfoliation Glaucoma	1 / 3	9
Age-related Cataract	0 / 1	10
Age-related Macular Degeneration	1 / 2	11
New Myopia	1 / 1	12
New Hyperopia	1 / 2	13
New Corneal Astigmatism	0 / 1	14
New Diabetic Retinopathy	0 / 3	15
New Vogt-Koyanagi-Harada Disease	1 / 3	16
SKIN ISSUES		
Atopic Dermatitis	3 / 8	18,19
Psoriasis	0 / 1	20
Keloid	1 / 1	21
Generalized Vitiligo	1 / 5	22
RESPIRATORY SYSTEM		
Asthma	1 / 2	24
Allergic Rhinitis	0 / 1	25
Chronic Obstructive Pulmonary Disease	0 / 2	26
Pulmonary Fibrosis	4 / 4	27
New Emphysema	0 / 1	28
New Obstructive Sleep Apnea	0 / 1	29
NERVOUS SYSTEM		
Alzheimer's Disease	0 / 1	31
Parkinson's Disease	1 / 2	32
Multiple Sclerosis	1 / 2	33
New Glioma	1 / 4	34
New Migraine	3 / 8	35
New Essential Tremor	0 / 1	36

Disease Name	Variant Detected	Page
ENDOCRINE SYSTEM		
Hypothyroidism	1 / 3	38
Graves' Disease	2 / 5	39
Type 2 Diabetes	2 / 3	40
CARDIOVASCULAR SYSTEM		
Hypertension	2 / 4	42
Coronary Heart Disease	3 / 5	43
Acute Myocardial Infraction	3 / 4	44
Ischemic Stroke	0 / 2	45
Atrial Fibrillation	1 / 5	46
Dilated Cardiomyopathy	0 / 1	47
Hypertriglyceridemia	1 / 2	48
New Coronary Spasm	0 / 1	49
New Intracranial Aneurysm	6 / 6	50
New Peripheral Artery Disease	1 / 2	51
New Venous Thromboembolism	1 / 3	52
New Abdominal Aortic Aneurysm	3 / 3	53
New Kawasaki Disease	2 / 5	54
New Orthostatic Hypotension	0 / 1	55
New Carotid Artery Disease	0 / 2	56
New Brugada Syndrome	2 / 2	57
URINARY SYSTEM		
Chronic Kidney Disease	1 / 1	59
Renal Calculus	2 / 4	60
IgA Nephropathy	3 / 3	61
Acquired Nephrotic Syndrome	0 / 1	62

GENETIC HEALTH RISKS: **SUMMARY**

Disease Name	Variant Detected	Page
INFECTION		
New Tuberculosis	0 / 2	64
New Chronic Hepatitis B	2 / 2	65
New Chronic Hepatitis C	0 / 1	66
New Leprosy	4 / 5	67,68
New Dengue Shock Syndrome	0 / 1	69
New Severe Malaria	2 / 2	70
GENDER		
New Endometriosis	4 / 8	72
New Uterine Fibroids	1 / 1	73
New Polycystic Ovary Syndrome	8 / 11	74,75
New Gestational Diabetes	2 / 3	76
New Intrahepatic Cholestasis of Pregnancy	1 / 1	77
New Erectile Dysfunction	-- / 4	78
New Postpartum Depression	1 / 4	79
New Postpartum Stretch Marks	1 / 4	80
New Postpartum Scarring	0 / 1	81
New Arachidonic Acid Deficiency	0 / 2	82
BONE		
New Hip Fracture	3 / 7	84,85
New Lumbar Spine Stenosis	2 / 7	86
New Osteoporosis	1 / 2	87
EAR		
New Age-related Hearing Impairment	1 / 2	89
New Otosclerosis	1 / 2	90

Disease Name	Variant Detected	Page
NOSE		
Rhinosinusitis	0 / 2	92
MOUTH		
New Dental Caries	1 / 2	94
New Periodontitis	1 / 3	95
MENTAL		
New ADHD	3 / 4	97
New Depression	0 / 4	98
New Autism	1 / 4	99
New Panic Disorder	0 / 1	100
New Eating Disorder	0 / 5	101
New Tourette Syndrome	3 / 4	102
New Chronic Fatigue Syndrome	2 / 3	103
New Obsessive Compulsive Disorder	1 / 3	104
OTHERS		
Rheumatoid Arthritis	2 / 5	106
Gout	1 / 4	107
Scoliosis	1 / 1	108
Restless Legs Syndrome	0 / 1	109
Alcohol Dependence	2 / 2	110
Bipolar Disorder	1 / 3	111
Hypersomnia	2 / 2	112
Hemochromatosis (HFE-related)	0 / 2	113
Gallstones	0 / 1	114
Primary Biliary Cirrhosis	1 / 2	115
Celiac Disease	0 / 1	116

CARRIER STATUS TESTS: **SUMMARY**

Disease Name	Variant Detected	Page	Disease Name	Variant Detected	Page
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)		118	Limb-Girdle Muscular Dystrophy Type 2E (LGMD2E)		142
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)		119	Limb-Girdle Muscular Dystrophy Type 2I (LGMD2I)		143
Autosomal Recessive Polycystic Kidney Disease (ARPKD)		120	Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD deficiency)		144
Beta Thalassemia and Related Hemoglobinopathies		121,122	Maple Syrup Urine Disease Type 1B (MSUD 1B)		145
Bloom Syndrome		123	Mucolipidosis Type IV		146
Canavan Disease		124	Neuronal Ceroid Lipofuscinosis (CLN5-Related)		147
Congenital Disorder of Glycosylation Type 1a (PMM2-CDG)		125	Neuronal Ceroid Lipofuscinosis (PPT1-Related)		148
Cystic Fibrosis		126,127	Niemann-Pick Disease Type A		149
D-Bifunctional Protein Deficiency (DBPD)		128	Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related)		150
Dihydrolipoamide Dehydrogenase Deficiency (DLD deficiency)		129	Pendred Syndrome and DFNB4 Hearing Loss (SLC26A4-Related)		151
Familial Dysautonomia		130	Phenylketonuria and Related Disorders (PKU)		152,153
Familial Hyperinsulinism (ABCC8-Related)		131	Primary Hyperoxaluria Type 2 (PH2)		154
Familial Mediterranean fever (FMF)		132	Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1)		155
Fanconi Anemia Group C		133	Salla Disease		156
GRACILE Syndrome		134	Sickle Cell Anemia		157
Gaucher Disease Type 1		135	Sjögren-Larsson Syndrome		158
Glycogen Storage Disease Type Ia (GSDIa)		136	Tay-Sachs Disease		159
Glycogen Storage Disease Type Ib (GSDIb)		137	Tyrosinemia Type I		160
Hereditary Fructose Intolerance		138	Usher Syndrome Type 1F (Usher 1F)		161
Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related)		139	Usher Syndrome Type 3A (Usher 3A)		162
Leigh Syndrome, French Canadian Type (LSFC)		140	Zellweger Syndrome Spectrum (ZSS) (PEX1-Related)		163
Limb-Girdle Muscular Dystrophy Type 2D (LGMD2D)		141			

Variant not detected Variant detected

DRUG SENSITIVITY: SUMMARY

Category	Drug Name	Results	Page
Common Drugs and Hospitalized Medication	Clopidogrel	Ultra Good Response - Increase the starting dosage	165
	Warfarin	Good Response - Use as directed	166
	Isoniazid	Good Response - Use as directed	167,168
	Omeprazole	Ultra Good Response - Increase the starting dosage	169
	Simvastatin	Low Response - Use with caution	170
	Sulfonylureas	Good Response - Use as directed	171
	Allopurinol	Good Response - Use as directed	172
	Citalopram	Good Response - Use as directed	173
	Diazepam	Ultra Good Response - Increase the starting dosage	174
	Caffeine	Good Response - Use as directed	175
	Ethanol	Good Response - Use as directed	176
	Voriconazole Tablets	Good Response - Use as directed	177,178
	Tacrolimus	Good Response - Use as directed	179
Common Drugs for Cancer	Fluorouracil	Normal Response - Use as directed	181
	Thiopurines	Good Response - Use as directed	182,183
	Capecitabine	Normal Response - Use as directed	184
Others	Abacavir	Good Response - Use as directed	186
	Celecoxib	Good Response - Use as directed	187
	Sildenafil	Low Response - Use with caution	188

HEALTH RISK
VISION

Health Risk

Exfoliation Glaucoma

Exfoliation Glaucoma (XFG) is characterized by a group of symptoms with optic atrophy and visual field defects. The most significant risk factor is pathologically elevated intraocular pressure (eye pressure). The elevated level of intraocular pressure and the tolerance of the optic nerve over pressure damage are associated with the occurrence and development of glaucomatous optic atrophy and visual field loss.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The signs and symptoms of glaucoma vary depending on the type and stage of your condition. For example:

- Open-angle glaucoma
- Patchy blind spots in your side (peripheral) or central vision, frequently in both eyes
- Tunnel vision in the advanced stages
- Acute angle-closure glaucoma
- Severe headache
- Eye pain
- Nausea and vomiting
- Blurred vision
- Halos around lights
- Eye redness

RISK FACTORS

Certain factors may increase your risk of developing exfoliation glaucoma which include:

- Having high internal eye pressure (intraocular pressure)
- Being over age 60
- Being black, Asian or Hispanic
- Having a family history of glaucoma
- Having corneas that are thin in the center
- Being extremely nearsighted or farsighted
- Having had an eye injury or certain types of eye surgery
- Taking corticosteroid medications, especially eyedrops, for a long time

Gene	Your Genotype	Your Result	Explanation
COL11A1	AA	Normal Risk	When this site carries G allele, it is significantly negatively correlated with the depth of the anterior chamber of the eye, which may increase the risk of exfoliation glaucoma.
PLEKHA7	TC	Higher Risk	If gene locus carries the risk C allele can increase the risk of disease.
Intergenic	CC	Normal Risk	According to large sample analysis, this site that carries the risk T allele can increase disease susceptibility.

Health Risk

Age-related Cataract

Age-related cataract is the most common type of cataract, with the incidence increased with age. It is thought to be associated with a slower metabolism in elderly due to degenerative changes, but in most cases the condition progresses slowly and does not affect vision. In some cases, it is true that the lens opacity (cloudiness) affects vision, while the diagnosis of age-related cataract is of clinical significance.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of cataracts include:
· Clouded, blurred or dim vision
· Increasing difficulty with vision at night
· Sensitivity to light and glare
· Need for brighter light for reading and other activities
· Seeing "halos" around lights
· Frequent changes in eyeglass or contact lens prescription
· Fading or yellowing of colors
· Double vision in a single eye

RISK FACTOR

Factors that increase your risk of cataracts include:
· Increasing age
· Diabetes
· Excessive exposure to sunlight
· Smoking
· Obesity
· High blood pressure
· Previous eye injury or inflammation
· Previous eye surgery
· Prolonged use of corticosteroid medications
· Drinking excessive amounts of alcohol

COMPLICATIONS

No studies have proved how to prevent cataracts or slow the progression of cataracts. But doctors think several strategies may be helpful, including:
· Have regular eye examinations
· Quit smoking
· Manage other health problems
· Choose a healthy diet that includes plenty of fruits and vegetables
· Wear sunglasses
· Reduce alcohol consumption

Gene	Your Genotype	Your Result	Explanation
WRN	AA	Normal Risk	When the A>G mutation occurs at this site, the risk of disease increases.

Health Risk

Age-related Macular Degeneration

Age-related macular degeneration (AMD) is a natural consequence of aging with the deterioration of the macular area of the retina. It is one of the leading causes of irreversible blindness in the elderly. This type of eye disease mainly damages the central vision, which causes darkness, dark spots and image distortion in your vision, but does not cause pain. To date, there is no specific treatment for AMD, therefore prevention is important for this disease.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Macular degeneration symptoms usually appear suddenly and worsen rapidly. They may include:
· Visual distortions, such as straight lines seeming bent
· Reduced central vision in one or both eyes
· Decreased intensity or brightness of colors
· A well-defined blurry spot or blind spot in your field of vision
· A general haziness in your overall vision
· Abrupt onset and rapid worsening of symptoms

RISK FACTOR

Factors that may increase your risk of macular degeneration include:
· Age of over 50
· Family history
· Smoking
· Obesity
· Cardiovascular disease

COMPLICATIONS

People whose wet macular degeneration has progressed to central vision loss have a higher risk of depression and social isolation. With profound loss of vision, people may see visual hallucinations (Charles Bonnet syndrome).

Gene	Your Genotype	Your Result	Explanation
ARMS2	TG	Higher Risk	The G>T mutation of this site may cause oxidative damage and apoptosis and eventually promote the occurrence of AMD.
CFH	TT	Normal Risk	Variant that carries the C allele may cause an over-reactive immune response, attack normal cells and promote AMD.

Health Risk

Myopia

Myopia (near-sightedness) is a common eye disorder that occurs when the shape of the eye causes light to refract incorrectly, focusing images in front of, instead of on, the retina. As a result, near objects can be seen clearly, and distant objects look blurred.

1

variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Myopia symptoms may include:
· Blurry vision when looking at distant objects
· Needing to squint to see clearly
· Headaches caused by eyestrain
· Difficulty seeing while driving a vehicle, especially at night (night myopia)

RISK FACTORS

Certain risk factors may increase the likelihood of developing myopia, such as:
· Genetics
· Environmental conditions: focus on close objects for a long period of time, greater time spent indoors.

COMPLICATIONS

Nearsightedness is associated with a variety of complications from mild to severe, such as:
· Reduced quality of life
· Eyestrain
· Other eye problems: retinal detachment, glaucoma, cataracts and myopic maculopathy

Gene	Your Genotype	Your Result	Explanation
BLID	AG	Higher Risk	Individuals with AG and GG genotypes of this site tend to have a higher genetic risk of myopia.

Health Risk

Hyperopia

Hyperopia (farsightedness) is a common eye disorder that occurs when the shape of the eye causes light to refract incorrectly, focusing images behind of, instead of on, the retina. As a result, distant objects can be seen clearly, and near objects look blurred.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Hyperopia symptoms may include:
· Trouble focusing on nearby objects
· Needing to squint to see clearly
· Having eyestrain, including burning eyes, and aching in or around the eyes
· Fatigue or headache after doing close-up tasks, such as reading, writing or computer work, for a period of time

RISK FACTORS

Risk factor of developing hyperopia:
· Genetics

COMPLICATIONS

Hyperopia can be associated with several problems, such as:
· Crossed eyes
· Lazy eyes
· Eyestrain

Gene	Your Genotype	Your Result	Explanation
TOX	CC	Normal Risk	Individuals with AA and AC genotypes of this gene tend to have a higher genetic risk of hyperopia.
GJD2	AC	Higher Risk	Individuals with AA and AC genotypes of this gene tend to have a higher genetic risk of hyperopia.

Health Risk

Corneal Astigmatism

Astigmatism is a disorder that occurs when the surface of the eye (cornea) is unevenly curved. When that happens, light entering the eye is not correctly focused on the retina. Instead, multiple focal points occur resulting in an unclear image. This causes blurred vision at all distances.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of astigmatism may include:
· Blurred or distorted vision
· Eyestrain or discomfort
· Headaches
· Difficulty with night vision
· Squinting

RISK FACTORS

Risk factor of developing astigmatism:
· Genetics
· Eye surgery such as cataract removal
· Corneal scarring or thinning
· A history of near-sightedness or farsightedness

COMPLICATIONS

Astigmatism can be associated with several problems, such as:
· Lazy eyes
· Keratoconus

Gene	Your Genotype	Your Result	Explanation
PDGFRA	CC	Normal Risk	Reports indicate that individuals with TT and TC genotypes of this gene tend to have a higher genetic risk of corneal astigmatism.

Health Risk

Diabetic Retinopathy

Diabetic retinopathy is an eye condition that can cause vision loss and blindness in people who have diabetes. High blood sugar levels is the cause of it, damaging blood vessels in the retina that plays a vital role in vision. The condition can develop in anyone who has type 1 or type 2 diabetes.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Diabetic retinopathy symptoms may include:
- Spots or dark strings floating in the vision (floaters)
- Blurred vision
- Fluctuating vision
- Impaired color vision
- Dark or empty areas in the vision
- Vision loss

RISK FACTORS

Risk of developing the eye condition can increase as a result of:
- Duration of diabetes — the longer the diabetes condition, the greater the risk of developing diabetic retinopathy
- Poor control of blood sugar level
- High blood pressure
- High cholesterol
- Pregnancy
- Tobacco use

COMPLICATIONS

Diabetic retinopathy involves the abnormal growth of blood vessels in the retina. Complications can lead to serious vision problems:
- Vitreous hemorrhage
- Retinal detachment
- Glaucoma
- Blindness

Gene	Your Genotype	Your Result	Explanation
TBX18	CC	Normal Risk	Individuals with TT and TC genotypes of this gene tend to have a higher genetic risk of diabetic retinopathy.
MYSM1	CC	Normal Risk	Individuals with TT genotype of this gene tend to have a higher genetic risk of diabetic retinopathy.
PLXDC2	AA	Normal Risk	Individuals with GG genotype of this gene tend to have a higher genetic risk of diabetic retinopathy.

Health Risk

Vogt-Koyanagi-Harada Disease

Vogt-Koyanagi-Harada disease (VKH disease) affects the eyes, ears, nervous system, and skin. The exact cause of VKH disease is unknown, but the symptoms are thought to be due to an abnormal response of the immune system to a viral infection. Genetic factors may be involved.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

In the early phase, symptoms may include:
· Headache
· Deep pain in the eyes
· Dizziness (vertigo)
· Nausea and vomiting
· Hearing loss
· Ringing in the ears (tinnitus)

In next phase, symptoms may include:
· Blurry vision in one or both eyes
· Inflammation of the eyes (uveitis)
· Floating spots in the vision that are signs of retinal detachment

The convalescent phase usually occurs a few weeks to months after the uveitis phase. In this phase, symptoms may include:
· Vitiligo
· Patches of white hair, eyelashes, and eyebrows (poliosis)
· Hair loss

The recurrent phase occurs in about half of the people with VKH disease. Symptoms may include:
· Cataracts
· Glaucoma
· Abnormal blood vessels growth under the retina (choroidal neovascularization)

RISK FACTORS

Risk factors include genetics and it affects more pigmented groups such as Hispanics, Asians, Native Americans, Middle Easterners, and Asian Indians

COMPLICATIONS

The most common complications are:
· Cataracts
· Glaucoma
· Choroidal Neovascularization
· Subretinal Fibrosis
· Choroidal Atrophy
· Posterior Synechiae
· Optic Atrophy

Gene	Your Genotype	Your Result	Explanation
IL23R	AA	Normal Risk	Individuals with AG and GG genotypes tend to have a higher genetic risk of Vogt-Koyanagi-Harada syndrome.
HLA-DRB1	GG	Higher Risk	Individuals with CG and GG genotypes tend to have a higher genetic risk of Vogt-Koyanagi-Harada syndrome.
ZNF365	CC	Normal Risk	Individuals with CT and TT genotypes tend to have a higher genetic risk of Vogt-Koyanagi-Harada syndrome.

HEALTH RISK

SKIN ISSUES

Health Risk

Atopic Dermatitis

Atopic dermatitis is a chronic inflammatory skin disease associated with inherited allergen. It is characterized by itching and skin redness. This disease is most common in infants and young children but can occur at any age.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Atopic dermatitis (eczema) signs and symptoms vary widely from person to person and include:
- Dry skin
- Itching, which may be severe, especially at night
- Red to brownish-gray patches, especially on the hands, feet, ankles, wrists, neck, upper chest, eyelids, inside the bend of the elbows and knees, and in infants, the face and scalp
- Small, raised bumps, which may leak fluid and crust over when scratched
- Thickened, cracked, scaly skin
- Raw, sensitive, swollen skin from scratching

CAUSES

Healthy skin helps retain moisture and protects you from bacteria, irritants and allergens. Eczema is related to a gene variation that affects the skin's ability to provide this protection. This allows your skin to be affected by environmental factors, irritants and allergens. In some children, food allergies may play a role in causing eczema.

RISK FACTORS

The primary risk factor for atopic dermatitis is having a personal or family history of eczema, allergies, hay fever or asthma.

COMPLICATIONS

Complications of atopic dermatitis (eczema) may include:
- Asthma and hay fever
- Chronic itchy, scaly skin
- Skin infections
- Irritant hand dermatitis
- Allergic contact dermatitis
- Sleep problems

Atopic Dermatitis

Gene	Your Genotype	Your Result	Explanation
ZNF365	TT	Normal Risk	When this site carries C allele, the risk of disease increases.
MHC	AA	Higher Risk	When this site carries A allele, the risk of disease increases.
IL13	CC	Normal Risk	When this site carries A allele, the risk of disease increases.
HLA-B	GG	Higher Risk	When this site carries G allele, the risk of disease increases.
C6orf10	GG	Higher Risk	When this site carries G allele, the risk of disease increases.
GLB1	AG	Moderate Risk	G>A mutation occurring at this site can increase the risk of disease.
Intergenic	TT	Normal Risk	When this site carries C allele, the risk of disease increases.
Intergenic	CC	Normal Risk	C>T mutation occurring at this site can increase the risk of disease.

Health Risk

Psoriasis

Psoriasis can develop at any age. It is mainly characterized by red, flaky, crusty patches of skin covered with silvery scales. It is a common chronic skin disease thought to be caused by autoimmune and inflammatory reactions. For those with psoriasis, it could have a significant impact on quality of life and mental well-being.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Psoriasis signs and symptoms are different for everyone. Common signs and symptoms include:
· Red patches of skin covered with thick, silvery scales
· Small scaling spots (commonly seen in children)
· Dry, cracked skin that may bleed
· Itching, burning or soreness
· Thickened, pitted or ridged nails
· Swollen and stiff joints

RISK FACTOR

Anyone can develop psoriasis, but these factors can increase your risk of developing the disease:
· Family history
· Viral and bacterial infections
· Stress
· Obesity
· Smoking

COMPLICATIONS

If you have psoriasis, you're at greater risk of developing certain diseases. These include:
· Psoriatic arthritis
· Eye conditions
· Obesity
· Type 2 diabetes
· Cardiovascular disease
· Metabolic syndrome
· Other autoimmune diseases
· Parkinson's disease
· Kidney disease
· Emotional problems

Gene	Your Genotype	Your Result	Explanation
MHC	GG	Normal Risk	When this variant carries C allele, it may increase the possibility of misidentification of the immune system, thereby increasing the risk of psoriasis.

Health Risk

Keloid

Keloid is an inevitable outcome when the scar tissue undergoes wound healing process. A keloid scar is an enlarged, raised scar that can be pink, red, skin-colored or darker than the surrounding skin. Keloid scars are more common on the upper chest, shoulders, head (especially the earlobes after a piercing) and neck, but they can happen anywhere.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Keloids come from the overgrowth of scar tissue. Keloid scars tend to be larger than the original wound itself. They may take weeks or months to develop fully.
The symptoms of a keloid can include:
· A localized area that is flesh-colored, pink, or red
· A lumpy or ridged area of skin that's usually raised
· An area that continues to grow larger with scar tissue over time
· An itchy patch of skin

CAUSES

Most skin injury types can contribute to scarring. This includes burns, acne scars, chickenpox scars, ear piercing, scratches, surgical incisions, and vaccination sites.
According to the (US) National Center for Biotechnology Information, keloid scarring is common in young people between the ages of 10 and 20. Studies have shown that those with darker complexions are at a higher risk of keloid scarring as a result of skin trauma. They occur in 15 – 20% of individuals with sub-Saharan African, Asian or Latino ancestry, significantly less in those of a Caucasian background and there are no reported cases in patients with albinism. Keloids tend to have a genetic component, which means one is more likely to have keloids if one or both of their parents have them.

Gene	Your Genotype	Your Result	Explanation
Intergenic	CC	Higher Risk	GWAS study found that this site carries a significant correlation with the occurrence of keloid.

Health Risk

Generalized Vitiligo

Vitiligo is a common skin pigmentation disease. This disease is characterized by pale white patches developed on the skin, caused by the lack of melanin - the pigment responsible for skin, hair and eyes color. Depigmentation usually first shows on sun-exposed skin, such as the hands, feet, arms, face and lips.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Vitiligo signs include:
· Patchy loss of skin color
· Premature whitening or graying of the hair on your scalp, eyelashes, eyebrows or beard
· Loss of color in the tissues that line the inside of your mouth and nose (mucous membranes)
· Loss of or change in color of the inner layer of the eyeball (retina)

Depending on the type of vitiligo you have, the discolored patches may cover:
· Many parts of your body. With this most common type, called generalized vitiligo, the discolored patches often progress similarly on corresponding body parts (symmetrically).
· Only one side or part of your body. This type, called segmental vitiligo, tends to occur at a younger age, progress for a year or two, then stop.
· One or only a few areas of your body. This type is called localized (focal) vitiligo.

It is difficult to predict how your disease will progress. Sometimes the patches stop forming without treatment. In most cases, pigment loss spreads and eventually involves most of your skin. Rarely, the skin gets its color back.

COMPLICATIONS

Social or psychological distress, sunburn, skin cancer, eye problems (such as inflammation of the iris (iritis)) and hearing loss.

Gene	Your Genotype	Your Result	Explanation
Intergenic	AA	Normal Risk	Research analysis found that G allele at this locus is related to the early-onset and severe clinical type of vitiligo.
HLA	AG	Normal Risk	Big data analysis found that GG genotype at this locus is related to the early-onset and severe clinical type of vitiligo.
RNASET2	CC	Normal Risk	This variant is located on the RNASET2 gene and by carrying T allele, it may cause an increase in the expression of the RNASET2 gene, thereby increasing the risk of disease.
HLA	CC	Normal Risk	Big data analysis found that the site carries T allele is associated with the early-onset and moderate-to-severe clinical types of vitiligo.
Intergenic	AG	Higher Risk	Big data analysis found that when the site carries G allele, it increases the risk of vitiligo.

HEALTH RISK

RESPIRATORY SYSTEM

Health Risk

Asthma

Bronchial asthma, also known as asthma, is one of the most common chronic diseases worldwide nowadays. It is also one of the most common chronic childhood diseases that typically presents with wheezing, shortness of breath, chest tightness and/or coughing. Maintaining a healthy lifestyle, avoiding environmental triggers and taking the right medications are some of the ways to manage symptoms of asthma.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Asthma symptoms vary from person to person. You may have infrequent asthma attacks, have symptoms only at certain times — such as when exercising — or have symptoms all the time. Some of the symptoms of asthma include:

- Shortness of breath
- Chest tightness or pain
- Trouble sleeping caused by shortness of breath, coughing or wheezing
- A whistling or wheezing sound when exhaling (wheezing is a common sign of asthma in children)
- Coughing or wheezing attacks that are worsened by a respiratory virus, such as a cold or the flu

RISK FACTORS

Certain factors may increase your risk of developing asthma which include:

- Having a blood relative (such as a parent or sibling) with asthma
- Having another allergic condition, such as atopic dermatitis or allergic rhinitis (hay fever)
- Being overweight
- Being a smoker
- Exposure to secondhand smoke
- Exposure to exhaust fumes or other types of pollution
- Exposure to occupational triggers, such as chemicals used in farming, hairdressing and manufacturing

COMPLICATIONS

Asthma may lead to the following complications:

- Signs and symptoms that interfere with sleep, work or recreational activities
- Sick days from work or school during asthma flare-ups
- Permanent narrowing of the bronchial tubes (airway remodeling) that affects how well you can breathe
- Emergency room visits and hospitalizations for severe asthma attacks
- Side effects from long-term use of some medications used to stabilize severe asthma

Gene	Your Genotype	Your Result	Explanation
SLC6A7	CC	Higher Risk	CC genotype at this site is associated with an increased risk of asthma.
ORMDL3	CC	Normal Risk	TT genotype at this site is associated with an increased risk of asthma in children and young adult.

Health Risk

Allergic Rhinitis

People with allergic rhinitis, also called hay fever, develop cold-like signs and symptoms, such as a runny nose, itchy eyes, congestion, sneezing and sinus pressure. But unlike a cold, allergic rhinitis is not caused by a virus. It is caused by an allergic response to environmental allergens, such as pollen, dust mites, or tiny flecks of skin and saliva shed by cats, dogs, and other animals with fur or feathers (pet dander).

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common symptoms of allergic rhinitis include:
· Sneezing, runny, stuffy and itchy nose
· Coughing, sore or scratchy throat
· Itchy, watery and dark circles under the eyes
· Frequent headaches
· Eczema-type symptoms, such as having extremely dry, itchy skin that can blister and weep
· Hives
· Excessive fatigue

RISK FACTORS

The primary risk factor for allergic rhinitis is having a personal or family history of asthma, eczema and food allergies. These can be caused by genetics. Allergic rhinitis most often appears at childhood and it may not last into adulthood. If allergic rhinitis started after age of 20, it may last through middle age.

COMPLICATIONS

Unfortunately, allergic rhinitis itself cannot be prevented. Treatment and management are keys to achieving a good quality of life with allergies. Some complications that can arise from hay fever include:
· Inability to sleep from symptoms keeping you up at night
· Development or worsening of asthma symptoms
· Frequent ear infections
· Sinusitis or frequent sinus infections
· Absences from school or work because of reduced productivity
· Frequent headaches

Gene	Your Genotype	Your Result	Explanation
HLA	TT	Normal Risk	When T>C mutation occurs at this site, the risk of disease increases.

Health Risk

Chronic Obstructive Pulmonary Disease

Chronic obstructive pulmonary disease (COPD) is an inflammatory lung disease. Its condition is related to the exposure to harmful gases and particles. It can further develop into pulmonary heart disease and respiratory failure with high disability and mortality rate. Although environmental toxins and pollution can also cause chronic obstructive pulmonary disease, such conditions are often associated with long-term smoking. Therefore, smoking cessation (quit smoking) reduces the risk of chronic obstructive pulmonary disease.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

COPD symptoms often do not appear until significant lung damage has occurred, and they usually worsen over time, particularly if smoking exposure continues. For chronic bronchitis, the main symptom is a daily cough and mucus (sputum) production at least three months a year for two consecutive years. Other signs and symptoms of COPD may include:

· Shortness of breath, especially during physical activities
· Wheezing
· Chest tightness
· Having to clear your throat first thing in the morning, due to excess mucus in your lungs
· A chronic cough that may produce mucus (sputum) that may be clear, white, yellow or greenish
· Blueness of the lips or fingernail beds (cyanosis)
· Frequent respiratory infections
· Lack of energy
· Unintended weight loss (in later stages)
· Swelling in ankles, feet or legs

RISK FACTORS

Certain factors may increase your risk of developing chronic obstructive pulmonary disease which include:

· Exposure to tobacco smoke
· People with asthma who smoke
· Occupational exposure to dusts and chemicals
· Exposure to fumes from burning fuel
· Genetics

COMPLICATIONS

COPD can cause many complications, including:

· Respiratory infections
· Heart problems
· Lung cancer
· High blood pressure in lung arteries
· Depression

Gene	Your Genotype	Your Result	Explanation
FAM13A	CC	Normal Risk	Studies have shown that C>T mutation may be associated with changes in FEV1/FVC and susceptibility to COPD in the general population.
HYKK	TT	Normal Risk	Studies have shown that T>C mutation may be associated with COPD in the general population.

Health Risk

Pulmonary Fibrosis

Pulmonary fibrosis is a condition in which the lungs become scarred and damaged. As the disease progresses, it gets more difficult for your lungs to work properly, and breathing becomes increasingly difficult.

4 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of pulmonary fibrosis may include:
· Shortness of breath (dyspnea)
· A dry cough
· Fatigue
· Unexplained weight loss
· Aching muscles and joints
· Widening and rounding of the tips of the fingers or toes (clubbing)

RISK FACTORS

Factors that make you more susceptible to pulmonary fibrosis include:
· Age: the disorder is much more likely to affect middle-aged and older adults
· Gender: idiopathic pulmonary fibrosis is more likely to affect men than women
· Smoking
· Having radiation treatments to your chest or using certain chemotherapy drugs can increase your risk of pulmonary fibrosis
· Genetic factors

COMPLICATIONS

Complications of pulmonary fibrosis may include:
· High blood pressure in your lungs (pulmonary hypertension)
· Right-sided heart failure (cor pulmonale)
· Respiratory failure
· Lung cancer
· Lung complications

Gene	Your Genotype	Your Result	Explanation
TERT	AC	Higher Risk	Studies have shown that C>A mutations increase the risk of idiopathic pulmonary fibrosis.
DPP9	AG	Higher Risk	Studies have shown that A>G mutations in this position increases the risk of pulmonary fibrosis.
DSP	TG	Higher Risk	Studies have shown that T>G mutations at this site increase the risk of idiopathic pulmonary fibrosis.
ATP11A	GG	Higher Risk	Studies have shown that A>G mutation in this location increases the risk of pulmonary fibrosis.

Health Risk

Emphysema

Emphysema is a lung condition that causes shortness of breath. In people with emphysema, the air sacs in the lungs are damaged. Over time, the inner walls of the air sacs weaken and rupture — creating larger air spaces instead of many small ones. This reduces the surface area of the lungs and the amount of oxygen that reaches bloodstream.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The main symptom of emphysema is shortness of breath, which usually begins gradually. Other signs and symptoms include:

- Wheezing
- Ongoing feeling of not being able to get enough air
- Long-term cough
- Long-term mucus production

RISK FACTORS

Factors that increase the risk of developing emphysema include:

- Smoking
- Age: most people experience the symtoms between the ages of 40-60
- Exposure to secondhand smoke
- Occupational exposure to fumes or dust
- Exposure to indoor and outdoor pollution

COMPLICATIONS

People who have emphysema are more likely to develop:

- Collapsed lung
- Heart problems
- Large holes in the lungs (bullae)

Gene	Your Genotype	Your Result	Explanation
SERPINE2	TT	Normal Risk	Reports indicate that individuals with the CC genotype tend to have a higher genetic risk of pulmonary emphysema.

Health Risk

Obstructive Sleep Apnea

Obstructive sleep apnea causes breathing to repeatedly stop and start during sleep. It occurs when the throat muscles intermittently relax and block the airway during sleep. A noticeable sign of obstructive sleep apnea is snoring.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of obstructive sleep apnea include:
· Excessive daytime sleepiness
· Loud snoring
· Observed episodes of stopped breathing during sleep
· Abrupt awakenings accompanied by gasping or choking
· Awakening with a dry mouth or sore throat
· Morning headache
· Difficulty concentrating during the day
· Experiencing mood changes
· High blood pressure
· Nighttime sweating
· Decreased libido

RISK FACTORS

Factors that increase the risk of developing obstructive sleep apnea include:
· Excess weight
· Narrowed airway
· Hypertension
· Chronic nasal congestion
· Smoking
· Diabetes
· Genetics
· Asthma
· Gender: male are more likely to develop this disease than women

COMPLICATIONS

Obstructive sleep apnea is considered a serious medical condition. Complications may include:
· Daytime tiredness
· Cardiovascular problems
· Complications with medications and surgery

Gene	Your Genotype	Your Result	Explanation
NRG1	AG	Normal Risk	Reports indicate that individuals with AG and GG genotypes tend to have a lower genetic risk of obstructive sleep apnea.

HEALTH RISK

NERVOUS SYSTEM

Health Risk

Alzheimer's Disease

Alzheimer's disease, a common cause of dementia, is an irreversible, progressive brain disorder that slowly destroys memory and thinking skills, and, eventually, the ability to carry out the simplest tasks. In most people with Alzheimer's, symptoms first appear in their mid-60s.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Memory loss is the key symptom of Alzheimer's disease. An early sign of the disease is usually difficulty remembering recent events or conversations. As the disease progresses, memory impairments worsen and other symptoms develop. At first, a person with Alzheimer's disease may be aware of having difficulty with remembering things and organizing thoughts. A family member or friend may be more likely to notice how the symptoms worsen.

RISK FACTOR

Risk factors for Alzheimer's disease include:
· Age
· Family history and genetics
· Down syndrome
· Mild cognitive impairment
· Past head trauma
· Lifestyle and heart health

COMPLICATIONS

Memory and language loss, impaired judgment, and other cognitive changes caused by Alzheimer's can complicate treatment for other health conditions. A person with Alzheimer's disease may not be able to:
· Communicate that he or she is experiencing pain — for example, from a dental problem
· Report symptoms of another illness
· Follow a prescribed treatment plan
· Notice or describe medication side effects

As Alzheimer's disease progresses to its last stages, brain changes begin to affect physical functions, such as swallowing, balance, and bowel and bladder control. These effects can increase vulnerability to additional health problems such as:
· Inhaling food or liquid into the lungs (aspiration)
· Pneumonia and other infections
· Falls
· Fractures
· Bedsores
· Malnutrition or dehydration

Gene	Your Genotype	Your Result	Explanation
APOE	TT	Normal Risk	This site that carries C allele may accelerate the beta-amyloid deposition while slowing down its degradation. It may cause neurofibrillary tangles, which increases the risk of disease.

Health Risk

Parkinson's Disease

Parkinson's disease is a neurological disease with a high incidence among elderly over 60 years old. Symptoms in the early stages of the disease usually manifest as tremor in one limb, lack of facial expression, soft or slurred speech and stiff or slowed movement.

1 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Parkinson's disease signs and symptoms can be different for everyone. Early signs may be mild and go unnoticed. Symptoms often begin on one side of your body and usually remain worse on that side, even after symptoms begin to affect both sides. Parkinson's signs and symptoms may include:

· Tremor
· Slowed movement (bradykinesia)
· Rigid muscles
· Impaired posture and balance
· Loss of automatic movements
· Speech changes
· Writing changes

RISK FACTOR

Risk factors for Parkinson's disease include:

· Age: people usually develop the disease around age 60 or older
· Heredity
· Gender: men are more likely to develop Parkinson's disease than are women
· Exposure to toxins

COMPLICATIONS

Parkinson's disease is often accompanied by these additional problems, which may be treatable:

· Thinking difficulties
· Depression and emotional changes
· Swallowing problems
· Chewing and eating problems
· Sleep problems and sleep disorders
· Bladder problems
· Constipation

Gene	Your Genotype	Your Result	Explanation
LRRK2	GG	Normal Risk	A allele promotes neuronal necrosis and increases inflammatory response, thereby increasing the risk of Parkinson's disease.
SCNA	TT	Higher Risk	When this variant site is TT genotype, it may lead to structural changes in alpha-synuclein, thereby increasing the risk of Parkinson's disease.

Health Risk

Multiple Sclerosis

Multiple sclerosis (MS) is an autoimmune disease involving the brain and spinal cord (central nervous system). It causes a wide range of potential symptoms, including problems with vision, arm or leg movement, sensation or balance. It is a lifelong condition that can sometimes cause serious disability, although it can occasionally be mild. Although MS cannot currently be cured, there are some treatments that can delay the progression of the disease and improve the quality of life of patients.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Symptoms often affect movement, such as:
- Numbness or weakness in one or more limbs that typically occurs on one side of your body at a time, or the legs and trunk
- Electric-shock sensations that occur with certain neck movements, especially bending the neck forward (Lhermitte sign)
- Tremor, lack of coordination or unsteady gait

Vision problems are also common, including:
- Partial or complete loss of vision, usually in one eye at a time, often with pain during eye movement
- Prolonged double vision
- Blurry vision

Other symptoms for MS:
- Slurred speech
- Fatigue
- Dizziness
- Tingling or pain in parts of your body
- Problems with sexual, bowel and bladder function

RISK FACTOR

These factors may increase your risk of developing multiple sclerosis:
- Age: usually affects people between the ages of 16 and 55
- Family history
- Certain infections
- Having low levels of vitamin D and low exposure to sunlight is associated with a greater risk of MS
- Certain autoimmune diseases: thyroid disease, type 1 diabetes or inflammatory bowel disease
- Smoking

COMPLICATIONS

People with multiple sclerosis may also develop:
- Muscle stiffness or spasms
- Paralysis, typically in the legs
- Problems with bladder, bowel or sexual function
- Mental changes, such as forgetfulness or mood swings
- Depression
- Epilepsy

Gene	Your Genotype	Your Result	Explanation
HLA-DRA	GG	Normal Risk	Variant site carrying A allele is associated with the increase in the number of immunoglobulin IgG that may lead to an increased risk of multiple sclerosis.
IL7R	CC	Higher Risk	The CC genotypes at this site may result in a decrease in lymphocyte proliferation that increase the risk of disease.

Health Risk

Glioma

Glioma is a common type of tumor originating in the brain. About 33 percent of all brain tumors are gliomas, which originate in the glial cells that surround and support neurons in the brain.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common signs and symptoms of gliomas include:
· Headaches
· Nausea or vomiting
· Confusion or a decline in brain function
· Memory loss
· Personality changes or irritability
· Difficulty with balance
· Vision problems
· Speech difficulties
· Seizures

RISK FACTORS

The exact cause of gliomas is not known. But there are some factors that may increase your risk of a brain tumor. Risk factors include:
· Age: most common in adults between ages 45 and 65 years old
· Exposure to radiation
· Genetics

COMPLICATIONS

Complications of gliomas include:
· Chest pain
· Erectile dysfunction
· Loss of muscle control
· Loss of bowel or bladder control
· Numbness
· Weakness in the limbs or upper body

Gene	Your Genotype	Your Result	Explanation
TERT	AC	Higher Risk	Reports indicate that individuals with CC and AC genotypes tend to have a higher genetic risk of glioma.
CCDC26	TT	Normal Risk	Reports indicate that individuals with GT and GG genotypes tend to have a higher genetic risk of glioma.
CDKN2B-AS1	AA	Normal Risk	Reports indicate that individuals with GA and GG genotypes tend to have a higher genetic risk of glioma.
PHLDB1	AG	Normal Risk	Reports indicate that individuals with GT and GG genotypes tend to have a higher genetic risk of glioma.

Health Risk

Migraine

A migraine can cause severe throbbing pain or a pulsing sensation, usually on one side of the head. It is often accompanied by nausea, vomiting, and extreme sensitivity to light and sound. Migraine attacks can last for hours to days, and the pain can be so severe that it interferes with daily activities.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common signs and symptoms of migraine include:
· Pain usually on one side of the head
· Pain that throbs or pulses
· Sensitivity to light, sound, and sometimes smell and touch
· Nausea and vomiting

RISK FACTORS

Factors that increase the risk of developing migraines include:
· Genetics
· Age: migraines tend to peak during 30s
· Gender: women are three times more likely to have migraines
· Hormonal changes

COMPLICATIONS

Complications of migraines include:
· Migrainous infarction (stroke)
· Persistent aura without infarction
· Migraine-triggered seizure
· Depression and anxiety
· Vertigo
· Nausea and vomiting
· Sleeplessness

Gene	Your Genotype	Your Result	Explanation
PRDM16	TT	Normal Risk	Reports indicate that individuals with AC and CC genotypes tend to have a higher genetic risk of migraine.
TSPAN2	CC	Normal Risk	Reports indicate that individuals with AC and AA genotypes tend to have a higher genetic risk of migraine.
MEF2D	AA	Normal Risk	Reports indicate that individuals with AC and CC genotypes tend to have a higher genetic risk of migraine.
TRPM8	AG	Normal Risk	Reports indicate that individuals with GG genotype tend to have a higher genetic risk of migraine.
PHACTR1	AG	Normal Risk	Reports indicate that individuals with AA genotype tend to have a higher genetic risk of migraine.
SUGCT	TC	Higher Risk	Reports indicate that individuals with TC and TT genotypes tend to have a higher genetic risk of migraine.
ASTN2	GG	Higher Risk	Reports indicate that individuals with AG and GG genotypes tend to have a higher genetic risk of migraine.
LRP1	TT	Higher Risk	Reports indicate that individuals with TT genotype tend to have a higher genetic risk of migraine.

Health Risk

Essential Tremor

Essential tremor is a nervous system (neurological) disorder that causes involuntary and rhythmic shaking. It can affect almost any part of the body, but the trembling occurs most often in hands — especially when doing simple tasks, such as drinking from a glass or tying shoelaces.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Essential tremor signs and symptoms:
· Begin gradually, usually more prominently on one side of the body
· Worsen with movement
· Usually occur in the hands first, affecting one hand or both hands
· Aggravated by emotional stress, fatigue, caffeine or temperature extremes

RISK FACTORS

Known risk factors for essential tremor include:
· Genetics
· Age: more common in people age 40 and older

COMPLICATIONS

If the tremors become severe, patients might find it difficult to:
· Hold a cup or glass without spilling
· Eat normally
· Put on makeup or shave
· Talk, if the voice box or tongue is affected
· Write legibly

Gene	Your Genotype	Your Result	Explanation
LRRK2	GG	Normal Risk	Reports indicate that individuals with GC and CC genotypes tend to have a higher genetic risk of essential tremor.

HEALTH RISK

ENDOCRINE SYSTEM

Health Risk

Hypothyroidism

An underactive thyroid gland (hypothyroidism) is where your thyroid gland does not produce enough hormones. Common signs of an underactive thyroid are tiredness, weight gain and feeling depressed. An underactive thyroid can often be successfully treated by taking daily hormone tablets to replace the hormones your thyroid is not making. There is no way of preventing hypothyroidism. Most cases are caused either by the immune system attacking the thyroid gland and damaging it, or by damage to the thyroid that occurs during some treatments for an overactive thyroid or thyroid cancer.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The signs and symptoms of hypothyroidism vary, depending on the severity of the hormone deficiency. Problems tend to develop slowly, often over a number of years.

Hypothyroidism signs and symptoms may include:
· Fatigue
· Increased sensitivity to cold
· Constipation
· Dry skin
· Weight gain
· Muscle weakness, aches, tenderness and stiffness
· Heavier than normal or irregular menstrual periods
· Thinning hair
· Slowed heart rate
· Enlarged thyroid gland (goiter)

RISK FACTOR

Although anyone can develop hypothyroidism, you're at an increased risk if you:
· Are a woman
· Are older than 60
· Have a family history of thyroid disease
· Have an autoimmune disease, such as type 1 diabetes or celiac disease
· Have been treated with radioactive iodine or anti-thyroid medications
· Received radiation to your neck or upper chest
· Have had thyroid surgery (partial thyroidectomy)
· Have been pregnant or delivered a baby within the past six months

COMPLICATIONS

Untreated hypothyroidism can lead to a number of health problems such as goiter, heart problems, mental health issues, peripheral neuropathy, myxedema, infertility and birth defects.

Gene	Your Genotype	Your Result	Explanation
VAV3	TT	Normal Risk	When the T>C mutation occurs at this site, the risk of disease increases.
HLA	GG	Higher Risk	When the site carries the risk gene G allele, the risk of disease increases.
SH2B3	CC	Normal Risk	When the site carries T allele, the disease risk increases.

Health Risk

Graves' disease

Graves' disease is an autoimmune disease. It results in the overproduction of thyroid hormones (hyperthyroidism). Because thyroid hormones affect a number of different body systems, signs and symptoms associated with Graves' disease can be wide ranging and significantly influence your overall well-being.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Graves' disease signs and symptoms include:
· Anxiety and irritability
· A fine tremor of your hands or fingers
· Heat sensitivity and an increase in perspiration or warm, moist skin
· Weight loss, despite normal eating habits
· Enlargement of your thyroid gland (goiter)
· Change in menstrual cycles
· Erectile dysfunction or reduced libido
· Frequent bowel movements
· Bulging eyes (Graves' ophthalmopathy)
· Fatigue
· Thick, red skin usually on the shins or tops of the feet (Graves' dermopathy)
· Rapid or irregular heartbeat (palpitations)

RISK FACTOR

These factors might increase your risk of developing this condition: family history, age (Graves' disease usually develops in people younger than 40), gender (women are much more likely to develop Graves' disease than are men), presence of other autoimmune disorders (people with other disorders of the immune system, such as type 1 diabetes or rheumatoid arthritis, have an increased risk), emotional or physical stress, pregnancy and smoking.

COMPLICATIONS

Graves' disease can cause complications including pregnancy issues, heart disorders, thyroid storm and brittle bones.

Gene	Your Genotype	Your Result	Explanation
SLAMF6	AA	Normal Risk	The study found that when this site carries C allele, the risk of disease increases.
HLA-B	TT	Higher Risk	It is found that the C>T mutation occurs at this site can increase the risk of illness.
ABO	TC	Normal Risk	Related studies have found that TT homozygotes at this site increases the risk of disease in people with type O-blood.
TG	GG	Normal Risk	In vitro studies have shown that the AA genotype in this gene locus can influence the splicing of TG, leading to the increased expression of non-e46 TG isoform.
C1QTNF6	CC	Higher Risk	Large number of literatures showed that this site that carrying C allele is associated with the disease risk of GD.

Health Risk

Type 2 Diabetes

Type 2 diabetes is a common condition that causes the level of sugar (glucose) in the blood to become too high. It is caused by problems with a chemical in the body (hormone) called insulin. It is often linked to being overweight or inactive, or having a family history of type 2 diabetes. It is a lifelong condition that can affect your everyday life. You may need to change your diet, take medicines and have regular check-ups.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of type 2 diabetes often develop slowly. In fact, you can have type 2 diabetes for years and not know it. Look for:

- Increased thirst
- Frequent urination
- Increased hunger
- Unintended weight loss
- Fatigue
- Blurred vision
- Slow-healing sores
- Frequent infections
- Areas of darkened skin, usually in the armpits and neck

RISK FACTORS

Being overweight, accumulation of abdominal fat, inactivity, family history, age, prediabetes and medical history of gestational diabetes are some of the risk factors of type 2 diabetes.

COMPLICATIONS

Although long-term complications of diabetes develop gradually, they can eventually be disabling or even life-threatening. Some of the potential complications of diabetes include:

- Heart and blood vessel disease
- Nerve damage (neuropathy)
- Kidney damage
- Eye damage
- Slow wound healing
- Hearing impairment
- Skin conditions
- Sleep apnea
- Alzheimer's disease

Gene	Your Genotype	Your Result	Explanation
ADIPOQ	TG	Higher Risk	The gene locus carrying T allele can reduce adiponectin expression, reduce insulin sensitivity and increase the risk of diabetes.
KCNQ1	TC	Higher Risk	The gene locus carrying C allele can cause continuous opening of the potassium channel that reduces insulin secretion and increases the risk of diabetes.
TCF7L2	CC	Normal Risk	The C>T mutation at this gene locus may result in a decrease in GLP-1 secretion leading to a decrease in insulin secretion, which in turn increases the risk of type 2 diabetes.

HEALTH RISK

CARDIOVASCULAR SYSTEM

Health Risk

Hypertension

Hypertension, or high blood pressure, rarely has noticeable symptoms. But if untreated, it increases your risk of serious problems such as heart attacks and strokes. Hence, it is important to monitor your blood pressure routinely. Ask your doctor for a blood pressure reading at least every two years starting at age 18. If you are age 40 or older, or you are 18 to 39 with a high risk of high blood pressure, ask your doctor for a blood pressure reading every year.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Most people with high blood pressure have no signs or symptoms, even if blood pressure readings reach dangerously high levels. A few people with high blood pressure may have headaches, shortness of breath or nosebleeds, but these signs and symptoms are not specific and usually do not occur until high blood pressure has reached a severe or life-threatening stage.

RISK FACTORS

Certain factors may increase your risk of developing hypertension which include:
· Age: risk of high blood pressure increases as you age
· Family history: high blood pressure tends to run in families
· Being overweight or obese or not being physically active
· Using tobacco or drinking too much alcohol
· Too much salt (sodium) in your diet
· Too little potassium in your diet. If you do not get enough potassium in your diet or retain enough potassium, you may accumulate too much sodium in your blood
· Stress
· Certain chronic conditions such as kidney disease, diabetes and sleep apnea

COMPLICATIONS

The excessive pressure on your artery walls caused by high blood pressure can damage your blood vessels, as well as organs in your body. The higher your blood pressure and the longer it goes uncontrolled, the greater the damage. Uncontrolled high blood pressure can lead to complications including heart attack, stroke, aneurysm, heart failure; weakened and narrowed blood vessels in your kidneys; thickened, narrowed or torn blood vessels in the eyes; metabolic syndrome; trouble with memory or understanding and dementia.

Gene	Your Genotype	Your Result	Explanation
CYP17A1	TC	Normal Risk	The TT genotype at this gene locus can impair steroid hormone synthesis, which indirectly affects salt metabolism.
NEDD4L	TC	Higher Risk	The gene locus carrying T allele can increase sodium reabsorption and increase the risk of hypertension.
NEDD4L	GG	Normal Risk	The gene locus carrying A allele can increase sodium reabsorption and increase the risk of hypertension.
FGF5	TT	Higher Risk	GWAS study found that this site was significantly associated with the occurrence of hypertension.

Health Risk

Coronary Heart Disease

Coronary heart disease, also known as ischemic heart disease, is a type of heart disease caused by coronary atherosclerosis (plaque buildup inside the artery walls) leading to narrowing or clogging blood vessels, resulting in hypoxia (low oxygen in the blood) or necrosis (death) of the heart muscle. The onset of disease in male is earlier than female and it is a common disease in life.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

If your coronary arteries narrow, they cannot supply enough oxygen-rich blood to your heart — especially when it is beating hard, such as during exercise. At first, the decreased blood flow may not cause any coronary artery disease symptoms. As plaque continues to build up in your coronary arteries, however, you may develop coronary artery disease signs and symptoms, including:

· Chest pain (angina)
· Shortness of breath
· Heart attack

RISK FACTORS

Certain factors may increase your risk of developing coronary heart disease which include:

· Age: simply getting older increases your risk of damaged and narrowed arteries
· Gender: men are generally at greater risk of coronary artery disease. However, the risk for women increases after menopause
· Family history
· Smoking
· High blood pressure or high blood cholesterol levels
· Diabetes
· Overweight or obesity
· Physical inactivity
· High stress
· Unhealthy diet

COMPLICATIONS

Possible complications include chest pain (angina), heart attack, heart failure and abnormal heart rhythm (arrhythmia).

Gene	Your Genotype	Your Result	Explanation
CDKN2A/B	CC	Higher Risk	The CC genotype may promote cell proliferation, cause atherosclerosis and increase the likelihood of coronary heart disease.
CDKN2A/B	GG	Higher Risk	The GG genotype may promote cell proliferation, cause atherosclerosis and increase the likelihood of coronary heart disease.
APOA5	AG	Higher Risk	Gene locus that carries G allele may cause dyslipidemia and increase the risk of coronary heart disease.
LPA	TT	Normal Risk	The T>C mutation on this gene may increase serum lipoprotein levels, promote atherosclerosis and increase the likelihood of coronary heart disease.
ADTRP	GG	Normal Risk	GWAS study found that this site carries a significant correlation with the occurrence of coronary heart disease.

Health Risk

Acute Myocardial Infarction

Acute myocardial infarction or heart attack is a serious medical emergency in which the supply of blood to the heart is suddenly blocked, usually by a blood clot. The interrupted blood flow may damage or destroy part of the heart muscle and can be life threatening.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common myocardial Infarction signs and symptoms include:

- Pressure, tightness, pain, or a squeezing or aching sensation in your chest or arms that may spread to your neck, jaw or back
- Nausea, indigestion, heartburn or abdominal pain
- Shortness of breath
- Cold sweat
- Fatigue or lightheadedness

RISK FACTORS

Certain factors contribute to the unwanted buildup of fatty deposits (atherosclerosis) that narrows arteries throughout your body. Heart attack risk factors include:

- Age: men age 45 or older and women age 55 or older are more likely to have a heart attack than are younger men and women
- Lifestyle: tobacco, lack of physical activity, stress, illicit drug use
- Medical: high blood pressure, cholesterol and triglyceride levels, obesity, diabetes, autoimmune condition
- Family history of heart attack
- A history of preeclampsia

COMPLICATIONS

Complications are often related to the damage done to your heart during an attack, which can lead to abnormal heart rhythms (arrhythmias), heart failure, sudden cardiac arrest.

Gene	Your Genotype	Your Result	Explanation
CDKN2A/B	CC	Higher Risk	The CC genotype may promote cell proliferation, cause atherosclerosis and increase the likelihood of myocardial infarction.
PSRC1	AA	Higher Risk	The G>A mutation in this gene locus may reduce mRNA transcription of PSRC1, cause elevated serum total cholesterol levels and increase the likelihood of myocardial infarction.
LPL	GG	Higher Risk	The gene locus that carries G allele may affect the transcription of LPL, which in turn affects enzyme function. This causes elevated serum triglycerides and increase in risk of myocardial infarction.
BRAP	TT	Normal Risk	C allele affects the expression of BRAP gene to a certain extent, stimulates the production of inflammatory factors and causes myocardial infarction.

Health Risk

Ischemic Stroke

Ischemic stroke, commonly known as "stroke," is a type of disease due to a sudden rupture or blockage of blood vessels within the brain. It is not possible to completely prevent strokes because some factors that increase the risk of the condition cannot be changed, such as age, gender and ethnicity, But it is possible to significantly reduce the risk of having a stroke by making lifestyle changes to avoid problems such as atherosclerosis and high blood pressure.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Ischemic stroke signs and symptoms include:
· Trouble with speaking and understanding
· Paralysis or numbness of the face, arm or leg
· Trouble seeing in one or both eyes
· Headache
· Trouble with walking

RISK FACTOR

Many factors can increase your stroke risk. Potentially treatable stroke risk factors include:

Lifestyle risk factors
· Being overweight or obese
· Physical inactivity
· Heavy or binge drinking
· Use of illicit drugs such as cocaine and methamphetamines

Medical risk factors
· Blood pressure readings higher than 120/80 millimeters of mercury (mm Hg)
· Cigarette smoking or exposure to secondhand smoke
· High cholesterol
· Diabetes
· Obstructive sleep apnea
· Cardiovascular disease, including heart failure, heart defects, heart infection or abnormal heart rhythm
· Personal or family history of stroke, heart attack or transient ischemic attack

COMPLICATIONS

A stroke can sometimes cause temporary or permanent disabilities, depending on how long the brain lacks blood flow and which part was affected. Complications may include paralysis or loss of muscle movement, difficulty talking or swallowing, memory loss or thinking difficulties, emotional problems, pain, changes in behavior and self-care ability.

Gene	Your Genotype	Your Result	Explanation
ADIPOQ	TG	Normal Risk	The GG genotype in this gene locus may cause a decrease in adiponectin levels, leading to the formation of atherosclerosis and promoting ischemic stroke.
PRKCH	GG	Normal Risk	The G>A mutation in this gene locus may affect the process of atherosclerosis and promote the development of ischemic stroke.

Health Risk

Atrial Fibrillation

Atrial fibrillation (irregular heartbeat) can be hereditary; while those with high blood pressure, high cholesterol and high blood sugar as well as sleep apnea (sleep snoring) have a higher incidence rate. The symptoms of atrial fibrillation vary from person to person. The most common symptom is the sudden increase in heart rate leading to discomfort, some people may feel chest tightness, shortness of breath or sweating, some people may be accompanied by polyuria (frequent urination) and fatigue.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Some people with atrial fibrillation have no symptoms and are unaware of their condition until it is discovered during a physical examination. Those who do have atrial fibrillation symptoms may experience signs and symptoms such as:

- Palpitations, which are sensations of a racing, uncomfortable, irregular heartbeat or a flip-flopping in your chest
- Weakness
- Reduced ability to exercise
- Fatigue
- Lightheadedness
- Dizziness
- Shortness of breath
- Chest pain

RISK FACTOR

Certain factors may increase your risk of developing atrial fibrillation. These include age, heart disease, high blood pressure, other chronic conditions, drinking alcohol, obesity and a family history of atrial fibrillation.

COMPLICATIONS

Sometimes atrial fibrillation can lead to complications such as stroke and heart failure.

Gene	Your Genotype	Your Result	Explanation
PITX2c	TG	Higher Risk	The gene locus carrying T allele affects the function of PITX2c gene, thereby increasing the risk of atrial fibrillation.
PITX2c	TC	Normal Risk	The TT genotype in this gene locus affects the function of PITX2c gene, thereby increasing the risk of atrial fibrillation.
CYP11B2	AA	Normal Risk	This gene locus that carries G allele leads to increased risk of myocardial fibrosis and myocardial remodeling and providing the basis for the occurrence of atrial fibrillation.
ZFHX3	CC	Normal Risk	Gene locus carrying T allele leads to elevated levels of inflammatory responses and heterogeneity of atrial myocyte, thereby increasing the risk of atrial fibrillation.
NPPA-AS1	CC	Normal Risk	T allele increases the risk of atrial fibrillation.

Health Risk

Dilated Cardiomyopathy

Dilated cardiomyopathy is a general term for diseases of the heart muscle, where the walls of the heart chambers have become stretched and thin. This affects the heart's ability to pump blood around the body. Dilated cardiomyopathy might not cause symptoms, but for some people it can be life-threatening.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

If you have dilated cardiomyopathy, you are likely to have signs and symptoms of heart failure or arrhythmias caused by your condition. Signs and symptoms include:

- Fatigue
- Shortness of breath (dyspnea) when you are active or lying down
- Reduced ability to exercise
- Swelling (edema) in your legs, ankles and feet
- Swelling of your abdomen due to fluid buildup (ascites)
- Chest pain
- Extra or unusual sounds heard when your heart beats (heart murmurs)

RISK FACTOR

Dilated cardiomyopathy most commonly occurs in men, ages 20 to 50. But it can also occur in women. Other risk factors include:

- Damage to the heart muscle from a heart attack
- Family history of dilated cardiomyopathy
- Inflammation of heart muscle from immune system disorders, such as lupus
- Neuromuscular disorders, such as muscular dystrophy
- Obesity, diabetes, high blood pressure
- Abuse of alcohol or cocaine
- Certain cancer medications
- Infections
- Exposure to toxins
- Arrhythmias
- Complications of late-stage pregnancy

COMPLICATIONS

Complications from dilated cardiomyopathy include:

- Heart failure
- Heart valve regurgitation
- Fluid buildup (edema)
- Abnormal heart rhythms (arrhythmias)
- Sudden cardiac arrest
- Blood clots (emboli)

Gene	Your Genotype	Your Result	Explanation
BAG3	TT	Normal Risk	Data demonstrated that C>T mutations at this site can reduce the risk of dilated cardiomyopathy in individuals.

Health Risk

Hypertriglyceridemia

Hypertriglyceridemia refers to an elevated triglyceride level in the blood, which is affected by both genetic and environmental factors. Hypertriglyceridemia is one of the most common chronic diseases that interacts with other risk factors to cause cardiovascular disease, leading to atherosclerosis and increased morbidity and mortality of cardiovascular and cerebrovascular diseases.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Most people with elevated triglycerides experience no symptoms. Some signs and symptoms of hypertriglyceridemia include:
· Skin symptoms (eruptive xanthoma)
· Eye abnormalities (corneal arcus, lipemia retinalis, xanthelasmas)
· Hepatosplenomegaly (enlargement of the liver and spleen)
· Neurological symptoms (memory loss,dementia, depression)
· Mild episodes of pancreatitis

RISK FACTORS

The risk of familial hypertriglyceridemia is higher if one or both of your parents have the gene defect that causes it.

Hypertriglyceridemia also tend to be more common in people who have:
· Obesity
· Diabetes mellitus and insulin resistance
· Kidney failure and nephrotic syndrome
· Lipoprotein deficiency
· Hypothyroidism
· Systemic Lupus Erythematosus
· Glycogen storage disease Type 1

Certain medications can also increase the risk of getting hypertriglyceridemia:
· Thiazide diuretics
· Beta-blockers
· Protease inhibitors for HIV
· Some cholesterol-lowering drugs
· Estrogen therapy
· Isotretinoin for acne
· Immunosuppressants
· Corticosteroids
· Some antipsychotics

Gene	Your Genotype	Your Result	Explanation
Chr 11 Intergenic	GC	Higher Risk	The C>G mutation reduces HDL (high-density lipoprotein) concentration, increases TG (triglyceride) concentration and increases the risk of hypertriglyceridemia.
GCKR	TC	Normal Risk	The TT genotype at this gene locus activates glucokinase activity and promotes glucose uptake. Sugars and lipids can be mutually transformed and thereby promoting the synthesis of triglycerides that increase the risk of hypertriglyceridemia.

Health Risk

Coronary Spasm

Coronary spasm is a sudden tightening of the muscles within the arteries of the heart. When this occurs, arteries are narrow and prevent blood from flowing to the heart. Coronary artery spasms are brief and temporary. However, it can potentially lead to further heart complications, such as a heart attack.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Coronary spasms signs and symptoms:
· Chest pain or angina
· Pain on the left side of the chest
· Chest tightness
· Feeling of constriction

RISK FACTORS

Known risk factors for coronaryspasm include:
· Smoking
· Excess use of stimulants, such as cocaine and amphetamines
· Extreme stress
· Extreme cold
· Alcohol withdrawal

COMPLICATIONS

Complications of coronary spasm include:
· Heart arrhythmias, when heart beats irregularly or too fast or too slow
· Heart attacks, when there is a complete blockage of blood flow to heart
· Cardiac arrest
· Death

Gene	Your Genotype	Your Result	Explanation
eNOS	TT	Normal Risk	eNOS gene produces a protein that synthesizes carbon monoxide. Reports indicate that individuals with TC and CC genotypes tend to have a higher genetic risk of coronary spasm.

Health Risk

Intracranial Aneurysm

Intracranial aneurysm, also known as brain aneurysm, is an abnormal dilation or ballooning of a blood vessel in the brain that results from a weakening of the blood vessel wall. A brain aneurysm can leak or rupture, causing bleeding into the brain (hemorrhagic stroke).

6 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common signs and symptoms of aneurysm include:
- Sudden, extremely severe headache
- Nausea and vomiting
- Stiff neck
- Blurred or double vision
- Sensitivity to light
- Seizure
- A drooping eyelid
- Loss of consciousness
- Confusion

RISK FACTORS

Risk factors of aneurysm nclude:
- Older age
- Cigarette smoking
- High blood pressure
- Drug abuse, particularly the use of cocaine
- Heavy alcohol consumption

COMPLICATIONS

Complications of aneurysm include:
- Re-bleeding
- Vasospasm
- Hydrocephalus
- Hyponatremia

Gene	Your Genotype	Your Result	Explanation
PLCL1	AG	Higher Risk	Reports indicate that individuals with AG and GG genotypes tend to have a higher genetic risk of intracranial aneurysm.
EDNRA	CC	Higher Risk	Reports indicate that individuals with AC and CC genotypes tend to have a higher genetic risk of intracranial aneurysm.
CDKN2B-AS1	TT	Higher Risk	Reports indicate that individuals with TC and TT genotypes tend to have a higher genetic risk of intracranial aneurysm.
CNNM2	AG	Higher Risk	Reports indicate that individuals with AG and GG genotypes tend to have a higher genetic risk of intracranial aneurysm.
STARD13	TC	Higher Risk	Reports indicate that individuals with TC and TT genotypes tend to have a higher genetic risk of intracranial aneurysm.
RBBP8	AC	Higher Risk	Reports indicate that individuals with AC and CC genotypes tend to have a higher genetic risk of intracranial aneurysm.

Health Risk

Peripheral Artery Disease

Peripheral artery disease (PAD) is a narrowing of the peripheral arteries that carry blood away from the heart to other parts of the body. The most common type of PAD affects the legs, in which blood flow is reduced to the legs and feet. PAD is usually caused by atherosclerosis (build-up of fatty plagues in the arteries).

1

variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Peripheral artery disease signs and symptoms include:
· Painful cramping in one or both hips, thighs or calf muscles after certain activities, such as walking or climbing stairs
· Leg numbness or weakness
· Coldness in lower leg or foot, especially when compared with the other side
· Sores on toes, feet or legs
· Change in the color of legs
· Slower growth of toenails
· Shiny skin on legs
· No pulse or a weak pulse in legs or feet
· Erectile dysfunction in men

RISK FACTORS

Factors that increase the risk of developing PAD include:
· Smoking
· Diabetes
· Obesity
· High blood pressure
· High cholesterol
· Increasing age, especially after reaching 50 years of age
· Genetics
· High levels of homocysteine, a protein component that helps build and maintain tissue

COMPLICATIONS

If your PAD is caused by a buildup of plaques in blood vessels, you are also at risk of developing:
· Critical limb ischemia
· Stroke
· Heart attack

Gene	Your Genotype	Your Result	Explanation
EDNRA	CC	Normal Risk	Reports indicate that individuals with AA genotype tend to have a higher genetic risk of peripheral artery disease.
HDAC9	TC	Higher Risk	Reports indicate that individuals with CT and CC genotypes tend to have a higher genetic risk of peripheral artery disease.

Health Risk

Venous Thromboembolism

Venous thromboembolism is a condition in which a blood clot forms in the deep veins of the leg, groin or arm (known as deep vein thrombosis) and travels in the circulation, lodging in the lungs (known as pulmonary embolism). It is a dangerous, potentially deadly medical condition.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of deep vein thrombosis (DVT):
- Pain or tenderness, often starting in the calf
- Swelling, including the ankle or foot
- Redness or noticeable discoloration
- Warmth

Signs and symptoms of pulmonary embolism (PE):
- Unexplained shortness of breath
- Rapid breathing
- Chest pain (may be worse upon deep breath)
- Rapid heart rate
- Light headedness or passing out

RISK FACTORS

Factors that increase the risk of developing venous thromboembolism include:
- Inheriting a blood-clotting disorder
- Prolonged bed rest, such as during a long hospital stay
- Injury or surgery
- Pregnancy
- Oral contraceptives or hormone replacement therapy
- Overweight or obese
- Smoking
- Cancer
- Genetics

COMPLICATIONS

One-third of people who have a DVT will have long-term complications called post-thrombotic syndrome (PTS). People with PTS have symptoms such as swelling, pain, discoloration, and in severe cases, scaling or ulcers in the affected part of the body. In some cases, the symptoms can be so severe that a person becomes disabled.

For some, DVT and PE can become a chronic illness; about 30% of people who have had a DVT or PE are at risk for another episode.

Gene	Your Genotype	Your Result	Explanation
F5	CC	Normal Risk	F5 gene produces a type of clotting factors. Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of venous thromboembolism.
FGG	GG	Normal Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of venous thromboembolism.
SLC44A2	AG	Higher Risk	Reports indicate that individuals with AG and GG genotypes tend to have a higher genetic risk of venous thromboembolism.

Health Risk

Abdominal Aortic Aneurysm

An abdominal aortic aneurysm is an enlarged area in the lower part of the major vessel that supplies blood to the body (aorta). The aorta runs from the heart through the center of the chest and abdomen.

3 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of an enlarging abdominal aortic aneurysm:
· Deep, constant pain in abdomen or on the side of abdomen
· Back pain
· A pulse near belly button

RISK FACTORS

Factors that increase the risk of developing abdominal aortic aneurysm include:
· Smoking
· Age: age 65 and older
· Gender: male have a higher risk
· High blood pressure
· High blood cholesterol
· Genetics

COMPLICATIONS

Tears in one or more of the layers of the wall of the aorta (aortic dissection) or a ruptured aneurysm are the main complications. Signs and symptoms that aortic aneurysm has ruptured include:
· Sudden, intense and persistent abdominal or back pain
· Dizziness
· Fast heartbeat

Gene	Your Genotype	Your Result	Explanation
DAB2IP	AG	Higher Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of abdominal aortic aneurysm.
LRP1	GG	Higher Risk	Reports indicate that individuals with GG and AG genotypes tend to have a higher genetic risk of abdominal aortic aneurysm.
LDLR	GG	Higher Risk	Reports indicate that individuals with GG genotype tend to have a higher genetic risk of abdominal aortic aneurysm.

Health Risk

Kawasaki Disease

Kawasaki disease causes swelling (inflammation) in the walls of medium-sized arteries throughout the body. It primarily affects children. The inflammation tends to affect the coronary arteries, which supply blood to the heart muscle. Kawasaki disease is sometimes called mucocutaneous lymph node syndrome because it also affects the lymph nodes, skin, and the mucous membranes inside the mouth, nose and throat.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Kawasaki disease signs and symptoms usually appear in three phases.

First phase:
· High fever that lasts more than three days
· Extremely red eyes
· Rashes on the body and in the genital area
· Red, dry, cracked lips and an extremely red, swollen tongue
· Swollen, red skin on the palms of the hands and the soles of the feet
· Swollen lymph nodes in the neck
· Irritability

Second phase:
· Peeling of the skin on the hands and feet, especially at the tips of the fingers and toes
· Joint pain
· Diarrhea
· Vomiting
· Abdominal pain

Third phase:
Signs and symptoms slowly go away unless complications develop. It may take as long as 8 weeks to reach this stage.

RISK FACTORS

Three things are known to increase the risk of developing Kawasaki disease:
· Age: children under 5 years old have higher risk
· Gender: male are slightly more likely to develop this disease
· Ethnicity: children of Asian or Pacific Island descent have higher rates

COMPLICATIONS

Kawasaki disease is a leading cause of acquired heart disease in children. However, with effective treatment, only a few have lasting damage. Heart complications include:
· Inflammation of blood vessels, usually the coronary arteries
· Inflammation of the heart muscle
· Heart valve problems

Gene	Your Genotype	Your Result	Explanation
MLF1IP	GG	Normal Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of Kawasaki disease.
HLA-DOB	GG	Higher Risk	Reports indicate that individuals with GG and AG genotypes tend to have a higher genetic risk of Kawasaki disease.
BLK	AG	Higher Risk	Reports indicate that individuals with GG and AG genotypes tend to have a higher genetic risk of Kawasaki disease.
MIA	GG	Normal Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of Kawasaki disease.
CD40	TT	Normal Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of Kawasaki disease.

Health Risk

Orthostatic Hypotension

Orthostatic hypotension (also called postural hypotension) is a form of low blood pressure that happens when standing up from a sitting or lying position. It may cause dizziness, light-headedness, and maybe even fainting. Each episode can last for a few minutes.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Orthostatic hypotension signs and symptoms include:
· Lightheadedness or dizziness upon standing
· Blurry vision
· Weakness
· Fainting
· Confusion
· Nausea

RISK FACTORS

Risk factors for orthostatic hypotension include:
· Age: age 65 and older
· Medications: medications used to treat high blood pressure or heart disease, such as diuretics and alpha blockers
· Bed rest
· Pregnancy
· Alcohol consumption

COMPLICATIONS

Persistent orthostatic hypotension can cause serious complications, especially in older adults. These include:
· Falls
· Stroke
· Cardiovascular diseases

Gene	Your Genotype	Your Result	Explanation
CTNNA2	AC	Normal Risk	CTNNA2 gene is suggested to function to regulate stability of synapses serving as hinges between neurons. Reports indicate that individuals with AC and CC genotypes tend to have a lower genetic risk of orthostatic hypotension.

Health Risk

Carotid Artery Disease

Carotid artery disease occurs when fatty deposits (plaques) clog the blood vessels that deliver blood to the brain and head (carotid arteries). The blockage increases the risk of stroke when blood supply to the brain is interrupted or seriously reduced.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Carotid artery disease does not produce any signs and symptoms in its early stages. The condition may go unnoticed until it is serious enough to deprive brain of blood, causing a stroke. Signs and symptoms of stroke include:

· Sudden numbness or weakness in the face or limbs, often on only one side of the body
· Sudden trouble speaking
· Sudden trouble seeing in one or both eyes
· Sudden dizziness or loss of balance
· Sudden, severe headache with no known cause

RISK FACTORS

Factors that increase the risk of carotid artery disease include:

· High blood pressure
· Smoking
· Diabetes
· High blood-fat levels
· Genetics
· Obesity
· Lack of exercise

COMPLICATIONS

Carotid artery disease can lead to stroke through:

· Reduced blood flow
· Ruptured plaques
· Blood clot blockage

Gene	Your Genotype	Your Result	Explanation
EDNRA	CC	Normal Risk	Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of carotid artery disease.
PIK3CG	GG	Normal Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of carotid artery disease.

Health Risk

Brugada Syndrome

Brugada syndrome is a rare, but potentially life-threatening heart rhythm disorder that is sometimes inherited. People with Brugada syndrome have an increased risk of having irregular heart rhythms beginning in the lower chambers of the heart (ventricles).

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms that may be associated with Brugada syndrome include:
· Dizziness
· Fainting
· Gasping, labored breathing, particularly at night
· Irregular heartbeats or palpitations
· Extremely fast and chaotic heartbeat
· Seizures

RISK FACTORS

Risk factors for Brugada syndrome include:
· Genetics
· Gender: male has higher risk of getting this disease
· Ethnicity: it occurs more frequently in Asians

COMPLICATIONS

Complications of Brugada syndrome require emergency medical care. These include:
· Sudden cardiac arrest
· Fainting

Gene	Your Genotype	Your Result	Explanation
SCN10A	TG	Higher Risk	Reports indicate that individuals with TT and TG genotypes tend to have a higher genetic risk of Brugada syndrome.
HEY2	TC	Higher Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of Brugada syndrome.

HEALTH RISK

URINARY SYSTEM

Health Risk

Chronic Kidney Disease

When the kidney is damaged, kidney function is reduced that causes the accumulation of waste, electrolytes and fluid in the body. In the early stages of chronic kidney disease, you may have few signs or symptoms. Chronic kidney disease may not become apparent until your kidney function is significantly impaired. Treatment for chronic kidney disease focuses on slowing the progression of kidney damage, usually by controlling the underlying cause. Chronic kidney disease can progress to end-stage kidney failure, which is fatal without dialysis or a kidney transplant.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of chronic kidney disease develop over time as kidney damage progresses slowly. Signs and symptoms of kidney disease may include nausea, vomiting, loss of appetite, fatigue and weakness, sleep problems, reduced urine amount, decreased mental sharpness, muscle twitches and cramps, swelling of feet and ankles, persistent itching, chest pain (if fluid builds up around the lining of the heart), shortness of breath (if fluid builds up in the lungs) and high blood pressure (hypertension).

RISK FACTORS

Certain factors may increase your risk of developing chronic kidney disease which include:
- Diabetes
- High blood pressure
- Heart and blood vessel (cardiovascular) disease
- Smoking
- Obesity
- Family history of kidney disease
- Abnormal kidney structure
- Older age

COMPLICATIONS

Chronic kidney disease may lead to the following complications:
- Fluid retention, which could lead to swelling in your arms and legs, high blood pressure, or fluid in your lungs (pulmonary edema)
- A sudden rise in potassium levels in your blood (hyperkalemia), which could impair your heart's ability to function and may be life-threatening
- Heart and blood vessel (cardiovascular) disease
- Weak bones and an increased risk of bone fractures
- Anemia
- Decreased sex drive, erectile dysfunction or reduced fertility
- Damage to your central nervous system, which can cause difficulty concentrating, personality changes or seizures
- Decreased immune response, which makes you more vulnerable to infection
- Pericarditis, an inflammation of the saclike membrane that envelops your heart (pericardium)
- Pregnancy complications that carry risks for the mother and the developing fetus
- Irreversible damage to your kidneys (end-stage kidney disease), eventually requiring either dialysis or a kidney transplant for survival

Gene	Your Genotype	Your Result	Explanation
UMOD	AA	Higher Risk	AA genotype at this gene locus may cause abnormal expression of UMOD gene that increases uromodulin excretion and the risk of chronic kidney disease.

Health Risk

Renal Calculus

A renal calculus (kidney stone) is a hard object that is made from chemicals in the urine. Urine has various wastes dissolved in it. When there is too much waste in too little liquid, crystals begin to form. The crystals attract other elements and join together to form a solid that will get larger unless it is passed out of the body with the urine. After it is formed, the stone may stay in the kidney or travel down the urinary tract into the ureter. Sometimes, tiny stones move out of the body in the urine without causing too much pain. But stones that do not move may cause a back-up of urine in the kidney, ureter, the bladder, or the urethra. This is what causes the pain.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Renal calculus may not cause symptoms until it moves around within your kidney or passes into your ureter - the tube connecting the kidney and bladder. At that point, you may experience these signs and symptoms:

- Severe pain in the side and back, below the ribs
- Pain that radiates to the lower abdomen and groin
- Pain that comes in waves and fluctuates in intensity
- Painful urination
- Pink, red or brown urine
- Cloudy or foul-smelling urine
- Nausea and vomiting
- Persistent need to urinate
- Urinating more often than usual
- Fever and chills if an infection is present
- Urinating small amounts

RISK FACTORS

Certain factors may increase your risk of developing renal calculus which include:

- Family or personal history
- Dehydration
- Certain diets: high in protein, sodium and sugar
- Being obese
- Digestive diseases and surgery
- Other medical conditions - renal tubular acidosis, cystinuria, hyperparathyroidism, certain medications and some urinary tract infections

Gene	Your Genotype	Your Result	Explanation
SLC34A1	AG	Higher Risk	The specific function of this site is still unclear,but study found that when the G>A mutation occurs at this site, the risk of disease increases.
DGKH	TC	Moderate Risk	When the T>C mutation occurs at this site, the risk of disease increases.
AQP1	TT	Normal Risk	When the T>C mutation occurs at this site, the risk of disease increases.
UMOD	AA	Higher Risk	When the G>A mutation occurs at this site, the risk of disease increases.

Health Risk

IgA Nephropathy

IgA Nephropathy is a condition of inflammation of the glomerulus, a filtering part of the kidney. We do not fully understand what caused IgA nephropathy, but it appears to relate to a type of antibody called IgA. IgA is a normal part of our natural defence against infection. In IgA nephropathy we believe this antibody behaves abnormally and collects in the glomerulus, causing it to become inflamed and scarred over time. No cure exists for IgA nephropathy, but certain medications can slow its course. Keeping your blood pressure under control and reducing your cholesterol levels also slow the disease.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

IgA nephropathy usually doesn't cause symptoms in the early stages, so the disease can go unnoticed for years or decades. It is sometimes suspected when routine tests reveal protein and red blood cells in your urine that cannot be seen without a microscope (microscopic hematuria). Signs and symptoms of IgA nephropathy include:

- Cola- or tea-colored urine (caused by red blood cells in the urine)
- Repeated episodes of cola- or tea-colored urine, and sometimes visible blood in your urine, usually during or after an upper respiratory or other infection and sometimes after strenuous exercise
- Foamy urine from protein leaking into your urine (proteinuria)
- Pain in the one or both sides of your back below your ribs
- Swelling (edema) in your hands and feet
- High blood pressure

RISK FACTOR

Although the exact cause of IgA nephropathy is unknown, these factors might increase your risk of developing this condition:

- Gender: IgA nephropathy affects at least twice as many men as it does women
- Age: it is most often develops between the late teens and late 30s
- Family history

COMPLICATIONS

The course of IgA nephropathy varies from person to person. Some people have the disease for years with few or no problems. In fact, many cases go undiagnosed. Other people develop one or more of the following complications:

- High blood pressure
- High cholesterol
- Acute kidney failure
- Chronic kidney disease
- Nephrotic syndrome

Gene	Your Genotype	Your Result	Explanation
PSMB8	TG	Higher Risk	G allele can increase the mRNA expression level of PSMB8 gene, directly increase the number of proteins that are involved in antigen processing and presentation and eventually increase the risk of disease.
CFH	GG	Higher Risk	The GG genotype in this gene locus may affect the activation of complement resulting in an increased risk of IgA nephropathy.
HLA-DPB2	GG	Higher Risk	The G allele at this gene locus may cause serum IgA deposition in patients with concomitant hematuria (blood in urine) and severe hypertension.

Health Risk

Acquired Nephrotic Syndrome

Nephrotic syndrome is a kidney disorder that causes your body to pass too much protein in your urine. Nephrotic syndrome is usually caused by damage to the clusters of small blood vessels in your kidneys that filter waste and excess water from your blood. The condition causes swelling, particularly in your feet and ankles, and increases the risk of other health problems. Treatment for nephrotic syndrome includes treating the condition that is causing it and taking medications. Nephrotic syndrome can increase your risk of infections and blood clots. Your doctor might recommend medications and dietary changes to prevent complications.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of nephrotic syndrome include:
· Severe swelling (edema), particularly around your eyes and in your ankles and feet
· Foamy urine, which may be caused by excess protein in your urine
· Weight gain due to excess fluid retention
· Fatigue
· Loss of appetite

RISK FACTOR

Factors that can increase your risk of nephrotic syndrome include:
· Medical conditions that can damage your kidneys
· Certain medications: nonsteroidal anti-inflammatory drugs and drugs used to fight infections
· Certain infections: HIV, hepatitis B, hepatitis C and malaria

COMPLICATIONS

Possible complications of nephrotic syndrome include:
· Blood clots
· High blood cholesterol and elevated blood triglycerides
· Poor nutrition
· High blood pressure
· Acute kidney failure
· Chronic kidney disease
· Infections

Gene	Your Genotype	Your Result	Explanation
GPC5	GG	Normal Risk	The G>A mutation at this site leads to an increase in the expression of GPC5 gene in podocytes, which is associated with an increased risk of secondary nephrotic syndrome.

HEALTH RISK
INFECTION

Health Risk

Tuberculosis

Tuberculosis (TB) is a potentially serious infectious disease that mainly affects the lungs. The bacteria that cause TB are spread through tiny droplets released into the air via coughs and sneezes. People infected with TB bacteria have a 5–10% lifetime risk of falling ill with TB.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

TB infections can be either latent or active. Signs and symptoms of active TB include:
· Coughing that lasts three or more weeks
· Coughing up blood
· Chest pain, or pain with breathing or coughing
· Unintentional weight loss
· Fatigue
· Fever
· Night sweats
· Chills
· Loss of appetite

RISK FACTORS

Anyone can get TB, but certain factors can increase risk of the disease. These factors include:
· Weakened immune system
· Substance use
· Traveling or living in a country where TB is common

COMPLICATIONS

Examples of tuberculosis complications include:
· Spinal pain
· Joint damage
· Meningitis
· Liver or kidney problems
· Heart disorders

Gene	Your Genotype	Your Result	Explanation
ASAP1	CC	Normal Risk	Reports indicate that individuals with AC and CC genotypes tend to have a lower genetic risk of tuberculosis.
WT1	AG	Normal Risk	Reports indicate that individuals with AG and AA genotypes tend to have a lower genetic risk of tuberculosis.

Health Risk

Chronic Hepatitis B

Hepatitis B is a serious liver infection caused by the hepatitis B virus (HBV). For some people, hepatitis B infection becomes chronic, meaning it lasts more than six months. Having chronic hepatitis B increases the risk of developing liver failure, liver cancer or cirrhosis — a condition that permanently scars the liver.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Hepatitis B signs and symptoms may include:
· Abdominal pain
· Dark urine
· Fever
· Joint pain
· Loss of appetite
· Nausea and vomiting
· Weakness and fatigue
· Jaundice

RISK FACTORS

HBV spreads through contact with blood, semen or other body fluids from an infected person.

Common ways HBV spreads:
· Having unprotected sex with an infected person
· Sharing of needles
· An infant born to an infected mother
· Travel to regions with high infection rates of HBV, such as Asia, the Pacific Islands, Africa and Eastern Europe

COMPLICATIONS

Having a chronic HBV infection can lead to serious complications, such as:
· Scarring of the liver (cirrhosis)
· Liver cancer
· Liver failure

Gene	Your Genotype	Your Result	Explanation
HLA-DQB1	TC	Higher Risk	Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of chronic hepatitis B.
HLA-DQB2	AG	Higher Risk	Reports indicate that individuals with GG and AG genotypes tend to have a higher genetic risk of chronic hepatitis B.

Health Risk

Chronic Hepatitis C

Hepatitis C is a viral infection that causes liver inflammation, sometimes leading to serious liver damage. The hepatitis C virus (HCV) spreads through contaminated blood.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Chronic hepatitis C is usually a "silent" infection for many years, until the virus damages the liver enough to cause the signs and symptoms of liver disease. Signs and symptoms include:

- Bleeding and bruising easily
- Fatigue
- Poor appetite
- Jaundice
- Dark urine
- Itchy skin
- Fluid buildup in abdomen (ascites)
- Swelling in legs
- Weight loss
- Confusion, drowsiness and slurred speech
- Spider angiomas

RISK FACTORS

HCV spreads through contact with blood from an infected person.

Common ways HCV spreads:

- Having unprotected sex with an infected person
- Sharing of needles
- Sharing personal items
- An infant born to an infected mother

COMPLICATIONS

Hepatitis C infection that continues over many years can cause significant complications, such as:

- Scarring of the liver (cirrhosis)
- Liver cancer
- Liver failure

Gene	Your Genotype	Your Result	Explanation
DEPDC5	TT	Normal Risk	Reports indicate that individuals with TG and GG genotypes tend to have a higher genetic risk of chronic hepatitis C.

Health Risk

Leprosy

Leprosy is a long-term infection by the bacteria Mycobacterium leprae. Infection can lead to damage of the nerves, respiratory tract, skin, and eyes. This nerve damage may result in a lack of ability to feel pain, which can lead to the loss of parts of a person's extremities from repeated injuries or infection due to unnoticed wounds.

4 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The disease can cause skin symptoms such as:
· Discolored patches of skin
· Growths (nodules) on the skin
· Thick, stiff or dry skin
· Painless ulcers on the soles of feet
· Painless swelling or lumps on the face or earlobes
· Loss of eyebrows or eyelashes

Symptoms caused by damage to the nerves are:
· Numbness of affected areas of the skin
· Muscle weakness or paralysis
· Enlarged nerves
· Eye problems that may lead to blindness

RISK FACTORS

Risk factors for leprosy include:
· Prolonged, close contact with someone with untreated leprosy over many months
· Immunosuppression
· Genetics

COMPLICATIONS

Examples of leprosy complications include:
· Blindness or glaucoma
· Inflammation of the iris
· Hair loss
· Infertility
· Disfigurement
· Erectile dysfunction
· Kidney failure
· Muscle weakness
· Permanent damage to the nose causing nosebleeds and stuffy nose
· Permanent nerve damage in the arms and legs

Health Risk

Leprosy

Gene	Your Genotype	Your Result	Explanation
IL23R	AA	Higher Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of leprosy.
HLA-DR-DQ	TC	Higher Risk	Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of leprosy.
RIPK2	TC	Higher Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of leprosy.
TNFSF15	TC	Higher Risk	Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of leprosy.
NOD2	TT	Normal Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of leprosy.

Health Risk

Dengue Shock Syndrome

Dengue fever is a mosquito-borne illness that occurs in tropical and subtropical areas. Mild dengue fever causes high fever and flu-like symptoms. The severe form of dengue fever, also called dengue hemorrhagic fever, can cause serious bleeding, a sudden drop in blood pressure (shock) and death.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Warning signs of dengue shock syndrome — which is a life-threatening emergency and can develop quickly. These include:
· Severe stomach pain
· Persistent vomiting
· Bleeding from gums or nose
· Blood in urine, stools or vomit
· Bleeding under the skin, which might look like bruising
· Difficult or rapid breathing
· Fatigue
· Irritability or restlessness

RISK FACTORS

Risk factors of developing dengue fever or dengue shock syndrome include:
· Live or travel in tropical areas
· Had dengue fever in the past

COMPLICATIONS

Dengue shock syndrome complications include:
· Internal bleeding
· Organ damage
· Blood pressure drop to dangerous level
· Death

Gene	Your Genotype	Your Result	Explanation
PLCE1	TT	Normal Risk	Reports indicate that individuals with TT and TC genotypes tend to have a lower genetic risk of dengue shock syndrome.

Health Risk

Severe Malaria

Malaria is a disease caused by parasites. The parasite is transmitted to humans through the bites of infected mosquitoes. People suffering from malaria usually feel very sick, accompanied by high fever and shaking chills.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

A malaria infection is generally characterized by the following signs and symptoms:
· Fever
· Chills
· General feeling of discomfort
· Nausea and vomiting
· Muscle pain and fatigue
· Sweating
· Chest or abdominal pain

RISK FACTORS

Risk factors for developing malaria:
· Living in tropical and subtropical regions
· Through mosquito transmission
· Exposure to infected blood

COMPLICATIONS

In most cases, malaria deaths are related to one or more serious complications, including:
· Cerebral malaria
· Breathing problems
· Organ failure
· Anemia
· Low blood sugar

Gene	Your Genotype	Your Result	Explanation
ABO	DI	Higher Risk	Reports indicate that individuals with the DD genotype tend to have a lower genetic risk of severe malaria.
HBB	GG	Higher Risk	Reports indicate that individuals with GC and CC genotypes of this site tend to have a lower genetic risk of severe malaria.

HEALTH RISK
GENDER

Health Risk

Endometriosis

[Only applicable for female]

Endometriosis is an often painful disorder in which tissue similar to the tissue that normally lines the inside of the uterus — the endometrium — grows outside of the uterus. Endometriosis most commonly involves ovaries, fallopian tubes and the tissue lining the pelvis. Rarely, endometrial tissue may spread beyond pelvic organs.

4 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common signs and symptoms of endometriosis include:
· Painful periods
· Pain during intercourse
· Pain during bowel movements or urination
· Excessive bleeding
· Infertility

RISK FACTORS

Several factors increase the risk of developing endometriosis, such as:
· Genetics
· Reproductive tract abnormalities

COMPLICATIONS

Examples of endometriosis complications include:
· Infertility
· Debilitating pelvic pain
· Cysts

Gene	Your Genotype	Your Result	Explanation
WNT4	AA	Higher Risk	Reports indicate that individuals with AA and AC genotypes tend to have a higher genetic risk of endometriosis.
RHOU	TT	Normal Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of endometriosis.
GREB1	GG	Higher Risk	Reports indicate that individuals with GG and AG genotypes tend to have a higher genetic risk of endometriosis.
IL1A	CC	Higher Risk	Reports indicate that individuals with CC and TC genotypes tend to have a higher genetic risk of endometriosis.
LOC100506885	TC	Higher Risk	Reports indicate that individuals with TT and TC genotypes tend to have a higher genetic risk of endometriosis.
Intergenic	TT	Normal Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of endometriosis.
CDKN2B	AA	Normal Risk	Reports indicate that individuals with CC and CA genotypes tend to have a higher genetic risk of endometriosis.
VEZT	AA	Normal Risk	Reports indicate that individuals with CC and CA genotypes tend to have a higher genetic risk of endometriosis.

Health Risk

Uterine Fibroids

[Only applicable for female]

Uterine fibroids are noncancerous growths of the uterus that often appear during childbearing years. Uterine fibroids are not associated with an increased risk of uterine cancer and almost never develop into cancer.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of uterine fibroids include:
· Heavy menstrual bleeding
· Menstrual periods lasting more than a week
· Painful periods
· Pain during intercourse
· Pelvic pressure or pain
· Frequent urination
· Backache or leg pains

RISK FACTORS

There are few known risk factors for uterine fibroids, other than being a woman of reproductive age. Factors that can have an impact on fibroid development include:
· Genetics
· Onset of menstruation at an early age
· Obesity
· Vitamin D deficiency

COMPLICATIONS

Examples of uterine fibroids complications include:
· Anemia
· Infertility or pregnancy loss
· Placental abruption
· Fetal growth restriction
· Preterm delivery

Gene	Your Genotype	Your Result	Explanation
TNRC6B	AG	Higher Risk	Reports indicate that individuals with AG and GG genotypes of this gene tend to have a higher genetic risk of uterine fibroids.

Health Risk

Polycystic Ovary Syndrome

[Only applicable for female]

Polycystic ovary syndrome (PCOS) is a hormonal disorder common among women of reproductive age. Women with PCOS produce higher-than-normal amounts of male hormones (androgen). This hormone imbalance makes it harder for them to get pregnant. The ovaries may develop numerous small cysts (fluid-filled sacs) and fail to regularly release eggs.

8

variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of PCOS vary. A diagnosis of PCOS is made when patient experiences at least two of these signs:
· Irregular periods
· Excess androgen
· Polycystic ovaries

RISK FACTORS

Risk factors of getting PCOS include:
· Genetics
· Diet
· Obesity
· Excessive androgen production
· Excessive insulin production

COMPLICATIONS

Complications of PCOS can include:
· Infertility
· Gestational diabetes or pregnancy-induced high blood pressure
· Miscarriage or premature birth
· Nonalcoholic steatohepatitis
· Metabolic syndrome
· Type 2 diabetes or prediabetes
· Sleep apnea
· Depression, anxiety and eating disorders
· Abnormal uterine bleeding
· Cancer of the uterine lining

Health Risk

Polycystic Ovary Syndrome

Gene	Your Genotype	Your Result	Explanation
THADA	AA	Higher Risk	Reports indicate that individuals with CC and AC genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
LHCGR	AA	Higher Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of polycystic ovary syndrome.
FSHR	TC	Higher Risk	Reports indicate that individuals with CC and CT genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
C9orf3	GG	Higher Risk	Reports indicate that individuals with GG and GA genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
DENND1A	GG	Higher Risk	Reports indicate that individuals with GG and GA genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
YAP1	AA	Normal Risk	Reports indicate that individuals with GG and GA genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
RAB5B	AA	Normal Risk	Reports indicate that individuals with GG and GA genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
HMGA2	AA	Higher Risk	Reports indicate that individuals with AA and AC genotypes tend to have a higher genetic risk of polycystic ovary syndrome.
TOX3	TT	Normal Risk	Reports indicate that individuals with GG and GT genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
INSR	GG	Higher Risk	Reports indicate that individuals with GG and GA genotypes tend to have a lower genetic risk of polycystic ovary syndrome.
SUMO1P1	AG	Higher Risk	Reports indicate that individuals with AA and AG genotypes tend to have a higher genetic risk of polycystic ovary syndrome.

Health Risk

Gestational Diabetes

[Only applicable for female]

Gestational diabetes is high blood sugar (glucose) that develops during pregnancy and usually disappears after giving birth. It can happen at any stage of pregnancy, but is more common in the second or third trimester. It happens when your body cannot produce enough insulin – a hormone that helps control blood sugar levels – to meet your extra needs in pregnancy.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

For most women, gestational diabetes doesn't cause noticeable signs or symptoms. Increased thirst and more-frequent urination are possible symptoms.

RISK FACTORS

Some women have a greater risk of gestational diabetes. Risk factors for gestational diabetes include the following:
· Overweight and obesity
· A lack of physical activity
· Previous gestational diabetes or prediabetes
· Polycystic ovary syndrome
· Diabetes in an immediate family member.
· Previously delivering a baby weighing more than 4.1 kg

COMPLICATIONS

Complications that may affect the baby:
· Excessive birth weight
· Preterm birth
· Breathing difficulties
· Low blood sugar
· Obesity and type 2 diabetes later in life
· Stillbirth

Complications that may affect the mother:
· High blood pressure and preeclampsia
· Having a surgical delivery
· Future diabetes

Gene	Your Genotype	Your Result	Explanation
IGF2BP2	AC	Higher Risk	Individuals with AC and CC genotypes of this gene tend to have a higher genetic risk of gestational diabetes.
CDKAL1	GG	Normal Risk	Individuals with GC and CC genotypes of this gene tend to have a higher genetic risk of gestational diabetes.
MTNR1B	GC	Higher Risk	Individuals with GC and GG genotypes of this gene tend to have a higher genetic risk of gestational diabetes.

Health Risk

Intrahepatic Cholestasis of Pregnancy

[Only applicable for female]

Intrahepatic Cholestasis of Pregnancy (ICP) is a liver disorder that occurs in late pregnancy. This condition affects the normal flow of bile. Bile acids are chemicals that help with digestion. With ICP, the bile flow begins to slow down and the bile acids build up in the blood. The condition triggers intense itching, but without a rash. Itching usually occurs on the hands and feet but can also affect other parts of the body.

1

variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of ICP include:
· Intense itching, most noticeable on the hands and feet
· dark urine
· light-coloured bowel movements
· Nausea
· Loss of appetite
· Jaundice

RISK FACTORS

Some factors that may increase the risk of developing ICP include:
· Genetics
· History of liver damage or disease
· Being pregnant with twins or more

COMPLICATIONS

Complications that may affect the baby:
· Preterm birth
· Fetal distress
· Stillbirth

Complications that may affect the mother:
· Temporarily affect fat absorption
· Decreased levels of vitamin K (important factor involved in blood clotting)

Gene	Your Genotype	Your Result	Explanation
ABCB11	GG	Higher Risk	Individuals with AG and GG genotypes of this gene tend to have a higher genetic risk of intrahepatic cholestasis of pregnancy.

Health Risk

Erectile Dysfunction

[Only applicable for male]

Erectile dysfunction (impotence) is the inability to get and keep an erection firm enough for sex. Having erection trouble from time to time is not necessarily a cause for concern. If erectile dysfunction is an ongoing issue, however, it can cause stress, affect self-confidence and contribute to relationship problems. Problems getting or keeping an erection can also be a sign of an underlying health condition that needs treatment.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Erectile dysfunction symptoms might include persistent:
· Trouble getting an erection
· Trouble keeping an erection
· Reduced sexual desire

RISK FACTORS

Various risk factors can contribute to erectile dysfunction, including:
· Medical conditions
· Tobacco use
· Being overweight
· Certain medical treatments
· Injuries
· Psychological conditions
· Drug and alcohol use

COMPLICATIONS

Complications resulting from erectile dysfunction can include:
· An unsatisfactory sex life
· Stress or anxiety
· Embarrassment or low self-esteem
· Relationship problems
· The inability to get partner pregnant

Gene	Your Genotype	Your Result	Explanation
eNOS	--	Not Available	Individuals with TT genotype tend to have a higher genetic risk of erectile dysfunction.
eNOS	--	Not Available	Individuals with CC genotype tend to have a higher genetic risk of erectile dysfunction.
FSHR	--	Not Available	Individuals with GG genotype tend to have a higher genetic risk of erectile dysfunction.
VEGF	--	Not Available	Individuals with AA genotype tend to have a higher genetic risk of erectile dysfunction.

Health Risk

Postpartum Depression

[Only applicable for female]

A condition of major depression with onset within 4 weeks after childbirth is defined as postpartum depression. Most new moms experience postpartum "baby blues" after childbirth, which commonly include mood swings, crying spells, anxiety and difficulty sleeping. Baby blues typically begin within the first two to three days after delivery, and may last for up to two weeks. Nonetheless, most cases of postpartum depression can occur anytime within the first year after delivery as shown in common evidence. At least 1 in 10 postnatal women is affected by this highly prevalent psychopathology.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of postpartum depression may include:
· Sleep and appetite disturbance, depressed mood, feelings of worthlessness, impaired concentration and suicidal ideation.
· Notable psychomotor agitation and lack of energy
· Exaggerated mood change and preoccupation with infant well-being
· Anxiety
Prevention, care and support are therefore crucial to ensure emotional wellbeing of mothers owing to the severity of postpartum depression.

COMPLICATIONS

The adverse impacts are far-reaching, it affects not only the mothers but disrupts bonding between mother and infant and also between family members. Furthermore, growth and development of the infant in cognitive, language and emotional processing are affected by the persistent symptoms shown.

Rarely, an extreme mood disorder called postpartum psychosis also may develop after childbirth. Prevention, care and support are therefore crucial to ensure emotional wellbeing of mothers owing to the severity of postpartum depression.

Gene	Your Genotype	Your Result	Explanation
COMT	CC	Normal Risk	Reports indicate that individuals with the TT genotype tend to have a higher genetic risk of postpartum depression.
COMT	AA	Normal Risk	Reports indicate that individuals with the GG genotype tend to have a higher genetic risk of postpartum depression.
NR3C2	CC	Higher Risk	Reports indicate that individuals with the CC genotype tend to have a higher genetic risk of postpartum depression.
COMT	GG	Normal Risk	Reports indicate that individuals with the AA genotype tend to have a higher genetic risk of postpartum depression.

Health Risk

Postpartum Stretch Marks

[Only applicable for female]

Common skin conditions that appear initially as red and after that white lines on the skin are known as stretch marks or striae distensae. Scars of the dermis are represented by these lines, which are characterised by linear bundles of collagens situated parallel to the skin surface and the eventual loss of both elastin and collagen.

1

variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

RISK FACTORS

Risk factors of postpartum stretch marks may include:
· Excessive skin distension (such as that which occurs during pregnancy
· Growth spurts in puberty, or rapid weight gain)
· Prolonged exposure to cortisol (such as in individuals with Cushing syndrome)
· Genetic factors

COMPLICATIONS

Abdomen and breasts are 2 areas that are susceptible to developing stretch marks particularly during the pregnancy stage. The prevalence of stretch marks ranges from an estimated 43-88% in pregnant women. Even mild stretch marks can lead to stress to the bearer even though stretch marks only cause harms in extreme cases.

Gene	Your Genotype	Your Result	Explanation
ELN	TC	Higher Risk	Reports indicate that individuals with the CC or CT genotypes tend to have a higher genetic risk of postpartum stretch mark.
SRPX	GG	Normal Risk	Reports indicate that individuals with the AA or AG genotypes tend to have a higher genetic risk of postpartum stretch mark.
HMCN1	GG	Normal Risk	Reports indicate that individuals with the CC or CG genotypes tend to have a higher genetic risk of postpartum stretch mark.
TMEM18	AA	Normal Risk	Reports indicate that individuals with the GG or AG genotypes tend to have a higher genetic risk of postpartum stretch mark.

Health Risk

Postpartum Scarring

[Only applicable for female]

Skin scarring encompasses a range of clinical phenotypes from normal fine lines to unusual widespread, atrophic, hypertrophic, keloid scars and scar contractures. Protein MG53 involves wound healing promotion and scar formation reduction by facilitating cell membrane repair and myofibroblast differentiation control. It has been shown that this protein travels throughout the bloodstream and is responsible to assist the body to fix skin, heart, lungs, kidneys and other organs without causing scars.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

RISK FACTORS

A surgical procedure used to deliver an infant through incisions in the abdomen and uterus is known as Cesarean delivery (C-section). C sections can include planned, unexpected or emergency surgery procedures. Certain C-sections are important for the health of both the mother and the baby.

After a cesarean delivery, a C-section scar forms part of the normal recovery process as it is the usual and unavoidable outcome of mammalian tissue repair. Scarring also forms part of the natural healing process. Every wound, for example after an accident, disease or surgery leads to some degree of scarring except the very minor lesions. But,

Gene	Your Genotype	Your Result	Explanation
TP53	CG	Normal Risk	Reports indicate that individuals with the CC genotype tend to have a higher genetic risk of postpartum scarring.

Health Risk

Arachidonic Acid Deficiency

[Only applicable for female]

During exclusive breastfeeding, breast milk is the sole nutrition while in infant physical and cognitive development, polyunsaturated fatty acids (FAs) are the important micronutrients. Therefore, to ensure the infant gets the correct balance of both the macronutrients and micronutrients, the breastmilk's composition is crucial especially the important arachidonic (AA, 20:4n6) and docosahexaenoic (DHA, 22:6 n-3) acids.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

RISK FACTOR

Maternal genetics play an important role in influencing breastmilk's FA composition besides body composition and diet. The identification of FA fractions which are influenced by genetic variation based on the first genome-wide association studies (GWAS) on breastmilk composition. In fact, AA is the main FA in breastmilk influenced by genetic variation at the FADS1/2/3 locus.

COMPLICATIONS

Growth and immune function at the earliest ages are directly influenced by AA levels, and AA is an important structural component of neuronal and brain tissue. With that, it poses significant consequences for an individual's survival and fitness through adulthood.

Gene	Your Genotype	Your Result	Explanation
FADS1	TT	Normal Risk	Reports indicate that individuals with CC genotype tend to have lower level of arachidonic acid in the serum.
FADS2	AA	Normal Risk	Reports indicate that individuals with GG genotype tend to have lower level of arachidonic acid in the serum.

HEALTH RISK
BONE

Health Risk

Hip Fracture

A hip fracture is a break in the upper portion of the femur (thighbone). Most hip fractures occur in elderly patients whose bones have become weakened by osteoporosis. Medications, poor vision and balance problems also make older people more likely to fall — one of the most common causes of hip fracture.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of a hip fracture include:
· Inability to get up from a fall or to walk
· Severe pain in hip or groin
· Not being able to lift, move or rotate (turn) leg
· Not being able to stand or put weight on leg
· Bruising and swelling around hip
· Injured leg appears shorter
· Injured leg turning outwards

RISK FACTORS

Some factors that may increase the risk of hip fracture are:
· Age: bone density and muscle mass decreases with age.
· Gender: women are 3 times more likely to experience hip fracture than men.
· Osteoporosis
· Underlying health conditions
· Certain medications
· Nutritional problems
· Physical inactivity
· Tobacco and alcohol use

COMPLICATIONS

The complications can include:
· Blood clots in legs or lungs
· Bedsores
· Urinary tract infections
· Pneumonia
· Further loss of muscle mass, increasing risk of falls and injuries
· Death

Health Risk

Hip Fracture

Gene	Your Genotype	Your Result	Explanation
ZBTB40	GG	Normal Risk	Individuals with CG and CC genotypes of this gene tend to have a higher genetic risk of hip fracture.
WLS	TT	Higher Risk	Individuals with CT and TT genotypes of this gene tend to have a higher genetic risk of hip fracture.
DNM3	TT	Normal Risk	Individuals with GT and TT genotypes of this gene tend to have a lower genetic risk of hip fracture.
GALNT3	AG	Normal Risk	Individuals with GA and AA genotypes of this gene tend to have a lower genetic risk of hip fracture.
CTNNB1	TC	Normal Risk	Individuals with CT and TT genotypes of this gene tend to have a lower genetic risk of hip fracture.
MEPE	TG	Higher Risk	Individuals with GT and TT genotypes of this gene tend to have a higher genetic risk of hip fracture.
MEF2C	AC	Higher Risk	Individuals with CA and AA genotypes of this gene tend to have a higher genetic risk of hip fracture.

Health Risk

Lumbar Spine Stenosis

Spinal stenosis is a narrowing of the spaces within the spine, which can put pressure on the nerves that travel through the spine. Spinal stenosis occurs most often in the lower back (lumbar) and the neck. Spinal stenosis is most commonly caused by wear-and-tear changes in the spine related to osteoarthritis. In severe cases of spinal stenosis, doctors may recommend surgery to create additional space for the spinal cord or nerves.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of a lumbar spine stenosis include:
· Numbness or tingling in a foot or leg
· Weakness in a foot or leg
· Pain or cramping in one or both legs when standing for long periods of time
· Back pain

RISK FACTORS

Some of the factors that may increase the risk of lumbar spine stenosis are:
· Being over the age of 50
· Trauma
· Scoliosis
· Genetics

COMPLICATIONS

Rarely, untreated severe spinal stenosis may progress and cause permanent issues, such as:
· Numbness
· Weakness
· Balance problems
· Incontinence
· Paralysis

Gene	Your Genotype	Your Result	Explanation
ZBTB40	GG	Normal Risk	Individuals with CG and CC genotypes of this gene tend to have a higher genetic risk of lumbar spine stenosis.
WLS	TT	Higher Risk	Individuals with CT and TT genotypes of this gene tend to have a higher genetic risk of lumbar spine stenosis.
SPTBN1	GG	Normal Risk	Individuals with CG and CC genotypes of this gene tend to have a higher genetic risk of lumbar spine stenosis.
GALNT3	AG	Normal Risk	Individuals with GA and AA genotypes of this gene tend to have a lower genetic risk of lumbar spine stenosis.
CTNNB1	TC	Normal Risk	Individuals with CT and TT genotypes of this gene tend to have a lower genetic risk of lumbar spine stenosis.
MEPE	TG	Higher Risk	Individuals with GT and TT genotypes of this gene tend to have a higher genetic risk of lumbar spine stenosis.
IDUA	AG	Normal Risk	Individuals with CT and TT genotypes of this gene tend to have a lower genetic risk of lumbar spine stenosis.

Health Risk

Osteoporosis

Osteoporosis causes bones to become weak and brittle — so brittle that a fall or even mild stresses such as bending over or coughing can cause a fracture. Osteoporosis-related fractures most commonly occur in the hip, wrist or spine.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms include:
· Back pain, caused by a fractured or collapsed vertebra
· Loss of height over time
· A stooped posture
· A bone that breaks much more easily than expected

RISK FACTORS

Risk factors for osteoporosis include:
· Inadequate amounts of dietary calcium
· Low vitamin D levels
· Cigarette smoking
· Alcohol intake of more than two standard drinks per day
caffeine intake of more than three cups of coffee or equivalent per day.
· Lack of physical activity.
· Early menopause (before the age of 45)

COMPLICATIONS

Bone fractures, particularly in the spine or hip, are the most serious complications of osteoporosis. Hip fractures often are caused by a fall and can result in disability and even an increased risk of death within the first year after the injury.

Gene	Your Genotype	Your Result	Explanation
FONG	AC	Higher Risk	Individuals with AA and AC genotypes tend to have a higher genetic risk of osteoporosis.
ALDH7A1	AA	Normal Risk	Individuals with GG and AG genotypes tend to have a higher genetic risk of osteoporosis.

HEALTH RISK

EAR

Health Risk

Age-related Hearing Impairment

Hearing loss that occurs gradually with age (presbycusis) is common. For those older than 75, that number is approximately 1 in 2. Hearing loss is defined as one of three types: conductive (involves outer or middle ear), sensorineural (involves inner ear) and mixed (combination of the two). Aging and chronic exposure to loud noises both contribute to hearing loss.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of hearing loss may include:
· Muffling of speech and other sounds
· Difficulty understanding words
· Trouble hearing consonants
· Frequently asking others to speak more slowly, clearly and loudly
· Needing to turn up the volume of the television or radio
· Withdrawal from conversations
· Avoidance of some social settings

RISK FACTORS

Factors that may damage or lead to loss of nerve cells in the inner ear include:
· Aging
· Loud noise
· Genetics
· Occupational noises
· Recreational noises

COMPLICATIONS

Complications that could occur are:
· Depression
· Feelings of isolation
· Cognitive impairment

Gene	Your Genotype	Your Result	Explanation
TNFRSF1B	AG	Higher Risk	Individuals with AA and AG genotypes of this gene tend to have a higher genetic risk of age-related hearing impairment.
TNF-α	CC	Normal Risk	Individuals with AA and AC genotypes of this gene tend to have a higher genetic risk of age-related hearing impairment.

Health Risk

Otosclerosis

Otosclerosis is a condition where the third bone of hearing does not move as well as it should. This causes conductive hearing loss. If the condition is mild, treatment may not be necessary. When hearing loss becomes more advanced, hearing aids or surgery may be effective treatment options.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Symptoms of otosclerosis include:
· Hearing loss that gradually worsen over time
· Difficulty in hearing low, deep sounds and whispers
· Speaking quietly because own voice sounds loud
· Finding it easier to hear when there is background noise
· Tinnitus
· Dizziness

RISK FACTORS

These risk factors may cause otosclerosis:
· Age: between the age of 10 and 45
· Genetics
· Race and ethnicity: Caucasians are more likely to get it

COMPLICATIONS

Complications may include:
· Complete deafness
· Funny taste in the mouth or loss of taste to part of the tongue, temporary or permanent
· Infection, dizziness, pain, or a blood clot in the ear after surgery
· Nerve damage

Gene	Your Genotype	Your Result	Explanation
RELN	TT	Higher Risk	Individuals with TT genotype tend to have a higher genetic risk of otosclerosis.
BMP4	AA	Normal Risk	Individuals with AG and GG genotypes tend to have a higher genetic risk of otosclerosis.

HEALTH RISK

NOSE

Health Risk

Rhinosinusitis

Rhinosinusitis occurs when the spaces inside the nose and head (sinuses) are swollen and inflamed for three months or longer, despite treatment. Chronic sinusitis can be brought on by an infection, by growths in the sinuses (nasal polyps) or swelling of the lining of the sinuses.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Common signs and symptoms of rhinosinusitis include:
· Nasal inflammation
· Thick, discolored discharge from the nose
· Drainage down the back of the throat
· Nasal obstruction or congestion, causing difficulty breathing through the nose
· Pain, tenderness and swelling around the eyes, cheeks, nose or forehead
· Reduced sense of smell and taste

RISK FACTORS

Increased risk of getting rhinosinusitis:
· A deviated septum
· Nasal polyps
· Asthma
· Aspirin sensitivity
· A dental infection
· An immune system disorder such as HIV/AIDS or cystic fibrosis
· Hay fever or another allergic condition
· Regular exposure to pollutants such as cigarette smoke

COMPLICATIONS

Serious complications of rhinosinusitis complications are rare, but may include:
· Vision problems
· Infections

Gene	Your Genotype	Your Result	Explanation
RYBP	CC	Normal Risk	Individuals with TT and TC genotypes tend to have a higher genetic risk of rhinosinusitis.
AOAH	TC	Normal Risk	Individuals with TC and CC genotypes tend to have a lower genetic risk of rhinosinusitis.

HEALTH RISK
MOUTH

Health Risk

Dental Caries

Cavities are permanently damaged areas in the hard surface of the teeth that develop into tiny openings or holes. Cavities, also called tooth decay or caries, are caused by a combination of factors, including bacteria in the mouth, frequent snacking, sipping sugary drinks and not cleaning teeth well.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The signs and symptoms of cavities include:
· Toothache
· Tooth sensitivity
· Mild to sharp pain when eating or drinking something sweet, hot or cold
· Visible holes or pits in the teeth
· Brown, black or white staining on any surface of a tooth
· Pain when biting down

RISK FACTORS

The following factors can increase the risk:
· Tooth location
· Certain foods and drinks
· Frequent snacking or sipping
· Bedtime infant feeding
· Inadequate brushing
· Not getting enough fluoride
· Younger or older age
· Dry mouth
· Heartburn
· Eating disorders

COMPLICATIONS

Complications of cavities may include:
· Pain
· Tooth abscess
· Swelling or pus around a tooth
· Damage or broken teeth
· Chewing problems

Gene	Your Genotype	Your Result	Explanation
VDR	CC	Normal Risk	Reports indicate that individuals with TC and TT genotypes tend to have a higher genetic risk of dental carries.
DLX3	AA	Higher Risk	Reports indicate that individuals with AG and AA genotypes tend to have a higher genetic risk of dental carries.

Health Risk

Periodontitis

Periodontitis, also called gum disease, is a serious gum infection that damages the soft tissue and, without treatment, can destroy the bone that supports the teeth. Periodontitis can cause teeth to loosen and lead to tooth loss.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of periodontitis can include:
· Swollen or puffy gums
· Bright red, dusky red or purplish gums
· Gums that feel tender when touched and bleed easily
· Spitting out blood when brushing or flossing the teeth
· Bad breath
· Pus between the teeth and gums
· Loose teeth or loss of teeth
· Painful chewing
· New spaces developing between the teeth

RISK FACTORS

Factors that can increase risk of periodontitis include:
· Gingivitis
· Poor oral health habits
· Smoking or chewing tobacco
· Hormonal changes, such as those related to pregnancy or menopause
· Obesity
· Inadequate nutrition
· Genetics
· Certain medications that cause dry mouth or gum changes
· Conditions that cause decreased immunity, such as leukemia, HIV/AIDS and cancer treatment
· Certain diseases, such as diabetes, rheumatoid arthritis and Crohn's disease

COMPLICATIONS

Periodontitis can cause tooth loss. The bacteria responsible for periodontitis can enter the bloodstream through gum tissue, possibly affecting other parts of the body. Periodontitis is linked with this diseases:
· Respiratory disease
· Rheumatoid arthritis
· Coronary artery disease
· Problems controlling blood sugar in diabetes

Gene	Your Genotype	Your Result	Explanation
CSF1	GG	Normal Risk	Individuals with AA and AG genotypes of this gene tend to have a higher genetic risk of periodontitis.
KCNQ5	AA	Higher Risk	Individuals with AG and GG genotypes of this gene tend to have a lower genetic risk of periodontitis.
GPR141	TC	Normal Risk	Individuals with TC and TT genotypes of this gene tend to have a lower genetic risk of periodontitis.

HEALTH RISK
MENTAL

Health Risk

ADHD

ADHD is a mental health disorder that includes a combination of persistent problems, such as difficulty paying attention, hyperactivity and impulsive behavior. Adult ADHD can lead to unstable relationships, poor work or school performance, low self-esteem, and other problems.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Adult ADHD symptoms may include:
· Impulsiveness
· Disorganization and problems prioritizing
· Poor planning and time management skills
· Problems focusing on a task
· Trouble multitasking
· Excessive activity or restlessness
· Low frustration tolerance
· Frequent mood swings
· Hot temper
· Trouble coping with stress

RISK FACTORS

Risk of ADHD may increase if:
· Genetics
· Exposed to environmental toxins as a child
· Born prematurely

COMPLICATIONS

ADHD has been linked to:
· Poor school or work performance
· Substance abuse
· Poor physical and mental health
· Poor self-image
· Delinquent or risky behavior
· Sleep problems

Gene	Your Genotype	Your Result	Explanation
BCL11A	GG	Normal Risk	Individuals with TT and GT genotypes tend to have a higher genetic risk of ADHD.
ELOVL6	TC	Higher Risk	Individuals with TT and CT genotypes tend to have a higher genetic risk of ADHD.
EMP2	AG	Higher Risk	Individuals with AA and AG genotypes tend to have a higher genetic risk of ADHD.
ITGA1	AG	Higher Risk	Individuals with AA and AG genotypes tend to have a higher genetic risk of ADHD.

Health Risk

Depression

Depression is a mood disorder that causes a persistent feeling of sadness and loss of interest. Also called major depressive disorder or clinical depression, it affects how one feels, thinks and behaves and can lead to a variety of emotional and physical problems. Patients may have trouble doing normal day-to-day activities, and sometimes may feel as if life is not worth living.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Symptoms of depression include:
- Feelings of sadness, tearfulness, emptiness or hopelessness
- Angry outbursts, irritability or frustration
- Loss of interest or pleasure in most or all normal activities
- Sleep disturbances
- Tiredness and lack of energy
- Reduced appetite and weight loss
- Anxiety, agitation or restlessness
- Slowed thinking, speaking or body movements
- Feelings of worthlessness or guilt, fixating on past failures or self-blame
- Trouble thinking, concentrating, making decisions and remembering things
- Frequent or recurrent thoughts of death and suicide
- Unexplained physical problems, such as back pain or headaches

RISK FACTORS

Risk factors of depression may increase due to:
- Genetics
- Brain chemistry
- Certain medical conditions
- Substance use
- Stress
- Poor nutrition

COMPLICATIONS

Examples of complications that are associated with depression are:
- Gaining excess weight or becoming obese
- Experiencing physical illness and pain
- Substance abuse
- Suffering from panic attacks, social phobia or anxiety
- Having suicidal thoughts, attempting suicide or committing suicide
- Mutilating oneself through cutting or other means

Gene	Your Genotype	Your Result	Explanation
ATXN7L2	TT	Normal Risk	Individuals with GG and GT genotypes tend to have a higher genetic risk of depression.
CCBE1	TT	Normal Risk	Individuals with CC and CT genotypes tend to have a higher genetic risk of depression.
GNAI3	CC	Normal Risk	Individuals with TT and CT genotypes tend to have a higher genetic risk of depression.
PAX5	AG	Normal Risk	Individuals with GG and GA genotypes tend to have a lower genetic risk of depression.

Health Risk

Autism

Autism spectrum disorder is a condition related to brain development that impacts how a person perceives and socializes with others, causing problems in social interaction and communication. The disorder also includes limited and repetitive patterns of behavior.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Individual with autism spectrum disorder may have these social communication/interaction problems:

· Fails to respond to his or her name
· Resists cuddling and holding
· Has poor eye contact and lacks facial expression
· Does not speak or has delayed speech
· Cannot start a conversation or keep one going
· Speaks with an abnormal tone or rhythm
· Repeats words or phrases verbatim
· Does not appear to understand simple questions or directions
· Inappropriately approaches a social interaction by being passive, aggressive or disruptive

RISK FACTORS

Autism spectrum disorder's risk factors are:

· Genetics
· Extremely preterm babies
· Being born to older parents
· Exposure to heavy metals and environmental toxins

COMPLICATIONS

Long-term effects of autism can include:

· Social isolation
· Difficulty forming and maintaining friendships
· Difficulty relating and empathizing with other people
· Some may have trouble living independently
· Sleep problems

Gene	Your Genotype	Your Result	Explanation
SEMA5A	TT	Higher Risk	Individuals with CC and CT genotypes tend to have a lower genetic risk of autism.
TBL1Y	--	Not Available	Individuals with GG and GA genotypes tend to have a higher genetic risk of autism.
DDX3Y	--	Not Available	Individuals with AA and CA genotypes tend to have a higher genetic risk of autism.
UTY	--	Not Available	Individuals with AA and GA genotypes tend to have a higher genetic risk of autism.

Health Risk

Panic Disorder

A panic attack is a sudden episode of intense fear that triggers severe physical reactions when there is no real danger or apparent cause. Panic attacks can be very frightening. When panic attacks occur, there might be a feeling of losing control, having a heart attack or even dying.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Panic attacks typically include some of these signs or symptoms:
· Sense of impending doom or danger
· Fear of loss of control or death
· Rapid, pounding heart rate
· Sweating
· Trembling or shaking
· Shortness of breath or tightness in the throat
· Chills
· Hot flashes
· Nausea
· Abdominal cramping
· Chest pain
· Headache
· Dizziness, lightheadedness or faintness
· Numbness or tingling sensation

RISK FACTORS

Factors that may increase the risk of developing panic disorder include:
· Genetics
· Major life stress
· A traumatic event
· Major changes in life
· Smoking or excessive caffeine intake
· History of childhood physical or sexual abuse

COMPLICATIONS

Complications that panic attacks may be linked to include:
· Development of specific phobias
· Frequent medical care for health concerns and other medical conditions
· Avoidance of social situations
· Problems at work or school
· Depression, anxiety disorders and other psychiatric disorders
· Increased risk of suicide or suicidal thoughts
· Substance abuse

Gene	Your Genotype	Your Result	Explanation
CNTN4	CC	Normal Risk	Individuals with TT and CT genotypes tend to have a higher genetic risk of panic disorder.

Health Risk

Eating Disorder

Eating disorders are serious conditions related to persistent eating behaviors that negatively impact health, emotions and ability to function in important areas of life. The most common eating disorders are anorexia nervosa, bulimia nervosa and binge-eating disorder.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Red flags that may indicate an eating disorder include:
· Skipping meals or making excuses for not eating
· Adopting an overly restrictive vegetarian diet
· Excessive focus on healthy eating
· Withdrawing from normal social activities
· Persistent worry or complaining about being fat and talk of losing weight
· Frequent checking in the mirror for perceived flaws
· Repeatedly eating large amounts of sweets or high-fat foods
· Use of laxatives or herbal products for weight loss
· Excessive exercise
· Calluses on the knuckles from inducing vomiting

RISK FACTORS

Certain factors may increase the risk of developing an eating disorder, including:
· Genetics
· Other mental health disorders
· Dieting and starvation
· Stress

COMPLICATIONS

Some examples of eating disorders complications are:
· Serious health problems
· Depression and anxiety
· Suicidal thoughts or behavior
· Problems with growth and development
· Social and relationship problems
· Work and school issues
· Death

Gene	Your Genotype	Your Result	Explanation
CAMK1D	TC	Normal Risk	Individuals with C genotype tend to have a lower genetic risk of eating disorder.
EMP2	GG	Normal Risk	Individuals with G genotype tend to have a lower genetic risk of eating disorder.
FLG-AS1	AG	Normal Risk	Individuals with A genotype tend to have a lower genetic risk of eating disorder.
Intergenic	AA	Normal Risk	Individuals with T genotype tend to have a lower genetic risk of eating disorder.
MACROD2	AA	Normal Risk	Individuals with A genotype tend to have a lower genetic risk of eating disorder.

Health Risk

Tourette Syndrome

Tourette syndrome is a disorder that involves repetitive movements or unwanted sounds (tics) that cannot be easily controlled. For instance, repeatedly blinking eyes, shrugging shoulders or blurt out unusual sounds or offensive words.

3 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Tics are classified as:
· Simple tics: sudden, brief and repetitive tics involve a limited number of muscle groups.
· Complex tics: distinct, coordinated patterns of movements involve several muscle groups.

Some examples of tics signs:
· Eye blinking
· Head jerking
· Shoulder shrugging
· Touching or smelling objects
· Nose twitching

RISK FACTORS

Risk factors for Tourette syndrome include:
· Genetics
· Gender: Males are about three to four times more likely than females to develop Tourette syndrome

COMPLICATIONS

Conditions often associated with Tourette syndrome include:
· Attention-deficit/hyperactivity disorder (ADHD)
· Obsessive-compulsive disorder (OCD)
· Autism spectrum disorder
· Learning disabilities
· Sleep disorders
· Depression
· Anxiety disorders
· Pain related to tics
· Anger-management problems

Gene	Your Genotype	Your Result	Explanation
COL27A1	GG	Normal Risk	Individuals with AA and AG genotypes tend to have a higher genetic risk of Tourette syndrome.
Intergenic	TT	Higher Risk	Individuals with AA and AC genotypes tend to have a higher genetic risk of Tourette syndrome.
LINC00351	AG	Higher Risk	Individuals with GG and AG genotypes tend to have a higher genetic risk of Tourette syndrome.
POLR3B	TC	Higher Risk	Individuals with TT and TC genotypes tend to have a higher genetic risk of Tourette syndrome.

Health Risk

Chronic Fatigue Syndrome

Chronic fatigue syndrome (CFS) is a complicated disorder characterized by extreme fatigue that lasts for at least six months and can't be fully explained by an underlying medical condition. The fatigue worsens with physical or mental activity, but does not improve with rest.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of chronic fatigue syndrome may include:
· Sleep problems
· Problems with memory or concentration
· Sore throat
· Headaches
· Enlarged lymph nodes in neck or armpits
· Unexplained muscle or joint pain
· Unrefreshing sleep
· Extreme exhaustion after physical or mental exercise

RISK FACTORS

Factors that may increase risk of chronic fatigue syndrome include:
· Age: most commonly affects young to middle-aged adults
· Gender: women are diagnosed with chronic fatigue syndrome much more often than men

COMPLICATIONS

Possible complications of chronic fatigue syndrome include:
· Lifestyle restrictions
· Increased work absences
· Social isolation
· Depression

Gene	Your Genotype	Your Result	Explanation
CFB	TT	Normal Risk	Individuals with AA and AT genotypes tend to have a higher genetic risk of chronic fatigue syndrome.
CFH	AC	Higher Risk	Individuals with AA and AC genotypes tend to have a higher genetic risk of chronic fatigue syndrome.
CXCL16	AG	Higher Risk	Individuals with GG and AG genotypes tend to have a higher genetic risk of chronic fatigue syndrome.

Health Risk

Obsessive Compulsive Disorder

Obsessive-compulsive disorder (OCD) features a pattern of unwanted thoughts and obsessions that lead to repetitive behaviors (compulsions). These obsessions and compulsions interfere with daily activities and cause significant distress.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Obsessions often have themes to them, such as:
· Fear of contamination or dirt
· Doubting and having difficulty tolerating uncertainty
· Needing things orderly and symmetrical
· Unwanted thoughts, including aggression, sexual or religious subjects

RISK FACTORS

Risk factors of OCD include:
· Genetics
· Stressful life events
· Other mental health disorders

COMPLICATIONS

Problems resulting from OCD may include:
· Excessive time spent engaging in ritualistic behaviors
· Health issues, such as contact dermatitis from frequent hand-washing
· Difficulty attending work, school or social activities
· Troubled relationships
· Overall poor quality of life

Gene	Your Genotype	Your Result	Explanation
TPH2	TT	Normal Risk	Individuals with GG genotype tend to have a higher genetic risk of OCD.
TPH2_2	CC	Higher Risk	Individuals with CC genotype tend to have a higher genetic risk of OCD.
SLC6A4	--	Not Available	Individuals with GG and GA genotypes tend to have a higher genetic risk of OCD.

HEALTH RISK

OTHERS

Health Risk

Rheumatoid Arthritis

Rheumatoid arthritis (RA) is a chronic inflammatory disease of unknown cause. It is an autoimmune disease that attacks the synovium (soft tissue that lines the joint). It is characterized by joint inflammation in multiple joints at different parts of the body, eventually result in bone erosion and joint deformity. The inflammation associated with rheumatoid arthritis can damage other parts of the body as well. While new types of medications have improved treatment options dramatically, severe rheumatoid arthritis can still cause physical disabilities.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of rheumatoid arthritis may include:
· Tender, warm, swollen joints
· Joint stiffness that is usually worse in the mornings and after inactivity
· Fatigue, fever and loss of appetite
Early rheumatoid arthritis tends to affect your smaller joints first, particularly the joints that attach your fingers to your hands and your toes to your feet. As the disease progresses, symptoms often spread to the wrists, knees, ankles, elbows, hips and shoulders. In most cases, symptoms occur in the same joints on both sides of your body.

RISK FACTORS

Certain factors may increase your risk of developing rheumatoid arthritis, these include your gender (women are more likely than men to develop rheumatoid arthritis), age, smoking, environmental exposures, obesity and a family history of RA.

COMPLICATIONS

Rheumatoid arthritis may lead to the following complications:
· Osteoporosis
· Rheumatoid nodules
· Dry eyes and mouth
· Infections
· Abnormal body composition
· Carpal tunnel syndrome
· Heart problems
· Lung disease
· Lymphoma

Gene	Your Genotype	Your Result	Explanation
CDK5RAP2	AG	Higher Risk	When this locus on the gene carries G allele, its risk is 1.5 times more than those carrying A allele.
DPP4	TC	Higher Risk	This variant is located on the DPPD4 gene and data indicates that carrying T allele will increase the risk of rheumatoid arthritis.
STAT4	TG	Moderate Risk	This variant is located on the STAT4 gene and data indicates that carrying T allele will increase the risk of rheumatoid arthritis.
CCR6	AG	Normal Risk	The A allele in this locus may be associated with the expression of the CCR6 gene, thus increasing the risk of rheumatoid arthritis.
RTKN2	CC	Normal Risk	T allele causes NFKBIE to participate in the encoded protein structural changes that increases the risk of rheumatoid arthritis.

Health Risk

Gout

Gout is a general term for a variety of conditions caused by a buildup of uric acid. Gout commonly affects the big toe joint, ankle joint, knee joint and so on. The prevalence is higher in men than in women- 95% of gout patients are male. Women generally do not develop gout before the age of 50, but have a higher risk of developing it after menopause.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The signs and symptoms of gout almost always occur suddenly, and often at night. They include:
· Intense joint pain
· Lingering discomfort lasting for a few days to weeks
· Inflammation and redness
· Limited range of motion

RISK FACTOR

You are more likely to develop gout if you have high levels of uric acid in your body. Factors that increase the uric acid level in your body include:
· Diet: eating a diet rich in meat and seafood; consumption of alcohol and beverages sweetened with fruit sugar (fructose)
· Obesity
· Family history of gout
· Age and gender: gout occurs more often in middle-aged men and postmenopausal women
· Recent surgery or trauma

COMPLICATIONS

People with gout can develop more severe conditions, such as:
· Recurrent gout
· Advanced gout
· Kidney stones

Gene	Your Genotype	Your Result	Explanation
Intergenic	TC	Higher Risk	When this site carries the T allele, the association with gout is stronger.
ABCG2	GG	Normal Risk	The G>T mutation at this site results in abnormal protein structure and reduces protein expression causing difficulty in uric acid transport, thereby increasing the risk of developing gout.
ALPK1	TC	Normal Risk	When the genotype is CC, it may increase the risk of gout.
SLC22A12	CC	Normal Risk	When this site carries the T allele, it causes more uric acid in the kidney to be absorbed into the blood.

Health Risk

Scoliosis

Scoliosis is where the spine twists and curves to the side. It can affect people of any age, from babies to adults, but most often starts in children aged 10 to 15. Scoliosis can improve with treatment, but it is not usually a sign of anything serious and treatment is not always needed if it is mild. However, severe scoliosis can be disabling. An especially severe spinal curve can reduce the amount of space within the chest, eventually impairing lung function.

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Signs and symptoms of scoliosis may include:
· Uneven shoulders
· One shoulder blade that appears more prominent than the other
· Uneven waist
· One hip higher than the other
If a scoliosis curve gets worse, the spine will also rotate or twist, in addition to curving side to side. This causes the ribs on one side of the body to stick out farther than on the other side.

CAUSES

Doctors do not know what causes the most common type of scoliosis — although it appears to involve hereditary factors, because the disorder tends to run in families. Less common types of scoliosis may be caused by:
· Neuromuscular conditions, such as cerebral palsy or muscular dystrophy
· Birth defects affecting the development of the bones of the spine
· Injuries to or infections of the spine

RISK FACTOR

Risk factors for developing the most common type of scoliosis include:
· Age: signs and symptoms typically begin during the growth spurt that occurs just prior to puberty
· Gender: although both boys and girls develop mild scoliosis at about the same rate, girls have a much higher risk of the curve worsening and requiring treatment
· Family history: scoliosis can run in families, but most children with scoliosis do not have a family history of the disease

COMPLICATIONS

While most people with scoliosis have a mild form of the disorder, scoliosis may sometimes cause complications, including:
· Lung and heart damage
· Back problems
· Appearance: uneven hips and shoulders, prominent ribs, and a shift of the waist and trunk to the side. Individuals with scoliosis often become self-conscious about their appearance

Gene	Your Genotype	Your Result	Explanation
LBX1	TT	Higher Risk	The data demonstrates that carrying T allele can increase the risk of scoliosis.

Health Risk

Restless Legs Syndrome

Restless legs syndrome (RLS) is mainly characterized by unpleasant or uncomfortable sensations in the legs at rest, with a strong desire to move the legs. It typically happens in the evening or nighttime hours when you are sitting or lying down. Moving eases the unpleasant feeling temporarily. Simple self-care steps and lifestyle changes may help relieve the symptoms. Medications also help many people with RLS.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The chief symptom is an urge to move the legs. Common accompanying characteristics of RLS include:

- Sensations that begin after rest. The sensation typically begins after you have been lying down or sitting for an extended time, such as in a car, airplane or movie theater
- Relief with movement. The sensation of RLS lessens with movement, such as stretching, jiggling your legs, pacing or walking
- Worsening of symptoms in the evening. Symptoms occur mainly at night
- Nighttime leg twitching. RLS may be associated with another, more common condition called periodic limb movement of sleep, which causes your legs to twitch and kick, possibly throughout the night, while you sleep

RISK FACTORS

RLS can develop at any age, even during childhood. The disorder is more common with increasing age and more common in women than in men. Restless legs syndrome usually is not related to a serious underlying medical problem. However, RLS sometimes accompanies other conditions, such as:

- Peripheral neuropathy. This damage to the nerves in your hands and feet is sometimes due to chronic diseases such as diabetes and alcoholism
- Iron deficiency. Even without anemia, iron deficiency can cause or worsen RLS. If you have a history of bleeding from your stomach or bowels, experience heavy menstrual periods, or repeatedly donate blood, you may have iron deficiency
- Kidney failure. If you have kidney failure, you may also have iron deficiency, often with anemia. When kidneys do not function properly, iron stores in your blood can decrease. This and other changes in body chemistry may cause or worsen RLS
- Spinal cord conditions. Lesions on the spinal cord have been linked to RLS. Having had anesthesia to the spinal cord, such as a spinal block, also increases the risk of developing RLS

COMPLICATIONS

Although RLS does not lead to other serious conditions, symptoms can range from barely bothersome to incapacitating. Many people with RLS find it difficult to fall or stay asleep. Severe RLS can cause marked impairment in life quality and can result in depression. Insomnia may lead to excessive daytime drowsiness, but RLS may interfere with napping.

Gene	Your Genotype	Your Result	Explanation
BTBD9	AG	Moderate Risk	BTBD9 gene plays a role in regulating iron homeostasis. This variant is located on the BTBD9 gene.

Health Risk

Alcohol Dependence

Alcohol dependence (chronic alcoholism) is caused by prolonged excessive alcohol use that severely disturbs the central nervous system. Thus, a person with alcohol dependence may not be able to control their actions. Experts have tried to pinpoint factors like genetics, gender, race, or socioeconomics that may predispose someone to alcohol addiction. But it has no single cause. Psychological, genetic, and behavioral factors can all contribute to having the disease.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Alcohol use disorder can be mild, moderate or severe, based on the number of symptoms you experience. Signs and symptoms may include:

- Being unable to limit the amount of alcohol you drink
- Wanting to cut down on how much you drink or making unsuccessful attempts to do so
- Spending a lot of time drinking, getting alcohol or recovering from alcohol use
- Feeling a strong craving or urge to drink alcohol
- Failing to fulfill major obligations at work, school or home due to repeated alcohol use
- Continuing to drink alcohol even though you know it is causing physical, social or interpersonal problems
- Giving up or reducing social and work activities and hobbies
- Using alcohol in situations where it is not safe, such as when driving or swimming
- Developing a tolerance to alcohol so you need more to feel its effect or you have a reduced effect from the same amount
- Experiencing withdrawal symptoms - such as nausea, sweating and shaking - when you do not drink, or drinking to avoid these symptoms

RISK FACTORS

Alcohol use may begin in the teens, but alcohol use disorder occurs more frequently in the 20s and 30s, though it can start at any age.

- Steady drinking over time
- Starting at an early age
- Family history
- Depression and other mental health problems
- History of trauma
- Having bariatric surgery
- Social and cultural factors

COMPLICATION

Alcohol depresses your central nervous system. In some people, the initial reaction may be stimulation. But as you continue to drink, you become sedated. Too much alcohol affects your speech, muscle coordination and vital centers of your brain. A heavy drinking binge may even cause a life-threatening coma or death. This is of particular concern when you are taking certain medications that also depress the brain's function.

Gene	Your Genotype	Your Result	Explanation
ALDH2	GG	Higher Risk	When this site carries G allele, an amino acid lysine is replaced with glutamic acid on ALDH2, while the aldehyde dehydrogenase is still well-functioning. Alcohol drinking will not produce adverse reactions easily but may lead to alcohol dependence.
DRD2	AA	Higher Risk	G>A mutation at this site increases the risk of alcohol dependence.

Health Risk

Bipolar Disorder

Bipolar disorder is characterized by extreme mood swings. These can range from extreme highs (mania) to extreme lows (depression). Episodes of mania and depression often last for several weeks or months. These mood swings can affect sleep, energy, activity, judgment, behavior and the ability to think clearly.

Although bipolar disorder is a lifelong condition, management of mood swings and other symptoms is possible by following a treatment plan. In most cases, bipolar disorder is treated with medications and psychological counseling (psychotherapy).

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

There are several types of bipolar and related disorders. They may include mania or hypomania and depression. Symptoms can cause unpredictable changes in mood and behavior, resulting in significant distress and difficulty in life.

Bipolar I disorder: You have had at least one manic episode that may be preceded or followed by hypomanic or major depressive episodes. In some cases, mania may trigger a break from reality (psychosis).
Bipolar II disorder: You have had at least one major depressive episode and at least one hypomanic episode, but you have never had a manic episode.
Cyclothymic disorder: You have had at least two years — or one year in children and teenagers — of many periods of hypomania symptoms and periods of depressive symptoms (though less severe than major depression).
Other types: These include, for example, bipolar and related disorders induced by certain drugs or alcohol or due to a medical condition, such as Cushing's disease, multiple sclerosis or stroke.

RISK FACTORS

Factors that may increase the risk of developing bipolar disorder or act as a trigger for the first episode include:
· Having a first-degree relative, such as a parent or sibling, with bipolar disorder
· Periods of high stress, such as the death of a loved one or other traumatic event
· Drug or alcohol abuse

COMPLICATIONS

Left untreated, bipolar disorder can result in serious problems that affect every area of your life, such as:
· Problems related to drug and alcohol use
· Suicide or suicide attempts
· Legal or financial problems
· Damaged relationships
· Poor work or school performance

Gene	Your Genotype	Your Result	Explanation
ZNF804A	AC	Higher Risk	AA genotype in this gene locus may increase the expression of ZNF804A, resulting in abnormalities in white matter density/volume, which leads to an increased risk of disease.
CACNA1C	GG	Normal Risk	The G>A mutation in this gene locus may result in dysfunction of CaV1.2 calcium channel protein products and thus is associated with bipolar disorder.
ANK3	CC	Normal Risk	The C>T mutation in this gene locus may affect the expression of ANK3 gene, which in turn affects occurrence of neuronal axon polarity and nerve excitability.

Health Risk

Hypersomnia

Hypersomnia refers to excessive sleep during the day. This type of excessive sleep is not due to the lack of sleep, alcohol, drugs, physical illness or a mental disorder (such as depression). The cause is currently unclear, but it is often related to psychological factors.

2 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The main symptom of hypersomnia is constant tiredness. People with hypersomnia may take naps throughout the day without ever relieving drowsiness. They also have difficulty waking from long periods of sleep. Other symptoms of hypersomnia include:

· Low energy
· Irritability
· Anxiety
· Loss of appetite
· Slow thinking or speech
· Difficulty remembering
· Restlessness
· Fall asleep during the day, often while eating or talking

RISK FACTOR

People with conditions that make them tired during the day are most at risk for hypersomnia. These conditions include sleep apnea, kidney conditions, heart conditions, brain conditions, atypical depression, and low thyroid function.

The American Sleep Association states that the condition affects men more than women. People who smoke or drink regularly are also at risk of developing hypersomnia. Medications that cause drowsiness can have side effects similar to hypersomnia.

TREATMENTS

Treatments for this condition can vary, depending on the cause of your hypersomnia. Many drugs intended for narcolepsy can treat hypersomnia. These include amphetamine, methylphenidate, and modafinil. These drugs are stimulants that help you feel more awake. Lifestyle changes are a critical part of the treatment process. A doctor may recommend getting on a regular sleeping schedule. Avoiding certain activities can also improve symptoms, especially around bedtime. Most people with hypersomnia should not drink alcohol or use drugs. A doctor may also recommend a high-nutrition diet to maintain energy levels naturally.

Gene	Your Genotype	Your Result	Explanation
TCRA	TG	Higher Risk	Study found that when this site carries G allele, the risk of disease increases.
P2RY11	AA	Higher Risk	The specific function of this site is unclear, but the study found that when the G>A mutation occurs at this site, the risk of disease increases.

Health Risk

Hemochromatosis (HFE-related)

Hereditary hemochromatosis is a hereditary disease that causes the body to absorb excessive iron from the diet. Build-up of iron in the body, known as iron overload, can cause unpleasant symptoms. If it is not treated, this can damage parts of the body such as the liver, joints, pancreas and heart.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Some people with hereditary hemochromatosis never have symptoms. Early signs and symptoms often overlap with those of other common conditions. Common symptoms include:
· Joint pain
· Abdominal pain
· Fatigue
· Weakness

Later signs and symptoms of the disease may include:
· Diabetes
· Loss of sex drive
· Impotence
· Heart failure
· Liver failure

RISK FACTOR

Factors that increase your risk of hereditary hemochromatosis include:
· Having 2 copies of a mutated HFE gene
· Family history
· Men are more likely than women to develop signs and symptoms of hemochromatosis at an earlier age
· Women after menopause or a hysterectomy

COMPLICATIONS

Untreated, hereditary hemochromatosis can lead to a number of complications, especially in your joints and in organs where excess iron tends to be stored - your liver, pancreas and heart. Complications can include:
· Liver problems
· Pancreas problems
· Heart problems
· Reproductive problems
· Skin color changes

Gene	Your Genotype	Your Result	Explanation
HFE	CC	Normal Risk	The C>G mutation of this gene locus causes a certain disruption in the HFE protein structure, thus increases the risk of disease.
HFE	GG	Normal Risk	The G>A mutation of this gene locus causes the disulfide bond cleavage in HF protein and the destruction of protein structure, eventually increases the risk of disease.

Health Risk

Gallstones

Gallstones are common diseases caused by stones that form in the gallbladder. In most cases, they do not cause any symptoms and hence do not need to be treated. Treatment is usually only necessary if gallstones are causing symptoms and complications.

0 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

Gallstones may cause no signs or symptoms. If a gallstone lodges in a duct and causes a blockage, the resulting signs and symptoms may include:

- Sudden and rapidly intensifying pain in the upper right portion of your abdomen
- Sudden and rapidly intensifying pain in the center of your abdomen, just below your breastbone
- Back pain between your shoulder blades
- Pain in your right shoulder
- Nausea or vomiting

RISK FACTOR

Factors that may increase your risk of gallstones include:

- Being female
- Taking medications that contains estrogen, such as oral contraceptives or hormone therapy drugs
- Being age 40 or older
- Being overweight or obese
- Have recently lost weight (from either dieting or weight loss surgery)
- Being pregnant
- Eating a high-fat diet
- Eating a high-cholesterol diet
- Eating a low-fiber diet
- Having a family history of gallstones
- Having diabetes
- Having liver disease

COMPLICATIONS

Complications of gallstones may include:

- Inflammation of the gallbladder
- Blockage of the common bile duct
- Blockage of the pancreatic duct
- Gallbladder cancer

Gene	Your Genotype	Your Result	Explanation
ABCG8	GG	Normal Risk	This variant is located on the ABCG8 gene and the G>C mutation at this site may affect the composition of lipids in bile and increase cholesterol saturation in bile.

Health Risk

Primary Biliary Cirrhosis

Bile is a liquid produced inside the liver that's used to help digest fats and remove waste products from the body. It passes out of the liver through bile ducts. Primary biliary cholangitis, previously called primary biliary cirrhosis, is a chronic disease in which the bile ducts in your liver are being attacked by the immune system. It is not clear why this happens, but it is thought to be caused by a combination of subtle differences in how the immune system works.The bile ducts become damaged and injured, causing bile to build up in the liver. This further damages the liver and may lead to scarring (cirrhosis).

1 **variant(s) detected**

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

More than half the people with primary biliary cholangitis do not have any noticeable symptoms when diagnosed. The disease may be diagnosed when blood tests are done for other reasons. Symptoms eventually develop over the next 5 to 20 years. Those who do have symptoms at diagnosis typically have poorer outcomes. Common early symptoms include fatigue, itchy skin, dry eyes and mouth. Later signs and symptoms may include:

· Pain in the upper right abdomen
· Swelling of the spleen
· Bone, muscle or joint (musculoskeletal) pain
· Swollen feet and ankles (edema)
· Buildup of fluid in the abdomen due to liver failure (ascites)
· Fatty deposits (xanthomas) on the skin around the eyes, eyelids or in the creases of the palms, soles, elbows or knees
· Yellowing of the skin and eyes (jaundice)
· Darkening of the skin that is not related to sun exposure (hyperpigmentation)
· Weak and brittle bones (osteoporosis), which can lead to fractures
· High cholesterol
· Diarrhea, which may include greasy stools (steatorrhea)
· Underactive thyroid (hypothyroidism)
· Weight loss

RISK FACTOR

The following factors may increase your risk of primary biliary cholangitis

· Gender: most people with primary biliary cholangitis are women
· Age: most likely to occur in people 30 to 60 years old
· Genetic factors

COMPLICATIONS

As liver damage worsens, primary biliary cholangitis can cause serious health problems, including liver scarring (cirrhosis), increased pressure in the portal vein (portal hypertension), enlarged spleen (splenomegaly), gallstones and bile duct stones, enlarged veins (varices), liver cancer, weak bones (osteoporosis), vitamin deficiencies and decreased mental function (hepatic encephalopathy).

Gene	Your Genotype	Your Result	Explanation
SPIB	AG	Normal Risk	The AA genotype in this variant may affect the normal function of T cells and B cells, causing the occurrence of primary biliary cirrhosis.
IL12A-AS	TT	Higher Risk	The TT genotype in this variant may affect the ability of IL12 to mediate cellular immunity properly and increase the possibility of primary biliary cirrhosis.

Health Risk

Celiac Disease

Celiac disease is an autoimmune disease caused by gluten contained in certain grains, such as wheat, barley and rye. Ingestion of gluten activates the immune system and attacks the small intestine. Over time, this reaction damages your small intestine's lining and prevents it from absorbing some nutrients (malabsorption). The intestinal damage often causes diarrhea, fatigue, weight loss, bloating and anemia, and can lead to serious complications. It is estimated that 1% of individuals worldwide is being affected.

0 variant(s) detected

Consult with a healthcare professional before making any major lifestyle changes.

SIGNS AND SYMPTOMS

The signs and symptoms of celiac disease can vary greatly and differ in children and adults. Common signs and symptoms include:

- Diarrhea
- Fatigue
- Weight loss
- Bloating and gas
- Abdominal pain
- Nausea and vomiting
- Constipation
- Pale, foul-smelling stools
- Anemia

Children with celiac disease may not grow at the expected rate and may have delayed puberty.

RISK FACTOR

Celiac disease tends to be more common in people who have:

- A family member with celiac disease or dermatitis herpetiformis
- Type 1 diabetes
- Down syndrome or Turner syndrome
- Autoimmune thyroid disease
- Microscopic colitis (lymphocytic or collagenous colitis)
- Addison's disease

COMPLICATIONS

Untreated, celiac disease can cause:

- Malnutrition
- Bone weakening
- Infertility and miscarriage
- Lactose intolerance
- Cancer
- Nervous system problems

Gene	Your Genotype	Your Result	Explanation
HLA-DQA1	CC	Normal Risk	The C>T mutation occuring in this variant may lead to the type classification of HLA-DQ2.5. This is the most common HLA-DQ typing found in celiac patients in the current study of certain populations.

CARRIER STATUS

Carrier Status

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is a condition characterized by the disruption of movement leading to progressive loss of sensation, muscle control and muscle stiffness. It is a rare genetic disorder occuring in individuals having two variants in the SACS gene.

You do not have the SACS variant we tested.

Your risk for ARSACS also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the SACS gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the SACS gene cause ARSACS. The SACS gene provides instructions for producing a protein called sacsin. Sacsin is found in the brain, skin cells, muscles used for movement (skeletal muscles), and at low levels in the pancreas, but the specific function of the protein is unknown. It is unclear how the abnormal sacsin protein affects the brain and skeletal muscles and results in the signs and symptoms of ARSACS.

SIGNS AND SYMPTOMS

ARSACS is usually diagnosed in early childhood, approximately 12–24 months of age when a child begins to take their first steps. At this time, it manifests as a lack of coordination and balance resulting in frequent falls. Some of the signs and symptoms include: stiffness of the legs, appendicular and trunk ataxia, hollow foot and hand deformities, ataxic dysarthria, distal muscle wasting and horizontal gaze nystagmus.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The A deletion variant in the SACS gene

Carrier Status

Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)

Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN) is a condition characterized by an absence of the tissue connecting between the left and right sides of the brain. It causes abnormal development, weakness and loss of sensation. It is a rare genetic disorder occurring in individuals having two variants in the SLC12A6 gene.

You do not have the SLC12A6 variant we tested.

Your risk for ACCPN also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the SLC12A6 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the SLC12A6 gene cause ACCPN. The SLC12A6 gene provides instructions for making a protein called a K-Cl cotransporter. This protein is involved in moving charged atoms (ions) of potassium (K) and chlorine (Cl) across the cell membrane. Mutations in the SLC12A6 gene disrupt the function of the K-Cl cotransporter protein. The lack of functional protein normally produced from the SLC12A6 gene is believed to interfere with the development of the corpus callosum and maintenance of the nerves that transmit signals needed for movement and sensation.

SIGNS AND SYMPTOMS

Symptoms begin in infancy and include: hypotonia, areflexia, amyotrophy, variable degrees of dysgenesis of the corpus callosum, mild to severe intellectual and developmental delay and psychiatric problems including paranoid delusions, depression, hallucinations and autistic-like behavior.

TREATMENT

There is currently no cure, but some symptoms may be treated with medications such as neuroleptics for psychiatric problems.

PROGNOSIS

The prognosis is poor. Patients are usually wheelchair bound by their 20s and die by their 30s.

INHERITANCE PATTERN

The inheritance pattern is autosomal recessive. Several genes have been associated with the disorder, including SLC12A6.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The C deletion variant in the SLC12A6 gene produces a shortened and nonfunctional protein

Carrier Status

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

Autosomal Recessive Polycystic Kidney Disease (ARPKD) is a condition characterized by a buildup of clusters of cysts in kidney and liver leading to kidney, liver and lung problems and complications including urinary tract infections and high blood pressure. It is a rare genetic disorder that occurs in individuals having two variants in the PKHD1 gene.

You do not have the PKHD1 variant we tested.

Your risk for ARPKD also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the PKHD1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

PKHD1 is a large gene and many different mutations to this gene cause ARPKD. The PKHD1 gene contains instructions for creating a protein known as fibrocystin. If patients have two mutations that result in no protein being generated, the result is usually lethal. However, majority of patients have at least one copy of the gene generates some functional protein. These cases are usually viable. The exact role and function of this protein in the body is unknown.

SIGNS AND SYMPTOMS

Symptoms and signs include abdominal discomfort, polyuria, polydipsia, incidental discovery of hypertension and abdominal mass. The classic presentation for ARPKD is systemic hypertension with progression to end-stage renal disease (ESRD) by the age of 15. in a typical presentation, a small number of ARPKD sufferers live to adulthood with some kidney function; but with significant deterioration in liver function. This outcome is postulated to result from expression of the polycystic kidney and hepatic disease gene PKHD1, which is located on chromosome 6p.

DIAGNOSIS

Ultrasonography is the primary method to evaluate autosomal recessive polycystic kidney disease, particularly in the perinatal and neonatal stages.

TREATMENT

The treatment options for autosomal recessive polycystic kidney disease, given there is no current cure, are for symptom management, such as medications for hypertension, medications and/or surgery for pain, antibiotics for infection, dialysis (if renal failure) and kidney transplantation (in serious cases).

INHERITANCE PATTERN

ARPKD is inherited in an autosomal recessive pattern, which means both copies of the PKHD1 gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the PKHD1 gene produces a shortened protein
2	II	Non-carrier	The T deletion variant in the PKHD1 gene

Carrier Status

Beta Thalassemia and Related Hemoglobinopathies

Beta thalassemia is a condition characterized by a reduced hemoglobin production that causes anemia, tiredness with some serious complications including bone abnormalities and organ damage. It is a genetic disorder that occurs in individuals having two variants in the HBB gene.

You do not have the HBB variant we tested.

Your risk for beta thalassemia and related hemoglobinopathies also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the HBB gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Thalassemia is caused by mutations in the DNA of cells that make hemoglobin — the substance in red blood cells that carries oxygen throughout your body. The mutations associated with thalassemia are passed from parents to children.

Thalassemia disrupts the normal production of hemoglobin and healthy red blood cells. This causes anemia. With anemia, your blood does not have enough red blood cells to carry oxygen to tissues — leaving patients fatigued.

SIGNS AND SYMPTOMS OF THALASSEMIA

Signs and symptoms of thalassemia may include fatigue, weakness, pale or yellowish skin, facial bone deformities, slow growth, abdominal swelling and dark urine.

COMPLICATIONS

Possible complications of thalassemia include:

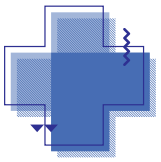
- **Iron overload.** People with thalassemia can get too much iron in their bodies, either from the disease or from frequent blood transfusions. Too much iron can result in damage to your heart, liver and endocrine system. This system includes hormone-producing glands that regulate processes throughout the body.
- **Infection.** People with thalassemia have an increased risk of infection. This is especially true if patients had their spleen removed.

In cases of severe thalassemia, the following complications can occur:

- **Bone deformities.** Thalassemia can make the bone marrow expand, which causes bones to widen. This can result in abnormal bone structure, especially in your face and skull. Bone marrow expansion also makes bones thin and brittle, increasing the chance of broken bones.
- **Enlarged spleen (splenomegaly).** The spleen helps the body fight infection and filter unwanted material, such as old or damaged blood cells. Thalassemia is often accompanied by the destruction of a large number of red blood cells. This causes the spleen to enlarge and work harder than normal. Splenomegaly can make anemia worse, and it can reduce the life of transfused red blood cells. If spleen are growing too big, doctor may suggest surgery to remove it (splenectomy).
- **Slowed growth rates.** Anemia can cause a child's growth to slow. And thalassemia may cause a delay in puberty.
- **Heart problems.** Heart problems - such as congestive heart failure and abnormal heart rhythms (arrhythmias) - may be associated with severe thalassemia.

INHERITANCE PATTERN

Thalassemia major and thalassemia intermedia are inherited in an autosomal recessive pattern. This means both copies of the HBB gene have mutations. Sometimes, people with only one HBB gene mutation (carrier) develop mild anemia. These mildly affected people are said to have thalassemia minor.



Carrier Status

Beta Thalassemia and Related Hemoglobinopathies

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to C substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
2	CC	Non-carrier	The C to G substitution variant in the HBB gene produce a deformed beta-globin protein
3	CC	Non-carrier	The C to G substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
4	AA	Non-carrier	The A to G substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
5	CC	Non-carrier	The C to T substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
6	GG	Non-carrier	The G to A substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
7	GG	Non-carrier	The G to C substitution variant in the HBB gene produces a reduced amount of the beta-globin protein important for red blood cell production
8	CC	Non-carrier	The C to T substitution variant in the HBB gene produces a shortened and nonfunctional protein
9	GG	Non-carrier	The G to A substitution variant in the HBB gene produces a shortened and nonfunctional protein
10	CC	Non-carrier	The C to T substitution variant in the HBB gene produces an abnormal hemoglobin protein that cannot bind to oxygen

Carrier Status

Bloom Syndrome

Bloom syndrome is a condition characterized by the sensitive skin after sun exposure leading to impaired growth and increased risk of cancer and infections. It is a rare genetic disorder that occurs in individuals having two variants in the BLM gene.

You do not have the BLM variant we tested.

Your risk for Bloom syndrome also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the BLM gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the BLM gene cause Bloom syndrome. BLM gene mutations result in the absence of functional BLM protein. Without the BLM protein, the cell is less able to repair DNA damage caused by ultraviolet light, which results in increased sun sensitivity. Genetic changes that allow cells to divide in an uncontrolled way lead to the cancers that occur in people with Bloom syndrome.

SIGNS AND SYMPTOMS

The most prominent feature of Bloom syndrome is proportional small size. The small size is apparent in utero. At birth, neonates exhibit rostral to caudal lengths, head circumferences, and birth weights that are typically below the third percentile. The second most commonly noted feature is a rash on the face that develops early in life as a result of sun exposure.

Some affected individuals may also have learning disabilities; an increased risk of diabetes; chronic obstructive pulmonary disease (COPD); and recurrent infections of the upper respiratory tract, ears, and lungs during infancy. Cancers may include any of those found in the general population, but develop much earlier in life in affected individuals.

TREATMENT

Bloom syndrome has no specific treatment; however, avoiding sun exposure and using sunscreens can help prevent some of the cutaneous changes associated with photo-sensitivity. Efforts to minimize exposure to other known environmental mutagens are also advisable.

INHERITANCE PATTERN

Bloom syndrome is an autosomal recessive disorder, caused by mutations in the maternally- and paternally-derived copies of the gene BLM.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The ATCTGA deletion and TAGATTC insertion variant in the BLM gene

Carrier Status

Canavan Disease

Canavan disease is a condition characterized by a progressive damage of nerve cells in the brain that disrupts brain to send and receive messages. It is a rare genetic disorder that occurs in individuals having two variants in the ASPA gene.

You do not have the ASPA variant we tested.

Your risk for Canavan disease also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the ASPA gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the ASPA gene cause Canavan disease. The ASPA gene provides instructions for making an enzyme called aspartoacylase that breaks down a compound called N-acetylaspartic acid (NAA), which is predominantly found in neurons in the brain. ASPA gene causes accumulation of NAA in brain. An excess of NAA in the brain is associated with the signs and symptoms of Canavan disease.

SIGNS AND SYMPTOMS

Symptoms of the most common (and most serious) form of Canavan disease typically appear in early infancy usually between the first three to six months of age. Canavan disease then progresses rapidly from that stage, with typical cases involving intellectual disability, loss of previously acquired motor skills, feeding difficulties, abnormal muscle tone (i.e., floppiness or stiffness; hypotonia), poor head control, and megalocephaly (abnormally enlarged head). Paralysis, blindness, or seizures may also occur.

DIAGNOSIS

The diagnosis of neonatal/ infantile Canavan disease relies on demonstration of very high concentration of N-acetylaspartic acid (NAA) in the urine. In mild/ juvenile Canavan disease, NAA may only be slightly elevated; thus, the diagnosis relies on molecular genetic testing of ASPA, the gene encoding the enzyme aspartoacylase, which functions to prevent the normal breakdown of NAA.

TREATMENT

No cure for Canavan disease is known, nor is there a standard course of treatment. Treatment is symptomatic and supportive. Physical therapy may help improve motor skills, and educational programs may help improve communication skills. Seizures are treated with antiepileptic drugs and gastrostomy is used to help maintain adequate food intake and hydration when swallowing difficulties exist.

INHERITANCE PATTERN

Canavan disease is an autosomal recessive disorder, caused by mutations in the maternally- and paternally-derived copies of the gene ASPA. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C/T to A substitution variant in the ASPA gene
2	AA	Non-carrier	The A to C substitution variant in the ASPA gene
3	CC	Non-carrier	The C to A substitution variant in the ASPA gene

Carrier Status

Congenital Disorder of Glycosylation Type 1a (PMM2-CDG)

Congenital Disorder of Glycosylation Type 1a (PMM2-CDG) is a condition that affects many other parts of the body leading to developmental delay, muscle weakness and poor weight gain. It is a rare genetic disorder that occurs in individuals having two variants in the PMM2 gene.

You do not have the PMM2 variant we tested.

Your risk for Congenital Disorder of Glycosylation Type 1a (PMM2-CDG) also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the PMM2 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

PMM2-CDG is caused by mutations in the PMM2 gene. This gene provides instructions for making an enzyme called phosphomannomutase 2 (PMM2). The PMM2 enzyme is involved in a process called glycosylation, which attaches groups of sugar molecules (oligosaccharides) to proteins. Glycosylation modifies proteins so they can perform a wider variety of functions. Mutations in the PMM2 gene lead to the production of an abnormal PMM2 enzyme with reduced activity. This leads to production of abnormally glycosylated protein in many organs and tissues, leading to a wide variety of signs and symptoms in PMM2-CDG.

SIGNS AND SYMPTOMS

Individuals with PMM2-CDG typically develop signs and symptoms of the condition during infancy. Affected infants may have weak muscle tone (hypotonia), retracted (inverted) nipples, an abnormal distribution of fat, eyes that do not look in the same direction (strabismus), developmental delay, and a failure to gain weight and grow at the expected rate (failure to thrive). Infants with PMM2-CDG also frequently have an underdeveloped cerebellum, which is the part of the brain that coordinates movement.

Distinctive facial features are sometimes present in affected individuals, including a high forehead, a triangular face, large ears, and a thin upper lip. Children with PMM2-CDG may also have elevated liver function test results, seizures, fluid around the heart (pericardialeffusion), and blood clotting disorders. About 20 percent of affected infants do not survive the first year of life due to multiple organ failure.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the PMM2 gene produces nonfunctional enzyme
2	CC	Non-carrier	The C to A substitution variant in the PMM2 gene produces less functional enzyme

Carrier Status

Cystic Fibrosis

Cystic fibrosis is a condition characterized by a buildup of thick mucus leading to respiratory and digestive problems. It is a rare genetic disorder that occurs in individuals having two variants in the CFTR gene.

You do not have the CFTR variant we tested.

Your risk for Cystic Fibrosis also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the CFTR gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

In cystic fibrosis, a defect (mutation) in a gene changes a protein that regulates the movement of salt in and out of cells. The result is thick, sticky mucus in the respiratory, digestive and reproductive systems, as well as increased salt in sweat.

Many different defects can occur in the gene. The type of gene mutation is associated with the severity of the condition. Children need to inherit one copy of the gene from each parent in order to have the disease. If children inherit only one copy, they will not develop cystic fibrosis. However, they will be carriers and possibly pass the gene to their own children.

SIGNS AND SYMPTOMS

Cystic fibrosis signs and symptoms vary, depending on the severity of the disease. Even in the same person, symptoms may worsen or improve as time passes. Some people may not experience symptoms until adolescence or adulthood.

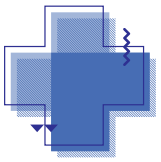
The thick and sticky mucus associated with cystic fibrosis clogs the tubes that carry air in and out of your lungs. There are some respiratory signs and symptoms: a persistent cough that produces thick mucus (sputum), wheezing, breathlessness, exercise intolerance, repeated lung infections and inflamed nasal passages or a stuffy nose.

The thick mucus can also block tubes that carry digestive enzymes from your pancreas to your small intestine. Without these digestive enzymes, your intestines are not able to completely absorb the nutrients in the food you eat. The result is often: foul-smelling, greasy stools, poor weight gain and growth, intestinal blockage, particularly in newborns (meconium ileus) and severe constipation.

COMPLICATIONS

Respiratory system complications: damaged airways, chronic infections, growths in the nose, coughing up blood, pneumothorax, respiratory failure, and acute exacerbations.

Digestive system complications: nutritional deficiencies, diabetes, blocked bile duct, intestinal obstruction and distal intestinal obstruction syndrome (DIOS).



Carrier Status

Cystic Fibrosis

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The CTT deletion variant in the CFTR gene
2	GG	Non-carrier	The G to A substitution variant in the CFTR gene
3	CC	Non-carrier	The C to T substitution variant in the CFTR gene
4	GG	Non-carrier	The G to A/C substitution variant in the CFTR gene
5	CC	Non-carrier	The C to A substitution variant in the CFTR gene
6	GG	Non-carrier	The G to T substitution variant in the CFTR gene
7	GG	Non-carrier	The G to T substitution variant in the CFTR gene
8	GG	Non-carrier	The G to A substitution variant in the CFTR gene
9	GG	Non-carrier	The G to A substitution variant in the CFTR gene
10	CC	Non-carrier	The C to T substitution variant in the CFTR gene
11	GG	Non-carrier	The G to C substitution variant in the CFTR gene
12	CC	Non-carrier	The C to T substitution variant in the CFTR gene
13	GG	Non-carrier	The G to A substitution variant in the CFTR gene

Marker	Your Genotype	Your Result	Explanation
14	CC	Non-carrier	The C to G substitution variant in the CFTR gene
15	II	Non-carrier	The TT deletion variant in the CFTR gene
16	GG	Non-carrier	The G to T substitution variant in the CFTR gene
17	GG	Non-carrier	The G to T substitution variant in the CFTR gene
18	II	Non-carrier	The T deletion variant in the CFTR gene
19	GG	Non-carrier	The G to A substitution variant in the CFTR gene
20	GG	Non-carrier	The G to A substitution variant in the CFTR gene
21	GG	Non-carrier	The G to A substitution variant in the CFTR gene
22	II	Non-carrier	The C deletion variant in the CFTR gene
23	DD	Non-carrier	The T insertion variant in the CFTR gene
24	CC	Non-carrier	The C to T substitution variant in the CFTR gene
25	GG	Non-carrier	The G to A substitution variant in the CFTR gene

Carrier Status

D-Bifunctional Protein Deficiency (DBPD)

D-Bifunctional Protein Deficiency (DBPD) is a condition characterized by the deterioration of nervous system functions leading to abnormal muscle tone, developmental disability, seizures and early death. It is a rare genetic disorder that occurs in individuals having two variants in the HSD17B4 gene.

You do not have the HSD17B4 variant we tested.

Your risk for DBPD also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the HSD17B4 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

HSD17B4 gene mutations that cause D-bifunctional protein deficiency can affect one or both of the protein's functions; however, this distinction does not seem to affect the severity or features of the disorder. Impairment of one or both of the protein's enzymatic activities prevents the D-bifunctional protein from breaking down fatty acids efficiently. As a result, these fatty acids accumulate in the body. It is unclear how fatty acid accumulation leads to the specific neurological and non-neurological features of D-bifunctional protein deficiency.

SIGNS AND SYMPTOMS

Individuals with D-bifunctional protein deficiency may have unusual facial features, including a high forehead, widely spaced eyes (hypertelorism), a lengthened area between the nose and mouth (philtrum), and a high arch of the hard palate at the roof of the mouth. Affected infants may also have an unusually large space between the bones of the skull (fontanelle). An enlarged liver (hepatomegaly) occurs in about half of affected individuals.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The A to T substitution variant in the HSD17B4 gene produces unstable and misfolded protein

Carrier Status

Dihydrolipoamide Dehydrogenase Deficiency (DLD deficiency)

Dihydrolipoamide Dehydrogenase Deficiency (DLD deficiency) is characterized by a buildup of lactic acid in tissues leading to decreased muscle tone and episodes of brain injury that can cause life-threatening liver disease. It is a rare genetic disorder that occurs in individuals having two variants in the DLD gene.

You do not have the DLD variant we tested.

Your risk for DLD deficiency also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the DLD gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the DLD gene impair the function of the DLD enzyme, which prevents the three enzyme complexes, which are responsible for energy productions in cells, from functioning properly. As a result, molecules that are normally broken down and their byproducts build up in the body, damaging tissues and leading to lactic acidosis and other chemical imbalances. In addition, the production of cellular energy is diminished. The brain is especially affected by the buildup of molecules and the lack of cellular energy, resulting in the neurological problems associated with dihydrolipoamide dehydrogenase deficiency. Liver problems are likely also related to decreased energy production in cells. The degree of impairment of each complex contributes to the variability in the features of this condition.

SIGNS AND SYMPTOMS

The signs and symptoms of dihydrolipoamide dehydrogenase deficiency occur in episodes that may be triggered by fever, injury, or other stresses on the body. Affected individuals are usually symptom-free between episodes. Many infants with this condition do not survive the first few years of life because of the severity of these episodes. Affected individuals who survive past early childhood often have delayed growth and neurological problems, including intellectual disability, muscle stiffness (spasticity), difficulty coordinating movements (ataxia), and seizures.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to T substitution variant in the DLD gene produces a less active enzyme

Carrier Status

Familial Dysautonomia

Familial dysautonomia is a condition characterized by the dysfunction of certain nerve cells in different parts of nervous system, such as in movement, senses and involuntary (autonomic) actions including breathing and digestion. It is a rare genetic disorder that occurs in individuals having two variants in the IKBKAP gene.

You do not have the IKBKAP variant we tested.

Your risk for familial dysautonomia also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the IKBKAP gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the ELP1 gene cause familial dysautonomia. The ELP1 gene provides instructions for making a protein that is found in a variety of cells throughout the body, including brain cells.

Nearly all individuals with familial dysautonomia have two copies of the same ELP1 gene mutation in each cell. This mutation can disrupt how information in the ELP1 gene is pieced together to make a blueprint for the production of ELP1 protein. As a result of this error, a reduced amount of normal ELP1 protein is produced. This mutation behaves inconsistently, however. Some cells produce near normal amounts of the protein, and other cells - particularly brain cells - have very little of the protein. Critical activities in brain cells are probably disrupted by reduced amounts or the absence of ELP1 protein, leading to the signs and symptoms of familial dysautonomia.

SIGNS AND SYMPTOMS

Problems related to this disorder first appear during infancy. Early signs and symptoms include poor muscle tone (hypotonia), feeding difficulties, poor growth, lack of tears, frequent lung infections, and difficulty maintaining body temperature. Older infants and young children with familial dysautonomia may hold their breath for prolonged periods of time, which may cause a bluish appearance of the skin or lips (cyanosis) or fainting. This breath-holding behavior usually stops by age 6. Developmental milestones, such as walking and speech, are usually delayed, although some affected individuals show no signs of developmental delay.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The A to G substitution variant in the ELP1 gene produces deformed enzyme

Carrier Status

Familial Hyperinsulinism (ABCC8-Related)

Familial Hyperinsulinism (ABCC8-related) is a condition characterized by the elevated insulin level leading to episodes of low blood sugar that causes low energy, seizures and if left untreated, causes serious complication like brain damage. It is a rare genetic disorder that occurs in individuals having two variants in the ABCC8 gene.

You do not have the ABCC8 variant we tested.
Your risk for familial hyperinsulinism (ABCC8-related) also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the ABCC8 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Congenital hyperinsulinism is caused by mutations in genes that regulate the release (secretion) of insulin, which is produced by beta cells in the pancreas. Insulin clears excess sugar (in the form of glucose) from the bloodstream by passing glucose into cells to be used as energy.

Mutations in at least nine genes have been found to cause congenital hyperinsulinism. Mutations in the ABCC8 gene are the most common known cause of the disorder. They account for this condition in approximately 40 percent of affected individuals.

SIGNS AND SYMPTOMS

People with this condition have frequent episodes of low blood sugar (hypoglycemia). In infants and young children, these episodes are characterized by a lack of energy (lethargy), irritability, or difficulty feeding. Repeated episodes of low blood sugar increase the risk for serious complications such as breathing difficulties, seizures, intellectual disability, vision loss, brain damage, and coma.

INHERITANCE PATTERN

Congenital hyperinsulinism can have different inheritance patterns, usually depending on the form of the condition. The most common form is the diffuse form, which occurs when all of the beta cells in the pancreas secrete too much insulin. The focal form of congenital hyperinsulinism occurs when only some of the beta cells over-secrete insulin. The diffuse form of congenital hyperinsulinism is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The GAA deletion variant in the ABCC8 gene produces a protein with impaired function
2	CC	Non-carrier	The G to A substitution variant in the ABCC8 gene produces a protein with impaired function
3	AA	Non-carrier	The T to A substitution variant in the ABCC8 gene produces a nonfunctional protein

Carrier Status

Familial Mediterranean Fever (FMF)

Familial Mediterranean fever (FMF) is a condition characterized by the frequent episodes of fever together with pain in the abdomen, chest, and joints and if left untreated, it triggers a buildup of protein especially in kidney that might lead to kidney failure. It is a genetic disorder that occurs in individuals having two variants in the MEFV gene.

You do not have the MEFV variant we tested.

Your risk for FMF also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the MEFV gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the MEFV gene cause familial Mediterranean fever. The MEFV gene provides instructions for making a protein called pyrin, which is found in white blood cells. Mutations in the MEFV gene reduce the activity of the pyrin protein, which disrupts control of the inflammation process. An inappropriate or prolonged inflammatory response can result, leading to fever and pain in the abdomen, chest, or joints.

SIGNS AND SYMPTOMS

Signs and symptoms of familial Mediterranean fever usually begin during childhood. They occur in bouts of attacks that last for one to three days. Arthritic attacks may last for weeks or months. Signs and symptoms of familial Mediterranean fever include:

- Fever
- Abdominal pain
- Chest pain
- Achy, swollen joints
- A red rash on your legs, especially below your knees
- Muscle aches
- A swollen, tender scrotum
- Between attacks, you'll likely feel normal. Symptom-free periods may be as short as a few days or as long as several years.

DIAGNOSIS

The diagnosis is clinically made on the basis of the history of typical attacks, especially in patients from the ethnic groups in which FMF is more highly prevalent. An acute phase response is present during attacks, with high C-reactive protein levels, an elevated white blood cell count and other markers of inflammation. In patients with a long history of attacks, monitoring the kidney function is of importance in predicting chronic kidney failure.

INHERITANCE PATTERN

Familial Mediterranean fever is almost always inherited in an autosomal recessive pattern, which means both copies of the MEFV gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The A to G substitution variant in the MEFV gene
2	AA	Non-carrier	The T to C substitution variant in the MEFV gene
3	CC	Non-carrier	The G to A substitution variant in the MEFV gene

Carrier Status

Fanconi Anemia Group C

Fanconi anemia group C is a condition characterized by a reduced blood cells production due to impaired bone marrow function leading to birth defects and an increased risk of certain infections and cancer. It is a rare genetic disorder that occurs in individuals having two variants in the FANCC gene.

You do not have the FANCC variant we tested.

Your risk for Fanconi anemia group C also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the FANCC gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

80 to 90% of cases of Fanconi anemia are due to mutations in one of three genes, FANCA, FANCC, and FANCG. These genes provide instructions for producing components of the FA pathway which is important in repairing damaged DNA. Mutations in these genes will cause those components to be nonfunctional and disrupt the entire FA pathway. As a result, DNA damage is not repaired efficiently and ultimately resulting in either abnormal cell death or uncontrolled cell growth due to a lack of DNA repair processes.

SIGNS AND SYMPTOMS

More than half of people with Fanconi anemia have physical abnormalities. These abnormalities can involve irregular skin coloring such as unusually light-colored skin (hypopigmentation) or café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area.

Other possible symptoms of Fanconi anemia include malformed thumbs or forearms and other skeletal problems including short stature; malformed or absent kidneys and other defects of the urinary tract; gastrointestinal abnormalities; heart defects; eye abnormalities such as small or abnormally shaped eyes; and malformed ears and hearing loss. Affected individuals experience extreme tiredness (fatigue) due to low numbers of red blood cells (anemia), frequent infections due to low numbers of white blood cells (neutropenia), and clotting problems due to low numbers of platelets (thrombocytopenia). People with Fanconi anemia may also develop myelodysplastic syndrome, a condition in which immature blood cells fail to develop normally.

INHERITANCE PATTERN

Fanconi anemia is most often inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to A substitution variant in the FANCC gene
2	GG	Non-carrier	The G to A substitution variant in the FANCC gene produces a shortened protein that cannot enter into proper location within cells
3	II	Non-carrier	The C deletion variant in the FANCC gene produces less functional protein that is important to repair damaged DNA

Carrier Status

GRACILE Syndrome

GRACILE Syndrome is a condition characterized by a buildup of iron and a reduced production of bile (digestive fluid) in the liver, excess amino acids in the urine and lactic acid in the body that leads to impaired growth before birth, liver damage and even early death in infancy. It is a rare genetic disorder that occurs in individuals having two variants in the BCS1L gene.

You do not have the BCS1L variant we tested.

Your risk for GRACILE Syndrome also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the BCS1L gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

GRACILE syndrome is caused by a mutation in the BCS1L gene. The protein produced from this gene is found in cell structures called mitochondria, which convert the energy from food into a form that cells can use. In mitochondria, the BCS1L protein plays a role in oxidative phosphorylation, which is a multistep process through which cells derive much of their energy. The BCS1L protein is critical for the formation of a group of proteins known as complex III, which is one of several protein complexes involved in oxidative phosphorylation.

The genetic change involved in GRACILE syndrome alters the BCS1L protein, and the abnormal protein is broken down more quickly than the normal protein. What little protein remains is able to help form some complete complex III, although the amount is severely reduced, particularly in the liver and kidneys. As a result, complex III activity and oxidative phosphorylation are decreased in these organs in people with GRACILE syndrome. Without energy, these organs become damaged, leading to many of the features of GRACILE syndrome. It is not clear why a change in the BCS1L gene leads to iron accumulation in people with this condition.

SIGNS AND SYMPTOMS

In GRACILE syndrome, growth before birth is slow (intrauterine growth retardation). Affected newborns are smaller than average and have an inability to grow and gain weight at the expected rate (failure to thrive). A characteristic of GRACILE syndrome is excess iron in the liver, which likely begins before birth. Iron levels may begin to improve after birth, although they typically remain elevated. Within the first day of life, infants with GRACILE syndrome have a buildup of a chemical called lactic acid in the body (lactic acidosis). They also have kidney problems that lead to an excess of molecules called amino acids in the urine (aminoaciduria). Babies with GRACILE syndrome have cholestasis, which is a reduced ability to produce and release a digestive fluid called bile. Cholestasis leads to irreversible liver disease (cirrhosis) in the first few months of life.

INHERITANCE PATTERN

GRACILE syndrome is inherited in an autosomal recessive pattern, which means both copies of the BCS1L gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The A to G substitution variant in the BCS1L gene produces a deformed protein that can quickly break down

Carrier Status

Gaucher Disease Type 1

Gaucher disease type 1 is a condition characterized by a buildup of fatty substrates to toxic levels within cells leading to bone abnormalities and enlargement of liver and spleen. Unlike type 2 and 3, GD1 does not usually involve the brain and spinal cord (central nervous system). It is a rare genetic disorder that occurs in individuals having two variants or two copies of a variant in the GBA gene.

You do not have the GBA variant we tested.

Your risk for Gaucher disease type 1 also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the GBA gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the GBA gene cause Gaucher disease. The GBA gene provides instructions for making an enzyme called beta-glucocerebrosidase. This enzyme breaks down a fatty substance called glucocerebroside into a sugar (glucose) and a simpler fat molecule (ceramide). Mutations in the GBA gene greatly reduce or eliminate the activity of beta-glucocerebrosidase. Without enough of this enzyme, glucocerebroside and related substances can build up to toxic levels within cells. Tissues and organs are damaged by the abnormal accumulation and storage of these substances, causing the characteristic features of Gaucher disease.

SIGNS AND SYMPTOMS

The features of this condition range from mild to severe and may appear anytime from childhood to adulthood. Major signs and symptoms include enlargement of the liver and spleen (hepatosplenomegaly), a low number of red blood cells (anemia), easy bruising caused by a decrease in blood platelets (thrombocytopenia), lung disease, and bone abnormalities such as bone pain, fractures, and arthritis.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	the T to C substitution variant in the GBA gene produces a less active enzyme
2	DD	Non-carrier	The C insertion variant in the GBA gene produces a shortened and nonfunctional enzyme
3	CC	Non-carrier	The C to A substitution variant in the GBA gene produces a less active enzyme

Carrier Status

Glycogen Storage Disease Type Ia (GSDIa)

Glycogen Storage Disease Type Ia (GSDIa) is a condition characterized by a buildup of glycogen (a complex sugar in storage form) in liver and kidneys leading to low blood sugar, impaired liver and kidney function and impaired growth. It is a rare genetic disorder that occurs in individuals having two variants in the G6PC gene.

You do not have the G6PC variant we tested.

Your risk for GSDIa also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the G6PC gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in G6PC gene causes GSDIa. The proteins produced from the G6PC and SLC37A4 genes work together to break down a type of sugar molecule called glucose 6-phosphate. The breakdown of this molecule produces the simple sugar glucose, which is the primary energy source for most cells in the body.

Mutations in the G6PC and SLC37A4 genes prevent the effective breakdown of glucose 6-phosphate. Glucose 6-phosphate that is not broken down to glucose is converted to glycogen and fat so it can be stored within cells. Too much glycogen and fat stored within a cell can be toxic. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys, leading to the signs and symptoms of GSDI.

SIGNS AND SYMPTOMS

Signs and symptoms of this condition typically appear around the age of 3 or 4 months, when babies start to sleep through the night and do not eat as frequently as newborns. Affected infants may have low blood sugar (hypoglycemia), which can lead to seizures. They can also have a buildup of lactic acid in the body (lactic acidosis), high blood levels of a waste product called uric acid (hyperuricemia), and excess amounts of fats in the blood (hyperlipidemia). As they get older, children with GSDI have thin arms and legs and short stature. An enlarged liver may give the appearance of a protruding abdomen. The kidneys may also be enlarged. Affected individuals may also have diarrhea and deposits of cholesterol in the skin (xanthomas).

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to T substitution variant in the G6PC gene produces a less active enzyme

Carrier Status

Glycogen Storage Disease Type Ib (GSDIb)

Glycogen Storage Disease Type Ib (GSDIb) is a condition characterized by a buildup of glycogen (a complex sugar in storage form) in liver and kidneys and a shortage of white blood cells leading to low blood sugar, impaired liver and kidney function and frequent bacterial infections. It is a rare genetic disorder that occurs in individuals having two variants in the SLC37A4 gene.

You do not have the SLC37A4 variant we tested.
Your risk for GSDIb also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the SLC37A4 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in SLC37A4 gene causes GSDIb. The proteins produced from the G6PC and SLC37A4 genes work together to break down a type of sugar molecule called glucose 6-phosphate. The breakdown of this molecule produces the simple sugar glucose, which is the primary energy source for most cells in the body.

Mutations in the G6PC and SLC37A4 genes prevent the effective breakdown of glucose 6-phosphate. Glucose 6-phosphate that is not broken down to glucose is converted to glycogen and fat so it can be stored within cells. Too much glycogen and fat stored within a cell can be toxic. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys, leading to the signs and symptoms of GSDI.

SIGNS AND SYMPTOMS

Many people with GSDIb have a shortage of white blood cells (neutropenia), which can make them prone to recurrent bacterial infections. Neutropenia is usually apparent by age 1. Many affected individuals also have inflammation of the intestinal walls (inflammatory bowel disease). People with GSDIb may have oral problems including cavities, inflammation of the gums (gingivitis), chronic gum (periodontal) disease, abnormal tooth development, and open sores (ulcers) in the mouth. The neutropenia and oral problems are specific to people with GSDIb and are typically not seen in people with GSDIa.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The AG deletion variant in the SLC37A4 gene produces a shortened protein with impaired function
2	AA	Non-carrier	The A to G substitution variant in the SLC37A4 gene

Carrier Status

Hereditary Fructose Intolerance

Hereditary fructose intolerance is a condition characterized by an inability to digest fructose (a simple sugar from fruits) leading to low blood sugar, stomach pain and vomiting after eating fructose. It is a rare genetic disorder that occurs in individuals having two variants in the ALDOB gene.

You do not have the ALDOB variant we tested.

Your risk for hereditary fructose intolerance also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the ALDOB gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the ALDOB gene cause hereditary fructose intolerance. The ALDOB gene provides instructions for making the aldolase B enzyme. This enzyme is found primarily in the liver and is involved in the breakdown (metabolism) of fructose so this sugar can be used as energy. Aldolase B is responsible for the second step in the metabolism of fructose, which breaks down the molecule fructose-1-phosphate into other molecules called glyceraldehyde and dihydroxyacetone phosphate.

SIGNS AND SYMPTOMS

After ingesting fructose, individuals with hereditary fructose intolerance may experience nausea, bloating, abdominal pain, diarrhea, vomiting, and low blood sugar (hypoglycemia). Affected infants may fail to grow and gain weight at the expected rate (failure to thrive). Repeated ingestion of fructose-containing foods can lead to liver and kidney damage. The liver damage can result in a yellowing of the skin and whites of the eyes (jaundice), an enlarged liver (hepatomegaly), and chronic liver disease (cirrhosis). Continued exposure to fructose may result in seizures, coma, and ultimately death from liver and kidney failure. Due to the severity of symptoms experienced when fructose is ingested, most people with hereditary fructose intolerance develop a dislike for fruits, juices, and other foods containing fructose.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to T substitution variant in the ALDOB gene
2	GG	Non-carrier	The G to C substitution variant in the ALDOB gene produces a less stable and less active enzyme
3	II	Non-carrier	The TTTG deletion variant in the ALDOB gene produces a shortened protein with impaired function

Carrier Status

Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related)

Herlitz Junctional Epidermolysis Bullosa (LAMB3-related) is a condition characterized by severe fragile and blistering on the skin and mucous membranes (layers of the mouth and digestive tract) leading to early death in infancy. It is a rare genetic disorder that occurs in individuals having two variants in the LAMB3 gene.

You do not have the LAMB3 variant we tested.

Your risk for Herlitz Junctional Epidermolysis Bullosa (LAMB3-related) also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the LAMB3 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

LAMB3 gene mutations are the most common cause (about 70 percent) of all cases of junctional epidermolysis bullosa. The gene provide instructions for making one part (subunit) of a protein called laminin 332. This protein plays an important role in strengthening and stabilizing the skin by helping to attach the top layer of skin (the epidermis) to underlying layers.

SIGNS AND SYMPTOMS

People with H-JEB lack anchors to hold the layers of their skin together. They develop large, fluid-filled blisters in response to any trauma, even something as minor as increased room temperature. Skin chafes and wears away, leaving the person open to infection.

Granulation tissue, a kind of soft, pink, bumpy, moist skin, is often seen around the nose, mouth, ears, fingers, and toes, as well as in areas that receive friction, such as the buttocks and back of the head. This tissue bleeds easily and can be a site of fluid loss.

Infants and children with the disease often develop a hoarse cry, cough, and other breathing problems. They are prone to developing fevers, often lose their fingernails and toenails, and have poorly-formed tooth enamel. They may also have abnormalities in their urinary tract and bladder which can lead to urinary tract infections and kidney failure.

TREATMENT

Even with the best of care, H-JEB is ultimately fatal. There are no successful treatments other than to protect the child as much as possible from skin damage and treat symptoms as they arise. A cesarean section may be recommended to protect the child from the skin trauma of birth.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the LAMB3 gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the LAMB3 gene
2	GG	Non-carrier	The G to A substitution variant in the LAMB3 gene
3	GG	Non-carrier	The G to A substitution variant in the LAMB3 gene

Carrier Status

Leigh Syndrome, French Canadian Type (LSFC)

Leigh Syndrome, French Canadian Type (LSFC) is a condition characterized by a buildup of lactic acid leading to damaged tissue in the brain and poor in weight gain. It is a rare genetic disorder that occurs in individuals having two variants in the LRPPRC gene.

You do not have the LRPPRC variant we tested.

Your risk for LSFC also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the LRPPRC gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the LRPPRC gene cause Leigh syndrome (French Canadian Type). The LRPPRC gene contains instructions for making a protein called leucine-rich PPR motif-containing protein. This protein controls the levels of an enzyme called complex IV (COX) that is necessary for the cell to generate energy. Certain variants in LRPPRC result in a form of the protein that cannot properly regulate COX levels, causing signs and symptoms of Leigh syndrome.

SIGNS AND SYMPTOMS

The first signs of Leigh syndrome seen in infancy are usually vomiting, diarrhea, and difficulty swallowing (dysphagia), which disrupts eating. These problems often result in an inability to grow and gain weight at the expected rate (failure to thrive). Severe muscle and movement problems are common in Leigh syndrome. Affected individuals may develop weak muscle tone (hypotonia), involuntary muscle contractions (dystonia), and problems with movement and balance (ataxia). Loss of sensation and weakness in the limbs (peripheral neuropathy), common in people with Leigh syndrome, may also make movement difficult.

INHERITANCE PATTERN

Leigh syndrome can have different inheritance patterns. It is most commonly inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. This pattern of inheritance applies to most of the Leigh syndrome-associated genes contained in nuclear DNA. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the LRPPRC gene produces protein with impaired function that cannot enter into proper location within cells

Carrier Status

Limb-Girdle Muscular Dystrophy Type 2D (LGMD2D)

Limb-Girdle Muscular Dystrophy Type 2D (LGMD2D) is a condition characterized by deterioration of the skeletal muscles, especially those around the hips and shoulders. Most of the time this disease is diagnosed in childhood, when the affected individual begins to have trouble with tasks like walking, climbing the stairs, and rising from a sitting position. LGMD2D does not affect intelligence or mental function and rarely includes weakening of the heart muscle (cardiomyopathy). It is a rare genetic disorder that occurs in individuals having two variants in the SGCA gene.

You do not have the SGCA variant we tested.

Your risk for LGMD2D also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the SGCA gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

The most common SGCA gene mutation occurs in about one-third of people with limb-girdle muscular dystrophy type 2D. This mutation replaces the protein building block (amino acid) arginine with the amino acid cysteine at position 77 in the alpha-sarcoglycan protein, written as Arg77Cys or R77C. The rest of the known SGCA gene mutations are specific to individual families or certain populations. SGCA gene mutations may prevent the sarcoglycan complex from forming or from binding to and stabilizing the dystrophin complex. Problems with these complexes reduce the strength and resilience of muscle fibers and result in the signs and symptoms of limb-girdle muscular dystrophy.

SIGNS AND SYMPTOMS

The severity, age of onset, and features of limb-girdle muscle dystrophy vary among the many subtypes of this condition and may be inconsistent even within the same family. Signs and symptoms may first appear at any age and generally worsen with time, although in some cases they remain mild.

In the early stages of limb-girdle muscular dystrophy, affected individuals may have an unusual walking gait, such as waddling or walking on the balls of their feet, and may also have difficulty running. They may need to use their arms to press themselves up from a squatting position because of their weak thigh muscles. As the condition progresses, people with limb-girdle muscular dystrophy may eventually require wheelchair assistance.

INHERITANCE PATTERN

Limb-girdle muscular dystrophy can have different inheritance patterns. Most forms of this condition are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to T substitution variant in the SGCA gene produces abnormal protein that cannot enter into proper location within cells

Carrier Status

Limb-Girdle Muscular Dystrophy Type 2E (LGMD2E)

Limb-Girdle Muscular Dystrophy Type 2E (LGMD2E) is a condition characterized by deterioration of the skeletal muscles, especially those around the hips and shoulders. Most of the time this disease is diagnosed in childhood, when the affected individual begins to have trouble with tasks like walking, climbing the stairs, and rising from a sitting position. However, mild cases may not be manifest until adulthood. In addition, patients may experience a weakening of the heart muscle, so individuals with LGMD2E should be monitored for cardiac function. It is a rare genetic disorder that occurs in individuals having two variants in the SGCB gene.

You do not have the SGCB variant we tested.

Your risk for LGMD2E also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the SGCB gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

LGMD2E is caused by variants in the SGCB gene. The SGCB gene contains instructions for making one part of a group of proteins. These proteins, called the sarcoglycan protein complex, are found in muscle tissue where they help strengthen and protect muscle fibers. Certain variants in the SGCB gene prevent the protein complex from working properly, causing signs and symptoms of LGMD2E.

SIGNS AND SYMPTOMS

The severity, age of onset, and features of limb-girdle muscle dystrophy vary among the many subtypes of this condition and may be inconsistent even within the same family. Signs and symptoms may first appear at any age and generally worsen with time, although in some cases they remain mild.

In the early stages of limb-girdle muscular dystrophy, affected individuals may have an unusual walking gait, such as waddling or walking on the balls of their feet, and may also have difficulty running. They may need to use their arms to press themselves up from a squatting position because of their weak thigh muscles. As the condition progresses, people with limb-girdle muscular dystrophy may eventually require wheelchair assistance.

INHERITANCE PATTERN

Limb-girdle muscular dystrophy can have different inheritance patterns.
Most forms of this condition are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to C substitution variant in the SGCB gene produces a less functional protein

Carrier Status

Limb-Girdle Muscular Dystrophy Type 2I (LGMD2I)

Limb-Girdle Muscular Dystrophy Type 2I (LGMD2I) is a condition characterized by a group of conditions that cause weakness and wasting of the muscles in the arms and legs. The proximal muscles (those closest to the body such as the upper arms and thighs) are generally most affected by the condition. In LGMD2I, specifically, signs and symptoms often develop in late childhood (average age 11.5 years) and may include difficulty running and walking. The symptoms gradually worsen overtime and affected people generally rely on a wheelchair for mobility approximately 23-26 years after onset. It is a rare genetic disorder that occurs in individuals having two variants in the FKRP gene.

You do not have the FKRP variant we tested.

Your risk for LGMD2I also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**

in the FKRP gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

LGMD2I is caused by variants in the FKRP gene. The FKRP gene contains instructions for making a protein that is found at especially high levels in muscle tissue. Its function is not fully understood, but it is believed to be involved in stabilizing and protecting muscle fibers.

SIGNS AND SYMPTOMS

The severity, age of onset, and features of limb-girdle muscle dystrophy vary among the many subtypes of this condition and may be inconsistent even within the same family. Signs and symptoms may first appear at any age and generally worsen with time, although in some cases they remain mild.

In the early stages of limb-girdle muscular dystrophy, affected individuals may have an unusual walking gait, such as waddling or walking on the balls of their feet, and may also have difficulty running. They may need to use their arms to press themselves up from a squatting position because of their weak thigh muscles. As the condition progresses, people with limb-girdle muscular dystrophy may eventually require wheelchair assistance.

INHERITANCE PATTERN

Limb-girdle muscular dystrophy can have different inheritance patterns. Most forms of this condition are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to A substitution variant in the FKRP gene

Carrier Status

Medium-chain acyl-CoA Dehydrogenase Deficiency

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD deficiency) is a condition characterized by an inability to convert fats to energy especially during fasting or under stress leading to low blood sugar and lack of energy. It is a rare genetic disorder that occurs in individuals having two variants in the ACADM gene.

You do not have the ACADM variant we tested.

Your risk for MCAD deficiency also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the ACADM gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the ACADM gene cause MCAD deficiency. This gene provides instructions for making an enzyme called medium-chain acyl-CoA dehydrogenase, which is required to break down (metabolize) a group of fats called medium-chain fatty acids. These fatty acids are found in foods and the body's fat tissues. Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues.

SIGNS AND SYMPTOMS

Signs and symptoms of MCAD deficiency typically appear during infancy or early childhood and can include vomiting, lack of energy (lethargy), and low blood sugar (hypoglycemia). In rare cases, symptoms of this disorder are not recognized early in life, and the condition is not diagnosed until adulthood. People with MCAD deficiency are at risk of serious complications such as seizures, breathing difficulties, liver problems, brain damage, coma, and sudden death.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to C substitution variant in the ACADM gene
2	AA	Non-carrier	The A to G substitution variant in the ACADM gene
3	CC	Non-carrier	The C to T substitution variant in the ACADM gene
4	CC	Non-carrier	The C to T substitution variant in the ACADM gene

Carrier Status

Maple Syrup Urine Disease Type 1B

Maple Syrup Urine Disease Type 1B (MSUD 1B) is a condition characterized by an inability to digest certain amino acids in the body leading to poor feeding, delayed development and urine with distinctive sweet odor. It is a rare genetic disorder that occurs in individuals having two variants in the BCKDHB gene.

You do not have the BCKDHB variant we tested.
Your risk for Maple Syrup Urine Disease Type 1B also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected
in the BCKDHB gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the BCKDHA, BCKDHB, and DBT genes can cause maple syrup urine disease. These three genes provide instructions for making proteins that work together as part of a complex. The protein complex is essential for breaking down the amino acids leucine, isoleucine, and valine, which are present in many kinds of food, particularly protein-rich foods such as milk, meat, and eggs. Because high levels of these substances are toxic to the brain and other organs, their accumulation leads to the serious health problems associated with maple syrup urine disease.

SIGNS AND SYMPTOMS

Maple syrup urine disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth.

Within several days, the infant will show poor feeding, vomiting, and irritability, followed by lack of energy, weight loss, seizures, a tense arched posture, muscle tone which alternates between stiff and limp, and swelling of the brain. If untreated, life-threatening coma or respiratory failure could occur within 7 to 10 days and most will die within several months.

Upon any lapse of treatment, classic MSUD can cause brain damage. People with the disease are particularly prone to crisis during illness, infection, fasting, or after surgery. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still lead to delayed development and other health problems if not treated.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to C substitution variant in the BCKDHB gene
2	GG	Non-carrier	The G to A substitution variant in the BCKDHB gene

Carrier Status

Mucolipidosis Type IV

Mucolipidosis Type IV is a condition characterized by delayed development and gradual loss of vision in childhood. It is a rare genetic disorder that occurs in individuals having two variants in the MCOLN1 gene.

You do not have the MCOLN1 variant we tested.

Your risk for Mucolipidosis Type IV also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**

in the MCOLN1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the MCOLN1 gene cause mucolipidosis type IV. This gene provides instructions for making a protein called mucolipin-1. This protein is located in the membranes of lysosomes and endosomes, compartments within the cell that digest and recycle materials. While its function is not completely understood, mucolipin-1 plays a role in the transport (trafficking) of fats (lipids) and proteins between lysosomes and endosomes. Mucolipin-1 appears to be important for the development and maintenance of the brain and retina. In addition, this protein is likely critical for normal functioning of the cells in the stomach that produce digestive acids.

SIGNS AND SYMPTOMS

Approximately 95 percent of individuals with this condition have the severe form. People with typical mucolipidosis type IV have delayed development of mental and motor skills (psychomotor delay). Motor skills include sitting, standing, walking, grasping objects, and writing.

Psychomotor delay is moderate to severe and usually becomes apparent during the first year of life. Affected individuals have intellectual disability, limited or absent speech, difficulty chewing and swallowing, weak muscle tone (hypotonia) that gradually turns into abnormal muscle stiffness (spasticity), and problems controlling hand movements. Most people with typical mucolipidosis type IV are unable to walk independently. In about 15 percent of affected individuals, the psychomotor problems worsen over time.

Vision may be normal at birth in people with typical mucolipidosis type IV, but it becomes increasingly impaired during the first decade of life. They are likely to also have impaired production of stomach acid (achlorhydria). They may not have enough iron in their blood, which can lead to a shortage of red blood cells (anemia). People with the severe form of this disorder usually survive to adulthood; however, they may have a shortened lifespan.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The A to G substitution variant in the MCOLN1 gene

Carrier Status

Neuronal Ceroid Lipofuscinosis (CLN5-Related)

Neuronal Ceroid Lipofuscinosis (CLN5-related) is a condition that primarily affects the nervous system, leading to worsening problems with vision, movement, and thinking ability. It is a rare genetic disorder that occurs in individuals having two variants in the CLN5 gene.

You do not have the CLN5 variant we tested.

Your risk for Neuronal Ceroid Lipofuscinosis (CLN5-related) also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the CLN5 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

CLN5 disease is caused by mutations in the CLN5 gene, which provides instructions for making a protein whose function is not well understood. After the CLN5 protein is produced, it is transported to cell compartments called lysosomes, which digest and recycle different types of molecules. Research suggests that the CLN5 protein may play a role in the process by which lysosomes break down or recycle damaged or unneeded proteins within the cell.

SIGNS AND SYMPTOMS

The signs and symptoms of this condition can begin anytime between childhood and early adulthood, but they typically appear around age 5. Children with CLN5 disease often have normal development until they experience the first signs of the condition, which are usually problems with movement and a loss of previously acquired motor skills (developmental regression).

Other features of the condition include recurrent seizures that involve uncontrollable muscle jerks (myoclonic epilepsy), difficulty coordinating movements (ataxia), vision loss, and a decline in intellectual function. The life expectancy of people with CLN5 disease varies; affected individuals usually survive into adolescence or mid-adulthood.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The AT deletion variant in the CLN5 gene produces an inactive protein that cannot be transported to lysosomes

Carrier Status

Neuronal Ceroid Lipofuscinosis (PPT1-Related)

Neuronal Ceroid Lipofuscinosis (PPT1-related) is a condition that primarily affects the nervous system, causing tissue loss in the brain and an unusually small head (microcephaly). This leads to worsening problems with vision, movement, and thinking ability. It is a rare genetic disorder that occurs in individuals having two variants in the PPT1 gene.

You do not have the PPT1 variant we tested.

Your risk for Neuronal Ceroid Lipofuscinosis (PPT1-related) also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the PPT1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

PPT1 gene mutations decrease or eliminate the production of functional enzyme in lysosomes. A reduction of this enzyme impairs the removal of fatty acids from certain proteins. These partially broken down fats and proteins accumulate in lysosomes. While accumulation of these substances occurs in cells throughout the body, nerve cells appear to be particularly vulnerable to damage caused by the abnormal cell materials. Early and widespread loss of nerve cells in this disease leads to severe signs and symptoms and death in childhood.

SIGNS AND SYMPTOMS

INFANTILE FORM

The infantile form of NCL (INCL) usually begin to cause noticeable symptoms between 6 months and 24 months of age. Initially, infants may show developmental delays, jerking movements, and / or seizures. In addition, these infants will typically have small heads. Blindness and seizures generally develop by 24 months of age, after which cognitive functions will deteriorate. The child's movement typically becomes spastic and uncontrolled, and will experience a loss of motor skills and intellectual abilities.

JUVENILE FORM

The symptoms of juvenile NCL (JNCL), also called Batten disease, often begin between the ages of 4 and 10. Children with JNCL rapidly lose their vision, which is often the first noticeable symptom. They typically become completely blind within two years. Most children with JNCL develop periodic seizures between the ages of 5 and 18, with cognitive functions declining between the ages of 8 and 14. They often experience speech and behavioral problems in childhood; psychiatric problems such as disturbed thoughts, attention difficulties, aggression; and dementia or memory problems. Individuals with JNCL also show a decline in motor function and may have difficulty controlling body movements.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the PPT1 gene cannot produce a fully functional palmitoyl-protein thioesterase 1 enzyme
2	TT	Non-carrier	The T to G substitution variant in the PPT1 gene produces a deformed and less active protein
3	TT	Non-carrier	The T to A substitution variant in the PPT1 gene produces a deformed protein that cannot enter into the proper location within cells

Carrier Status

Niemann-Pick Disease Type A

Niemann-Pick Disease Type A is a condition characterized by the enlargement of liver and spleen, developmental disability, frequent lung infections and early death. It is a rare genetic disorder that occurs in individuals having two variants in the SMPD1 gene.

You do not have the SMPD1 variant we tested.

Your risk for Niemann-Pick Disease Type A also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the SMPD1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Niemann-Pick disease types A and B is caused by mutations in the SMPD1 gene. This gene provides instructions for producing an enzyme called acid sphingomyelinase. This enzyme is found in lysosomes, which are compartments within cells that break down and recycle different types of molecules. Acid sphingomyelinase is responsible for the conversion of a fat (lipid) called sphingomyelin into another type of lipid called ceramide. Mutations in SMPD1 lead to a shortage of acid sphingomyelinase, which results in reduced break down of sphingomyelin, causing this fat to accumulate in cells. This fat buildup causes cells to malfunction and eventually die. Over time, cell loss impairs function of tissues and organs including the brain, lungs, spleen, and liver.

SIGNS AND SYMPTOMS

Infants with Niemann-Pick disease type A usually develop an enlarged liver and spleen (hepatosplenomegaly) by age 3 months and fail to gain weight and grow at the expected rate (failure to thrive). The affected children develop normally until around age 1 when they experience a progressive loss of mental abilities and movement (psychomotor regression). Children with Niemann-Pick disease type A also develop widespread lung damage (interstitial lung disease) that can cause recurrent lung infections and eventually lead to respiratory failure. All affected children have an eye abnormality called a cherry-red spot, which can be identified with an eye examination. Children with Niemann-Pick disease type A generally do not survive past early childhood.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to C substitution variant in the SMPD1 gene
2	II	Non-carrier	The C deletion variant in the SMPD1 gene
3	GG	Non-carrier	The G to T substitution variant in the SMPD1 gene

Carrier Status

Nonsyndromic Hearing Loss and Deafness, DFNB1

Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related) is a condition characterized by an inability to allow communication between neighboring cells leading to mild to severe hearing loss that is present from birth. It is an inherited disorder that occurs in individuals having two variants in the GJB2 gene.

You do not have the GJB2 variant we tested.

Your risk for DFNB1 also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the GJB2 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Most cases of nonsyndromic hearing loss are inherited in an autosomal recessive pattern. About half of all severe-to-profound autosomal recessive nonsyndromic hearing loss results from mutations in the GJB2 gene; these cases are designated DFNB1. The GJB2 gene provides instructions for making a protein called connexin 26, which is a member of the connexin protein family. Mutations in the GJB2 gene alter their respective connexin proteins, which changes the structure of gap junctions and may affect the function or survival of cells that are needed for hearing.

SIGNS AND SYMPTOMS

The characteristics of nonsyndromic hearing loss vary among the different types. Hearing loss can affect one ear (unilateral) or both ears (bilateral). Degrees of hearing loss range from mild (difficulty understanding soft speech) to profound (inability to hear even very loud noises). The term "deafness" is often used to describe severe-to-profound hearing loss. Hearing loss can be stable, or it may be progressive, becoming more severe as a person gets older. Particular types of nonsyndromic hearing loss show distinctive patterns of hearing loss. For example, the loss may be more pronounced at high, middle, or low tones.

INHERITANCE PATTERN

Nonsyndromic hearing loss has different patterns of inheritance. Between 75 and 80 percent of cases are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Usually, each parent of an individual with autosomal recessive hearing loss carries one copy of the mutated gene but does not have hearing loss.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The C deletion variant in the GJB2 gene produces a shortened protein with impaired function
2	II	Non-carrier	The A deletion variant in the GJB2 gene produces a shortened protein with impaired function

Carrier Status

Pendred Syndrome and DFNB4 Hearing Loss

Pendred Syndrome and DFNB4 Hearing Loss (SLC26A4-related) are conditions characterized by an inability to maintain ions levels in the inner ear and thyroid leading to hearing impairment, structural defects in the inner ear and the enlargement of the thyroid. It is a rare genetic disorder that occurs in individuals having two variants in the SLC26A4 gene.

You do not have the SLC26A4 variant we tested.

Your risk for Pendred Syndrome and DFNB4 Hearing Loss also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the SLC26A4 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the SLC26A4 gene cause about half of all cases of Pendred syndrome. The SLC26A4 gene provides instructions for making a protein called pendrin. This protein transports negatively charged particles (ions), including chloride, iodide, and bicarbonate, into and out of cells. Although the function of pendrin is not fully understood, this protein is important for maintaining the proper levels of ions in the thyroid and the inner ear. Mutations in the SLC26A4 gene alter the structure or function of pendrin, which disrupts ion transport. An imbalance of particular ions disrupts the development and function of the thyroid gland and structures in the inner ear, which leads to the characteristic features of Pendred syndrome.

SIGNS AND SYMPTOMS

In most people with Pendred syndrome, severe to profound hearing loss caused by changes in the inner ear (sensorineural hearing loss) is evident at birth. Less commonly, hearing loss does not develop until later in infancy or early childhood. Some affected individuals also have problems with balance caused by dysfunction of the vestibular system, which is the part of the inner ear that helps maintain the body's balance and orientation.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to C substitution variant in the SLC26A4 gene produces nonfunctional protein
2	AA	Non-carrier	The A to G substitution variant in the SLC26A4 gene produces nonfunctional protein
3	AA	Non-carrier	The A to C substitution variant in the SLC26A4 gene produces nonfunctional protein
4	GG	Non-carrier	The G to T substitution variant in the SLC26A4 gene produces protein that cannot enter into proper location within cells
5	AA	Non-carrier	The A to G substitution variant in the SLC26A4 gene produces protein that cannot enter into proper location within cells
6	TT	Non-carrier	The T to G substitution variant in the SLC26A4 gene produces protein that cannot enter into proper location within cells

Carrier Status

Phenylketonuria and related disorders

Phenylketonuria and related disorders (PKU) is a condition characterized by an elevated phenylalanine (an amino acid) level leading to seizures, intellectual disability and skin disorders. It is a genetic disorder that occurs in individuals having two variants in the PAH gene.

You do not have the PAH variant we tested.

Your risk for Phenylketonuria and related disorders also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the PAH gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the PAH gene cause phenylketonuria. The PAH gene provides instructions for making an enzyme called phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine to other important compounds in the body. If gene mutations reduce the activity of phenylalanine hydroxylase, phenylalanine from the diet is not processed effectively. As a result, this amino acid can build up to toxic levels in the blood and other tissues. Because nerve cells in the brain are particularly sensitive to phenylalanine levels, excessive amounts of this substance can cause brain damage.

Classic PKU, the most severe form of the disorder, occurs when phenylalanine hydroxylase activity is severely reduced or absent. People with untreated classic PKU have levels of phenylalanine high enough to cause severe brain damage and other serious health problems. Mutations in the PAH gene that allow the enzyme to retain some activity result in milder versions of this condition, such as variant PKU or non-PKU hyperphenylalaninemia.

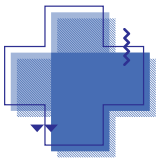
SIGNS AND SYMPTOMS

The signs and symptoms of PKU vary from mild to severe. The most severe form of this disorder is known as classic PKU. Infants with classic PKU appear normal until they are a few months old. Without treatment, these children develop permanent intellectual disability. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common. Untreated individuals may have a musty or mouse-like odor as a side effect of excess phenylalanine in the body. Children with classic PKU tend to have lighter skin and hair than unaffected family members and are also likely to have skin disorders such as eczema. Less severe forms of this condition, sometimes called variant PKU and non-PKU hyperphenylalaninemia, have a smaller risk of brain damage. People with very mild cases may not require treatment with a low-phenylalanine diet.

Babies born to mothers who have PKU and uncontrolled phenylalanine levels (women who no longer follow a low-phenylalanine diet) have a significant risk of intellectual disability because they are exposed to very high levels of phenylalanine before birth. These infants may also have a low birth weight and grow more slowly than other children. Other characteristic medical problems include heart defects or other heart problems, an abnormally small head size (microcephaly), and behavioral problems. Women with PKU and uncontrolled phenylalanine levels also have an increased risk of pregnancy loss.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.



Carrier Status

Phenylketonuria and related disorders

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to C substitution variant in the PAH gene produces a deformed enzyme that can be quickly broken down by the cell
2	AA	Non-carrier	The A to G substitution variant in the PAH gene produces an enzyme that can be quickly broken down in the cell
3	AA	Non-carrier	The A to G substitution variant in the PAH gene produces a less active enzyme
4	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a nonfunctional enzyme
5	CC	Non-carrier	The C to T substitution variant in the PAH gene produces a less stable enzyme
6	CC	Non-carrier	The C to T substitution variant in the PAH gene produces a less stable enzyme
7	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a nonfunctional enzyme
8	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a nonfunctional enzyme
9	CC	Non-carrier	The C to T substitution variant in the PAH gene produces a less active enzyme
10	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a nonfunctional enzyme
11	CC	Non-carrier	The C to A substitution variant in the PAH gene produces a shortened enzyme with impaired function
12	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a less active enzyme

Marker	Your Genotype	Your Result	Explanation
13	CC	Non-carrier	The C to A substitution variant in the PAH gene produces a less stable enzyme
14	GG	Non-carrier	The G to C substitution variant in the PAH gene produces a less active enzyme
15	TT	Non-carrier	The T to C substitution variant in the PAH gene produces a less active enzyme
16	GG	Non-carrier	The G to A substitution variant in the PAH gene produces a less stable enzyme
17	GG	Non-carrier	The G to A substitution variant in the PAH gene produces an unstable enzyme with impaired function
18	CC	Non-carrier	The C to T substitution variant in the PAH gene produces a less stable enzyme
19	TT	Non-carrier	The T to C substitution variant in the PAH gene produces a less stable enzyme
20	CC	Non-carrier	The C to T substitution variant in the PAH gene produces an unstable enzyme with impaired function
21	CC	Non-carrier	The C to T substitution variant in the PAH gene produces a nonfunctional enzyme

Carrier Status

Primary Hyperoxaluria Type 2 (PH2)

Primary Hyperoxaluria Type 2 (PH2) is a condition characterized by a buildup of stones in the kidney and urinary tract leading to kidney failure if left untreated. It is a rare genetic disorder that occurs in individuals having two variants in the GRHPR gene.

You do not have the GRHPR variant we tested.

Your risk for Primary Hyperoxaluria Type 2 also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the GRHPR gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Researchers have identified more than a dozen GRHPR mutations that cause this condition. These mutations either introduce signals that disrupt production of the glyoxylate reductase/hydroxypyruvate reductase enzyme or alter its structure. As a result, enzyme activity is absent or dramatically reduced. Glyoxylate builds up because of the enzyme shortage, and is converted to a compound called oxalate instead of glycolate. Oxalate, in turn, combines with calcium to form calcium oxalate, which the body cannot readily eliminate. Deposits of calcium oxalate can lead to the characteristic features of primary hyperoxaluria type 2.

SIGNS AND SYMPTOMS

Primary hyperoxaluria type 2 is characterized by recurrent nephrolithiasis (deposition of excess calcium oxalate in the kidney and urinary tract), nephrocalcinosis (deposition of calcium oxalate in the kidney tissue), and end-stage renal disease (ESRD). After ESRD, oxalosis (widespread tissue deposition of calcium oxalate) usually develops. Presenting symptoms are typically those associated with the presence of kidney stones, including hematuria, renal colic (a type of abdominal pain caused by kidney stones), or obstruction of the urinary tract. The symptoms of primary hyperoxaluria type 2 are typically less severe than primary hyperoxaluria type 1 and may be limited to kidney stone formation. Symptom onset may occur in childhood or adolescence. End stage renal disease is rarely observed in childhood.

TREATMENT

The current management strategy includes high fluid intake, treatment with inhibitors of calcium oxalate crystallization, and temporary intensive dialysis for end-stage renal disease (ESRD) followed by kidney transplantation. Varying success has been reported following transplantation, with recurrence being a real possibility since hyperoxaluria and elevated L-glycerate levels persist. Careful management in the postoperative period, with attention to brisk urine output and use of calcium oxalate urinary inhibitors may help prevent complications.

INHERITANCE PATTERN

Primary hyperoxaluria Type 2 is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	II	Non-carrier	The G deletion variant in the GRHPR gene

Carrier Status

Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1)

Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1) is a condition characterized by the shortening of the long bones (upper arms and thighs) leading to bone abnormalities, cataracts and intellectual disability. It is a rare genetic disorder that occurs in individuals having two variants in the PEX7 gene.

You do not have the PEX7 variant we tested.

Your risk for RCDP1 also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the PEX7 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Rhizomelic chondrodysplasia punctata results from mutations in one of three genes. Mutations in the PEX7 gene, which are most common, cause RCDP1. The genes associated with rhizomelic chondrodysplasia punctata are involved in the formation and function of structures called peroxisomes. Peroxisomes are sac-like compartments within cells that contain enzymes needed to break down many different substances, including fatty acids and certain toxic compounds. They are also important for the production of fats (lipids) used in digestion and in the nervous system.

SIGNS AND SYMPTOMS

Rhizomelic chondrodysplasia punctata is characterized by shortening of the bones in the upper arms and thighs (rhizomelia). Affected individuals also have a specific bone abnormality called chondrodysplasia punctata, which affects the growth of the long bones and can be seen on X-rays. People with rhizomelic chondrodysplasia punctata often develop joint deformities (contractures) that make the joints stiff and painful.

Distinctive facial features are also seen with rhizomelic chondrodysplasia punctata. These include a prominent forehead, widely set eyes (hypertelorism), a sunken appearance of the middle of the face (midface hypoplasia), a small nose with upturned nostrils, and full cheeks. Additionally, almost all affected individuals have clouding of the lenses of the eyes (cataracts). The cataracts are apparent at birth (congenital) or develop in early infancy.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to A substitution variant in the PEX7 gene produces protein that cannot enter into proper location within cells

Carrier Status

Salla Disease

Salla Disease is a condition characterized by an inability to store free sialic acid properly leading to weak muscle tone and coordination, poor growth, intellectual disability and seizures. It is a rare genetic disorder that occurs in individuals having two variants in the SLC17A5 gene.

You do not have the SLC17A5 variant we tested.

Your risk for Salla Disease also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**

in the SLC17A5 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the SLC17A5 gene cause all forms of sialic acid storage disease. This gene provides instructions for producing a protein called sialin that is located mainly on the membranes of lysosomes, compartments in the cell that digest and recycle materials. Sialin moves a molecule called free sialic acid, which is produced when certain proteins and fats are broken down, out of the lysosomes to other parts of the cell. Free sialic acid means that the sialic acid is not attached (bound) to other molecules. Researchers believe that sialin may also have other functions in brain cells, in addition to those associated with the lysosomes, but these additional functions are not well understood.

SIGNS AND SYMPTOMS

Babies with Salla disease usually begin exhibiting hypotonia during the first year of life and go on to experience progressive neurological problems. Signs and symptoms of Salla disease include intellectual disability and developmental delay, seizures, problems with movement and balance (ataxia), abnormal tensing of the muscles (spasticity), and involuntary slow, sinuous movements of the limbs (athetosis). Individuals with Salla disease usually survive into adulthood.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the SLC17A5 gene cannot produce a protein that can remove waste products from the cells.

Carrier Status

Sickle Cell Anemia

Sickle Cell Anemia is a condition characterized by an inability to carry oxygen with misshaped red blood cells leading to anemia, episodes of pain and frequent infections. It is a rare genetic disorder that occurs in individuals having two HbS variants in the HBB gene.

You do not have the HBB variant we tested.

Your risk for Sickle Cell Anemia also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the HBB gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the HBB gene cause sickle cell disease. Hemoglobin consists of four protein subunits, typically, two subunits called alpha-globin and two subunits called beta-globin. The HBB gene provides instructions for making beta-globin. Various versions of beta-globin result from different mutations in the HBB gene. One particular HBB gene mutation produces an abnormal version of beta-globin known as hemoglobin S (HbS). Other mutations in the HBB gene lead to additional abnormal versions of beta-globin such as hemoglobin C (HbC) and hemoglobin E (HbE). HBB gene mutations can also result in an unusually low level of beta-globin; this abnormality is called beta thalassemia.

SIGNS AND SYMPTOMS

Signs and symptoms of sickle cell disease usually begin in early childhood. Characteristic features of this disorder include a low number of red blood cells (anemia), repeated infections, and periodic episodes of pain. The severity of symptoms varies from person to person. Some people have mild symptoms, while others are frequently hospitalized for more serious complications.

The signs and symptoms of sickle cell disease are caused by the sickling of red blood cells. When red blood cells sickle, they break down prematurely, which can lead to anemia. Anemia can cause shortness of breath, fatigue, and delayed growth and development in children. The rapid breakdown of red blood cells may also cause yellowing of the eyes and skin, which are signs of jaundice. Painful episodes can occur when sickled red blood cells, which are stiff and inflexible, get stuck in small blood vessels. These episodes deprive tissues and organs of oxygen-rich blood and can lead to organ damage, especially in the lungs, kidneys, spleen, and brain.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	TT	Non-carrier	The T to A substitution variant in the HBB gene produces an abnormal hemoglobin protein that cannot bind to oxygen, which results in sickled or crescent-shaped red blood cells.

Carrier Status

Sjögren-Larsson Syndrome

Sjögren-Larsson Syndrome is a condition characterized by dry, scaly skin; neurological problems; and eye problems. It is a rare genetic disorder that occurs in individuals having two variants in the ALDH3A2 gene.

You do not have the ALDH3A2 variant we tested.

Your risk for Sjögren-Larsson Syndrome also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**

in the ALDH3A2 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

ALDH3A2 gene mutations disrupt the normal process of fatty acid oxidation. Most mutations result in the production of a FALDH enzyme that is unable to break down fatty aldehyde molecules. As a result, fats that cannot be broken down build up in cells. Within skin cells, excess fat accumulation can interfere with the formation of membranes that act as protective barriers to control water loss. As a result of the loss of these protective barriers, the skin has difficulty maintaining its water balance, resulting in dry, scaly skin.

SIGNS AND SYMPTOMS

Affected infants tend to be born prematurely. At birth the skin is red (erythema), but later in infancy the skin becomes dry, rough, and scaly with a brownish or yellowish tone. Mild to severe itchiness (pruritus) is also common. These skin abnormalities are generally dispersed over the whole body, most severely affecting the nape of the neck, the torso, and the extremities. The skin of the face is usually not affected.

People with this condition may also have neurological signs and symptoms. Most affected individuals have leukoencephalopathy, which is a change in a type of brain tissue called white matter. White matter consists of nerve fibers covered by a substance (myelin) that insulates and protects the nerves. In the brain, the consequences of excess fat accumulation are unclear, but it is likely that an abundance of fat disrupts the formation of myelin. Myelin is the covering that protects nerves and promotes the efficient transmission of nerve impulses. A lack of myelin can lead to neurological problems such as intellectual disability and walking difficulties.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to T substitution variant in the ALDH3A2 gene

Carrier Status

Tay-Sachs Disease

Tay-Sachs Disease is a condition characterized by the disruption of nerve cells in the brain and spinal cord leading to poor strength and coordination gradually, developmental disability, seizures and early death. It is a rare genetic disorder that occurs in individuals having two variants in the HEXA gene.

You do not have the HEXA variant we tested.

Your risk for Tay-Sachs Disease also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the HEXA gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the HEXA gene disrupt the activity of beta-hexosaminidase A, which prevents the enzyme from breaking down GM2 ganglioside. As a result, this substance accumulates to toxic levels, particularly in neurons in the brain and spinal cord. Progressive damage caused by the buildup of GM2 ganglioside leads to the destruction of these neurons, which causes the signs and symptoms of Tay-Sachs disease.

SIGNS AND SYMPTOMS

Infants with this disorder typically appear normal until the age of 3 to 6 months, when their development slowed and muscles used for movement weaken. Affected infants lose motor skills such as turning over, sitting, and crawling. They also develop an exaggerated startle reaction to loud noises. As the disease progresses, children with Tay-Sachs disease experience seizures, vision and hearing loss, intellectual disability, and paralysis. An eye abnormality called a cherry-red spot, which can be identified with an eye examination, is characteristic of this disorder. Children with this severe infantile form of Tay-Sachs disease usually live only into early childhood.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to T substitution variant in the HEXA gene produces a less functional enzyme
2	DD	Non-carrier	The GATA insertion variant in the HEXA gene cannot produce a fully functional hexosaminidase A enzyme
3	CC	Non-carrier	The C to G substitution variant in the HEXA gene cannot produce a fully functional hexosaminidase A enzyme
4	CC	Non-carrier	The C to T substitution variant in the HEXA gene cannot produce any hexosaminidase A enzyme

Carrier Status

Tyrosinemia Type I

Tyrosinemia Type I is a condition characterized by a buildup of tyrosine (an amino acid) leading to liver and kidney problems. It is a rare genetic disorder that occurs in individuals having two variants in the FAH gene.

You do not have the FAH variant we tested.

Your risk for Tyrosinemia Type I also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the FAH gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the FAH gene can cause tyrosinemia type I. In the liver, enzymes break down tyrosine in a five step process, resulting in molecules that are either excreted by the kidneys or used to produce energy or make other substances in the body. The FAH gene provides instructions for the fumarylacetoacetate hydrolase enzyme, which is responsible for the final step of tyrosine breakdown. The enzyme produced from the TAT gene, called tyrosine aminotransferase enzyme, is involved at the first step in the process. The HPD gene provides instructions for making the 4-hydroxyphenylpyruvate dioxygenase enzyme, which is responsible for the second step. Mutations in the FAH gene cause a decrease in the activity of one of the enzymes in the breakdown of tyrosine. As a result, tyrosine and its byproducts accumulate to toxic levels, which can cause damage and death to cells in the liver, kidneys, nervous system, and other organs.

SIGNS AND SYMPTOMS

Tyrosinemia type I, the most severe form of this disorder, is characterized by signs and symptoms that begin in the first few months of life. Affected infants fail to gain weight and grow at the expected rate (failure to thrive) due to poor food tolerance because high-protein foods lead to diarrhea and vomiting. Affected infants may also have yellowing of the skin and whites of the eyes (jaundice), a cabbage-like odor, and an increased tendency to bleed (particularly nosebleeds). Tyrosinemia type I can lead to liver and kidney failure, softening and weakening of the bones (rickets), and an increased risk of liver cancer (hepatocellular carcinoma). Some affected children have repeated neurologic crises that consist of changes in mental state, reduced sensation in the arms and legs (peripheral neuropathy), abdominal pain, and respiratory failure. These crises can last from 1 to 7 days. Untreated, children with tyrosinemia type I often do not survive past the age of 10.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the FAH gene cannot produce any fumarylacetoacetate hydrolase enzyme
2	CC	Non-carrier	The C to T substitution variant in the FAH gene
3	GG	Non-carrier	The G to A substitution variant in the FAH gene cannot produce a fully functional fumarylacetoacetate hydrolase enzyme
4	GG	Non-carrier	The G to T substitution variant in the FAH gene cannot produce a fully functional fumarylacetoacetate hydrolase enzyme

Carrier Status

Usher Syndrome Type 1F (Usher 1F)

Usher Syndrome Type 1F (Usher 1F) is a condition characterized by an inability to maintain normal retinal and hearing function leading to hearing impairment, poor balance and loss of vision gradually. It is a rare genetic disorder that occurs in individuals having two variants in the PCDH15 gene.

You do not have the PCDH15 variant we tested.

Your risk for Usher 1F also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**

in the PCDH15 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Usher syndrome can be caused by mutations in several different genes. Mutations in at least six genes can cause Usher syndrome type I. The genes associated with Usher syndrome provide instructions for making proteins involved in normal hearing, balance, and vision. In the inner ear, these proteins are involved in the development and function of specialized cells called hair cells, which help to transmit sound and signals from the inner ear to the brain. In the retina, the proteins contribute to the maintenance of light-sensing cells called rod photoreceptors (which provide vision in low light) and cone photoreceptors (which provide color vision and vision in bright light). For some of the proteins related to Usher syndrome, their exact role in hearing, balance, and vision is unknown.

SIGNS AND SYMPTOMS

Most individuals with Usher syndrome type I are born with severe to profound hearing loss. Progressive vision loss caused by retinitis pigmentosa becomes apparent in childhood. This type of Usher syndrome also causes abnormalities of the vestibular system, which is the part of the inner ear that helps maintain the body's balance and orientation in space. As a result of the vestibular abnormalities, children with the condition have trouble with balance. They begin sitting independently and walking later than usual, and they may have difficulty riding a bicycle and playing certain sports.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	GG	Non-carrier	The G to A substitution variant in the PCDH15 gene produces a shortened protein with impaired function that affects ear and eye development

Carrier Status

Usher Syndrome Type 3A (Usher 3A)

Usher Syndrome Type 3A (Usher 3A) is a condition characterized by an inability to deliver sounds and signals from the inner ear to the brain leading to loss of hearing and vision in late childhood and worsens gradually. It is a rare genetic disorder that occurs in individuals having two variants in the CLRN1 gene.

You do not have the CLRN1 variant we tested.

Your risk for Usher 3A also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0

variant(s) detected

in the CLRN1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Usher syndrome type III is most often caused by mutations in the CLRN1 gene. Most of the gene mutations responsible for Usher syndrome lead to a loss of hair cells in the inner ear and a gradual loss of rods and cones in the retina. Degeneration of these sensory cells causes the hearing loss, balance problems, and vision loss that occur with Usher syndrome.

SIGNS AND SYMPTOMS

People with Usher syndrome type III experience hearing loss and vision loss beginning somewhat later in life. Unlike the other forms of Usher syndrome, type III is usually associated with normal hearing at birth. Hearing loss typically begins during late childhood or adolescence, after the development of speech, and becomes more severe over time. By middle age, most affected individuals have profound hearing loss. Vision loss caused by retinitis pigmentosa also develops in late childhood or adolescence. Some people with Usher syndrome type III develop vestibular abnormalities that cause problems with balance.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	AA	Non-carrier	The A to C substitution variant in the CLRN1 gene produces a deformed protein that can quickly break down.

Carrier Status

Zellweger Syndrome Spectrum (PEX1-Related)

Zellweger Spectrum Syndrome (ZSS) (PEX1-related) is a condition characterized by a reduced functional peroxisomes (a functional unit in the cell to break down substances like fatty acids and certain toxic materials) leading to loss of hearing, vision and organ function, developmental disability and early death. It is a rare genetic disorder that occurs in individuals having two variants in the PEX1 gene.

You do not have the PEX1 variant we tested.

Your risk for ZSS also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 **variant(s) detected**
in the PEX1 gene

Consult with a healthcare professional before making any major lifestyle changes.

CAUSES

Mutations in the genes that cause Zellweger spectrum disorder prevent peroxisomes from forming normally. Diseases that disrupt the formation of peroxisomes, including Zellweger spectrum disorder, are called peroxisome biogenesis disorders. If the production of peroxisomes is altered, these structures cannot perform their usual functions. The signs and symptoms of Zellweger syndrome are due to the absence of functional peroxisomes within cells.

SIGNS AND SYMPTOMS

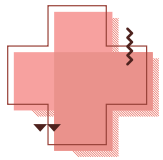
Individuals with Zellweger syndrome, at the severe end of the spectrum, develop signs and symptoms of the condition during the newborn period. These infants experience weak muscle tone (hypotonia), feeding problems, hearing and vision loss, and seizures. These problems are caused by the breakdown of myelin, which is the covering that protects nerves and promotes the efficient transmission of nerve impulses. The part of the brain and spinal cord that contains myelin is called white matter. Destruction of myelin (demyelination) leads to loss of white matter (leukodystrophy).

Children with Zellweger syndrome also develop life-threatening problems in other organs and tissues, such as the liver, heart, and kidneys. They may have skeletal abnormalities, including a large space between the bones of the skull (fontanelles) and characteristic bone spots known as chondrodysplasia punctata that can be seen on X-ray. Affected individuals have distinctive facial features, including a flattened face, broad nasal bridge, and high forehead. Children with Zellweger syndrome typically do not survive beyond the first year of life.

INHERITANCE PATTERN

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Marker	Your Genotype	Your Result	Explanation
1	CC	Non-carrier	The C to T substitution variant in the PEX1 gene produces a misfolded enzyme.



DRUG SENSITIVITY

COMMON DRUGS AND HOSPITALIZED MEDICATION

Drug Sensitivity

Clopidogrel

Clopidogrel is an antiplatelet agent that is widely used in atherothrombotic thrombosis-related diseases (coronary heart disease, cerebral thrombosis, peripheral arterial disease). CYP2C19 is the main gene affecting the efficacy of clopidogrel. According to the 2010 FDA "black-box warning", an individual who carries CYP2C19 gene variants (slow metabolizer or intermediate metabolizer) are prone to cardiovascular adverse events (such as cardiovascular death, reinfarction and others).

Your result:

Ultra Good Response

You have ultra good response to this drug.

When you use clopidogrel, the breakdown of this drug is very rapid.

Normal doses may cause suboptimal therapeutic response to this drug. However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

- Clopidogrel is a platelet P2Y12 receptor inhibitor, a thienopyridine prodrug.
- Both ticagrelor and prasugrel are platelet receptor inhibitors which are less affected by CYP2C19. Therefore, slow/intermediate CYP2C19 metabolizers can use the above two drugs when the clopidogrel effect is poor.
- Some drugs inhibit the activity of the CYP2C19 enzyme and clopidogrel is less effective when used in combination with these drugs.
- Drugs that inhibit the activity of CYP2C19 include: omeprazole, esomeprazole, voriconazole, ciprofloxacin, chloramphenicol and carbamazepine.

COMMON DRUGS

Common drugs containing clopidogrel:

- Plavix
- Clopidogrel hydrogen sulfate tablets

DRUG USAGE

Clopidogrel is commonly used for treatments as below:

- Coronary heart disease
- Cerebrovascular disease
- Peripheral artery disease

Gene Name	Variant's Name	Your Genotype	Description
CYP2C19	CYP2C19*2	GG	Rs4244285 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4244285 carries A mutations leading to splicing defects that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*3	GG	Rs4986893 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4986893 carries A mutation as a stop codon variation that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*17	GC	Rs12248560 is the only allele found that enhanced CYP2C19 enzymatic activity. Rs12778431 is highly linked to rs12248560. Rs12778431 carries G mutations that increase the rate of drug metabolism.

Drug Sensitivity

Warfarin

Warfarin is a classic oral anticoagulant agent that is the most commonly used drug for patients who requires long-term anticoagulant therapy. Children with paediatric diseases such as Kawasaki disease also require long-term oral warfarin therapy. However, the optimal dose of warfarin varies widely between patients and the therapeutic window is narrow (excessive dose leads to bleeding while insufficient dose leads to a risk of thrombosis). Genetic variant is an important factor affecting the dose. Patient's genotype is an important reference for doctors to develop a dosage regimen.

Your result:

Good Response

Based on your genotypes, you are likely predisposed to have a good response to warfarin administration.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

In 2008, the US FDA updated the warfarin drug specifications, suggesting that genetic variant testing can be used to decide the initial dose selection. There are many computational models that take into account of clinical indicators and genetic results that predict the warfarin dose required for the initial phase of administration. Gage (www.warfarindosing.org) and IWPC (International Warfarin Pharmacogenetics Consortium) are widely used models.

COMMON DRUGS

Common drug containing warfarin:

- Warfarin sodium tablets

DRUG USAGE

Warfarin is commonly used for treatments as below:

- Venous thrombosis
- Atrial fibrillation
- Heart valve replacement

Gene Name	Variant's Name	Your Genotype	Description
VKORC1	(-1639 G>A)	CC	The VKORC1 gene promoter region mutation -1639 G>A, that is, rs9923231 C mutation to T. This mutation affects the expression of the enzyme protein, resulting the warfarin efficacy is relatively increased. Therefore, the carrier's sensitivity to warfarin is increased and the dosage is required has to be reduced.
CYP2C19	CYP2C9*3	AA	Rs1057910 is a variant of A to C, which converts the lysine into leucine at position 359. This missense mutation leads to a decrease in the activity of the liver CYP2C9 enzyme and a decrease in the metabolic clearance rate of the drug. Therefore, carrier of this variant should reduce the dose.

Drug Sensitivity

Isoniazid

Isoniazid is an important first-line drug for the treatment of tuberculosis, which is mainly metabolized by N-acetyltransferase 2 (NAT2) in the body. The NAT2 gene mutation can affect the enzyme activity. Slow metabolizer accumulates toxic metabolites in the liver after using this drug, resulting in an increased risk of liver damage. Fast metabolizer may have insufficient drug efficacy, resulting in an increased risk of failure to fight tuberculosis treatment.

Your result:

Good Response

You have good response to this drug.

If you use isoniazid at a regular dose, it will not increase the risk of adverse reactions such as liver damage.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Local reports showed that patients with anti-tuberculosis drugs have a liver injury rate of 15%-30%. The occurrence of liver damage is significantly associated with the drug dosage. Patients with different type of NAT2 genetic variant may have significant difference in blood concentration even with the same dose of isoniazid. European studies have shown that blood concentration of slow metabolizers are 2-7 times higher than other metabolizers. East Asian population studies have shown that blood concentration of slow metabolizers are 2-3 times higher than those of fast metabolizers. Therefore, slow metabolizers have a higher proportion of liver damage and it is recommended to reduce the dose as appropriate (half the original dose or 2.5 mg/kg).

Fast metabolizer with lower blood drug concentration than other metabolizers is prone to have insufficient effective therapeutic dose that leads to an increased risk of anti-tuberculosis treatment failure.

COMMON DRUGS

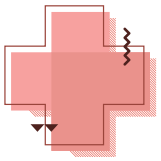
Common drugs containing isoniazid:

- Isoniazid tablets
- Isoniazid injection

DRUG USAGE

Isoniazid is commonly used for treatments as below:

- Tuberculosis



Drug Sensitivity

Isoniazid

Gene Name	Variant's Name	Your Genotype	Description
NAT2	341T>C	TT	Rs1801280 is the most important site for defining the NAT2 *5 haplotype. *5 is a common slow-metabolic variant that contains a variation of this site from T to C. The combined variants of other loci define each subtype. *5A to *5J subtypes may cause a decrease in NAT2 enzyme activity, which reduce the metabolism of isoniazid.
NAT2	590G>A	GG	Rs1799930 is the most important site for defining the NAT2 *6 haplotype. *6 is a common slow-metabolic variant that contains a variation of this site from G to A. The combined variants of other loci define each subtype. *6A to *6E subtypes may cause a decrease in NAT2 enzyme activity, which reduce the metabolism of isoniazid.
NAT2	857G>A	AG	Rs1799931 is the most important site for defining the NAT2 *7 haplotype. *7 is a common slow-metabolic variant that contains a variation of this site from G to A. The combined variants of other loci define each subtype. *7A to *7B subtypes may cause a decrease in NAT2 enzyme activity,which reduce the metabolism of isoniazid.
NAT2	803A>G	AA	Rs1208 is the most important site for defining the NAT2 *12 haplotype. *5 is a common fast-metabolic variant that contains a variation of this site from A to G. The combined variants of other loci define each subtype. *12A to *12C subtypes may cause an increase in NAT2 enzyme activity, which increase the metabolism of isoniazid.
NAT2	282C>T	TC	Rs1041983 is the most important site for defining the NAT2 *13 haplotype. *5 is a common fast-metabolic variant that contains a variation of this site from C to T. The combined variants of other loci define each subtype. The *13A subtype may cause an increase in NAT2 enzyme activity and increase the metabolism of isoniazid.

Drug Sensitivity

Omeprazole

Omeprazole is used to inhibit gastric acid secretion to treat gastroduodenal ulcer, gastroesophageal reflux disease (GERD), Zollinger-Ellison syndrome and others. CYP2C19 is the main gene affecting the efficacy of omeprazole. Ultra-rapid metabolizer may have poor drug efficacy affecting the eradication effect of Helicobacter pylori.

Your result:

Ultra Good Response

You have ultra good response to omeprazole.

When you use this drug at normal doeses, the breakdown of this drug is very rapid.

Normal doses may cause suboptimal therapeutic response to this drug. However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Omeprazole is the first proton pump inhibitor to inhibit gastric acid secretion by specifically acting on the site of the proton pump of the gastric wall.

Omeprazole is the proton pump inhibitor which is most affected by CYP2C19 enzyme activity. The effect of CYP2C19 enzyme on proton pump inhibitors is: Omeprazole > Pantoprazole > Lansoprazole > Esomeprazole > Rabeprazole. If you are an ultra-rapid metabolizer, in the event of poor efficacy when using omeprazole, you can switch to a similar drug that is less affected by CYP2C19 enzyme.

Omeprazole itself is also a CYP2C19 enzyme inhibitor. When combined with other drugs, it will affect the concentration of other drugs, such as clopidogrel, voriconazole, diazepam, phenytoin, warfarin and others. Special attention needs to be given in this case.

COMMON DRUGS

Common drugs containing omeprazole:

- Losec
- Oak
- Enteric coated omeprazole

DRUG USAGE

Omeprazole is commonly used for treatments as below:

- Gastroduodenal ulcer
- Reflux esophagitis
- Zollinger-Ellison syndrome

Gene Name	Variant's Name	Your Genotype	Description
CYP2C19	CYP2C19*2	GG	Rs4244285 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4244285 carries A mutations leading to splicing defects that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*3	GG	Rs4986893 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4986893 carries A mutation as a stop codon variation that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*17	GC	Rs12248560 is the only allele found that enhanced CYP2C19 enzymatic activity. Rs12778431 is highly linked to rs12248560. Rs12778431 carries G mutations that increase the rate of drug metabolism.

Drug Sensitivity

Simvastatin

Simvastatin belongs to the first generation of statins and statins are widely used in the prevention and treatment of hyperlipidemia, cardiovascular and cerebrovascular diseases. SLCO1B1 is the main gene affecting the concentration of simvastatin. Its genetic variant may lead to an increased risk of simvastatin-related adverse effects of muscle toxicity.

Your result:

Low Response

Based on your genotype, your likelihood of experiencing side effect from simvastatin administration is higher.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Common statins include simvastatin, pravastatin, atorvastatin, rosuvastatin, fluvastatin and others. After many people have died due to rheumatoid muscle caused by cerivastatin in 2001. The FDA quickly withdrew the drug from the market. In general, the incidence of adverse effects of statin-induced myotoxicity is about 0.1%-0.2%. Apart from the fact that cerivastatin is significantly higher than other statins, the risk of simvastatin is also quite high, about 1.1%-3.3%. It is associated with high doses.

Carrier of SLCO1B1 gene variant has impaired drug transportation leading to a reduced in drug clearance. It also causes a significant increase in blood concentration after simvastatin administration and an increased risk of myositis. Myotoxicity includes myalgia (pain without muscle dissolution - no increase in creatine kinase), myopathy (pain with muscle lysis - elevated creatine kinase) and rhabdomyolysis (severe muscle damage with acute renal failure) .

COMMON DRUGS

Common drugs containing simvastatin:

- Simvastatin tablets

DRUG USAGE

Simvastatin is commonly used for treatments as below:

- Hyperlipidemia
- Coronary heart disease
- Cerebrovascular disease

Gene Name	Variant's Name	Your Genotype	Description
SLCO1B1	c.521 T > C	TC	Rs4149056 contains a variation of this site from T to C that converts the proline to alanine at position 174, resulting in decreased transporter function. The haplotypes *5 (both rs2306283 is A) and *15 (while rs2306283 is G) can be defined. Both of which result in decreased transporter function. Studies have shown that the risk ratio (OR) for carrying a C-like myopathy is 4.5 and the OR with two C-borne myopathy is 16.9.
SLCO1B1	c.388 A > G	GG	Rs2306283 contains a variation of this site from A to G that converts the asparagine to aspartic acid at position 130. *1B (both rs4149056 is T) and *15 (while rs4149056 is C) haplotypes can be defined. *15 causes a decrease in transporter function.

Drug Sensitivity

Sulfonylureas

Sulfonylureas are a widely used oral hypoglycemic agent. Commonly used sulfonylurea drugs are gliclazide, glimepiride, glibenclamide, glipizide and others. Studies found that carrier of CYP2C9 gene variant is associated with the ability of the liver to clear sulfonylureas. Although a reduction in drug clearance results in an increased drug efficacy, it also increases the risk of adverse reactions.

Your result:

Good Response

Based on your genotype, your likelihood of experiencing side effect from sulfonylureas administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Sulfonylureas are the first oral hypoglycemic agents that are widely used and have been used for the longest time. The first-generation of sulfonylureas mainly consisted of tolbutamide while the second-generation included glibenclamide, glipizide, glimepiride, gliclazide and gliclazone. The hypoglycemic activity of the second-generation sulfonylurea was significantly stronger than that of the first-generation and the risk of adverse reactions was lower.

Common adverse reactions are gastrointestinal discomfort, skin irritation, jaundice, liver damage and weight gain. The more serious adverse reaction is persistent hypoglycemia.

COMMON DRUGS

Common drugs containing sulfonylureas:

- Glyburide
- Glimepiride
- Glyburide
- Amoli
- Gliclazide

DRUG USAGE

Sulfonylureas is commonly used for treatments as below:

- Diabetes

Gene Name	Variant's Name	Your Genotype	Description
CYP2C9	CYP2C9*3	AA	Rs1057910 contains a variation of this site from A to C that converts the lysine to leucine at position 359. This missense mutation leads to a decrease in the liver CYP2C9 enzyme activity and a decrease in the metabolic clearance rate of the drug.

Drug Sensitivity

Allopurinol

Allopurinol is the most common used drug in the treatment of hyperuricemia and gout. When people with human leukocyte antigen HLA-B*58:01 alleles use allopurinol, the risk of serious skin adverse reactions is significantly increased (OR = 73). Multi-country guidelines or health authorities recommend that Asian populations should be genetically tested before using the drug for the first time.

Your result:

Good Response

You are not the carrier of HLA-B*58:01 gene variant.

Your allopurinol metabolism is normal.

If you need to use this drug, it is recommended to use as directed.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Allopurinol is also known as sterol. The biggest safety consideration for taking allopurinol is that only 0.1%-0.4% of people are expected to have serious adverse reactions. Serious adverse reactions include hypersensitivity syndrome (fever, rash, digestive tract, respiratory tract and other systemic symptoms) and Stevens-Johnson syndrome/toxic epidermal necrolysis (SJS/TEN). The most severe TEN skin exfoliation area is >30% of body surface area, with a lethality risk as high as 25%-35%.

Who is more likely to have an adverse reaction to allopurinol? Carrier of the HLA-B*58:01 gene variant. The most common risk factors are the use of thiazide diuretics and renal insufficiency. The relationship between carrier of the HLA-B*58:01 gene and allopurinol adverse reactions was first seen in the study of Taiwanese Han population. In Asia, especially in East Asia, the proportion of this gene variant is the highest, about 5%-9%.

In 2017 version of multidisciplinary expert consensus on diagnosis and treatment of hyperuricaemia and related diseases" recommended to perform gene testing before taking allopurinol treatment. Carrier is prohibited from using this drug.

COMMON DRUGS

Common drugs containing allopurinol:

- Allopurinol tablets

DRUG USAGE

Allopurinol is commonly used for treatments as below:

- Gout
- Hyperuricemia
- Uric acid nephropathy

Gene Name	Variant's Name	Your Genotype	Description
HLA-B	HLA-B*58:01	CC	Rs9262570 is labeled as SNP of HLA-B*58:01. The positive predictive value for carrying the *58:01 allelic variation when carrying the T variant is approximately 76.4%, indicating that 76.4% of the individuals carrying the T variant carries the *58:01 variation. The sensitivity, specificity and negative predictive value of this site were both higher than 99%.

Drug Sensitivity

Citalopram

Citalopram is a selective 5-HT reuptake inhibitor (SSRIs), a first-line antidepressant that is also suitable for anxiety and obsessive-compulsive disorder. CYP2C19 is the main gene affecting the concentration of citalopram. The efficacy of citalopram is ineffective in ultra-rapid metabolizer. In addition, such drugs may have serious adverse reactions such as arrhythmia caused by prolonged QT interval. When slow CYP2C19 metabolizer use citalopram, the blood drug concentration increases leading to an increased risk of adverse reactions.

Your result:

Good Response

Based on your genotype, your likelihood of experiencing side effect from citalopram administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Selective 5-HT reuptake inhibitors (SSRIs) include citalopram, fluoxetine, fluvoxamine, paroxetine, sertraline and others. These drugs inhibits central neurons against 5-HT uptake to enhance the function of the central 5-HTergic nerve in order to produce an antidepressant effect.

The metabolism of these drugs is affected by both CYP2C19 and CYP2D6. Citalopram and escitalopram (see scientific details) are mainly affected by CYP2C19.

COMMON DRUGS

Common drugs containing citalopram:

- Citalopram hydrobromide tablets
- Escitalopram oxalate tablets

DRUG USAGE

Citalopram is commonly used for treatments as below:

- Depression
- Obsessive compulsive disorder (OCD)

Gene Name	Variant's Name	Your Genotype	Description
CYP2C19	CYP2C19*2	GG	Rs4244285 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4244285 carries A mutations leading to splicing defects that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*3	GG	Rs4986893 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4986893 carries A mutation as a stop codon variation that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*17	GC	Rs12248560 is the only allele found that enhanced CYP2C19 enzymatic activity. Rs12778431 is highly linked to rs12248560. Rs12778431 carries G mutations that increase the rate of drug metabolism.

Drug Sensitivity

Diazepam

Diazepam is a commonly used sedative, hypnotic, anti-epileptic drug that can eliminate the patient's nervousness and fear of surgery before surgery. CYP2C19 is the main gene affecting the drug efficacy. Slow CYP2C19 metabolizer may wake up slower from sedation and hypnosis.

Your result:

Ultra Good Response

You have ultra good response to this drug.

When you use diazepam, the breakdown of this drug is very rapid.

Normal doses may cause suboptimal therapeutic response to this drug. However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Diazepam is a long-acting benzodiazepine drug with a half-life of 20-50 hours. Since it was launched in 1963, it has become one of the highest-selling drugs in the United States. It can be widely used in anxiety, sleep disorders, alcohol withdrawal syndrome, tendon, epilepsy and restless legs syndrome and pre-anaesthesia administration. Through intravenous use, the effect initiates after 1-5 minutes and lasts for 1 hour while through oral administration, the effect initiates after 40 minutes.

The adverse reactions of diazepam are lethargy and memory loss. Serious adverse reactions are suicidal tendencies and respiratory depression, but they are very rare. Japanese population studies have shown that slow/intermediate CYP2C19 metabolizers have prolonged anaesthesia compared to the metabolizers.

COMMON DRUGS

Common drugs containing diazepam:

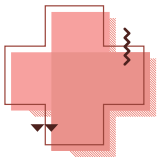
- Diazepam tablets
- Stable tablets
- Diazepam injection

DRUG USAGE

Diazepam is commonly used for treatments as below:

- Epilepsy
- Insomnia
- Anaesthesia
- Horror

Gene Name	Variant's Name	Your Genotype	Description
CYP2C19	CYP2C19*2	GG	Rs4244285 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4244285 carries A mutations leading to splicing defects that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*3	GG	Rs4986893 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4986893 carries A mutation as a stop codon variation that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*17	GC	Rs12248560 is the only allele found that enhanced CYP2C19 enzymatic activity. Rs12778431 is highly linked to rs12248560. Rs12778431 carries G mutations that increase the rate of drug metabolism.



Drug Sensitivity
Caffeine

Caffeine is a central nervous system stimulant that can be extracted from tea and coffee fruit. Common cold medicines usually contain caffeine. CYP1A2 is the main gene affecting caffeine metabolism. Slow CYP1A2 metabolizer are more prone to excessive caffeine leading to dizziness, headache, palpitation, vomiting, hallucinations and osteoporosis.

Your result:

Good Response

You have good response to caffeine.

Based on your genotype, your likelihood of experiencing side effect from caffeinated drugs administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

The CYP1A2 gene variants account 15.0% of CC type, 37.8% of AA type and 47.2% of AC type. People with the C allele have a reduced in caffeine metabolism.

Smoking, omeprazole, modafinil and others can induce CYP1A2 enzyme leading to the accelerated caffeine metabolism. The drug combination with caffeine may affect the efficacy, such as amiodarone, fluoroquinolone, fluvoxamine and ticlopidine are Inhibitors of the CYP1A2 enzyme. When used in combination, rate of caffeine metabolism is reduced leading to an increased risk of adverse reactions.

COMMON DRUGS

Common drugs containing caffeine:

- Compound acetaminophen
- Compound paracetamol
- Yellow ammonia capsule

DRUG USAGE

Caffeine is commonly used for treatments as below:

- Cold
- Bronchitis
- Asthma

Gene Name	Variant's Name	Your Genotype	Description
CYP1A2	(-163C>A)	AA	The CYP1A2 enzyme helps remove 95% of the caffeine ingested. When the C allele is carried in the rs762551 locus on the CYP1A2 gene, the activity of the individual CYP1A2 is decreased, resulting in a decrease in the rate of caffeine metabolism leading to an increased risk of adverse reactions when taking caffeinated drugs.

Drug Sensitivity

Ethanol

Ethanol (i.e. alcohol) is often used as an auxiliary and solvent for pharmaceutical preparations. Clinically drugs containing ethanol are Huoxiang Zhengqi Shui, ten drops of water, orthopaedic water, compound salicylic acid solution and others. ALDH (acetaldehyde dehydrogenase) is the main gene affecting the adverse reactions of ethanol drugs. The adverse reactions include nausea, vomiting, palpitations, decreased blood pressure, shortness of breath and even shock.

Your result:

Good Response

Your ethanol metabolism is faster.

When you use alcohol-containing drugs, your metabolic rate is fast.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Ethanol is a common auxiliary and solvent for pharmaceutical preparations. For example, Huoxiang Zhengqi water contains 45% to 55% of ethanol. Excessive intake may cause blushing and vomiting. In severe cases, it may cause blood pressure to drop, shortness of breath, confusion, shock and others.

ALDH is the main gene affecting the adverse reactions of ethanol drugs. About 40% of East Asian populations (about 500 million people) lack of ALDH2 enzyme activity or reduce enzyme activity due to the ALDH2*2 variant allele. Homozygous variation (ALDH2 *2/*2, AA type) has almost no enzymatic activity while the heterozygous variation (ALDH2 *1/*2, AG type) has reduced enzyme activity to 16% as compared to the wild type (ALDH2 *1/*1, GG type).

COMMON DRUGS

Common drugs containing ethanol:

- Orthopaedic water
- Compound salicylic acid solution

DRUG USAGE

Ethanol is commonly used for treatments as below:

- Cold
- Heat stroke

Gene Name	Variant's Name	Your Genotype	Description
ALDH2	ALDH2*2	GG	The variation of rs671 from G to A leads to the Glu504Lys mutation, i.e. replacement of glutamate with lysine that reduces the activity of ALDH enzyme. The enzyme activity was reduced by 74% when carrying one A allele. The enzyme was basically inactivated when carrying two A alleles. The decrease in enzyme activity results in a significant slowing down the rate of decomposition of acetaldehyde in the body, resulting in an increased risk of adverse reactions by taking alcohol-containing drugs.

Drug Sensitivity

Voriconazole

Voriconazole is a broad-spectrum triazole, an antifungal drug that targets progressive and potentially life-threatening fungal infections. CYP2C19 is the main gene affecting the drug metabolism. Slow metabolism of CYP2C19 enzymes (slow metabolizer or intermediate metabolizer) may increase the concentration of the drug in the blood circulation leading to an increased risk of adverse reactions (visual disorders, abnormal liver function and others).

Your result:

Good Response

Based on your genotype, your likelihood of experiencing side effect from voriconazole administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Voriconazole is a second-generation triazole antifungal drug. In recent years, global incidence of fungal infection has been severely increased. Voriconazole has gradually become the first-line antifungal drug with its good pharmacological properties and broad spectrum of antibacterial activity.

The clinical effect of voriconazole is significantly depending on concentration. When the plasma concentration is greater than a certain threshold, the incidence of adverse drug reactions such as liver damage is significantly increased.

COMMON DRUGS

Common drugs containing voriconazole:

- Voriconazole tablets
- Voriconazole injection

DRUG USAGE

Voriconazole is commonly used for treatments as below:

- Severe fungal infection
- Aspergillosis
- Candida infection

You need to pay attention to the use of the drug. Please communicate the following information to your doctor if:

- You are a intermediate CYP2C19 metabolizer.
- After you use voriconazole, your blood concentration may be higher than normal metabolizers and drug-related adverse reactions such as visual abnormalities and hepatotoxicity may increase. It is recommended to monitor the blood concentration and reduce the dose as appropriate.

Based on the above information, doctor may advise you:

- Monitor blood levels to further determine your drug response
- Reduce the amount of medication properly
- Do not make any changes and continue with the current medication plan

Drug Sensitivity

Voriconazole Tablets

Gene Name	Variant's Name	Your Genotype	Description
CYP2C19	CYP2C19*2	GG	Rs4244285 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4244285 carries A mutations leading to splicing defects that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*3	GG	Rs4986893 is one of the major alleles in the population that cause CYP2C19 enzyme deficiency. Rs4986893 carries A mutation as a stop codon variation that reduces the rate of drug metabolism.
CYP2C19	CYP2C19*17	GC	Rs12248560 is the only allele found that enhanced CYP2C19 enzymatic activity. Rs12778431 is highly linked to rs12248560. Rs12778431 carries G mutations that increase the rate of drug metabolism.

Drug Sensitivity

Tacrolimus

Tacrolimus is a macrolide antibiotic immunosuppressive agent used after organ and hematopoietic stem cell transplantation. The therapeutic index for tacrolimus is narrow and its pharmacokinetics vary among each individual. Therefore, inappropriate dose leads to toxicity risks such as nephrotoxicity, neurotoxicity, hypertension, hyperglycemia, and arterial vasoconstriction. Genetic variant is an important factor affecting the dose. Patient's genotype is an important reference for doctors to develop a dosage regimen.

Your result:

Good Response

You have good response to tacrolimus.

You are CT or TT type for CYP3A5, your body will metabolize tacrolimus at normal rate.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Compared with CT type and TT type, CC type patients who underwent transplantation and received tacrolimus treatment have decreased in their metabolism, resulting in an increased in blood concentration. Therefore, the dose needs to be lowered. In addition to genetic factors, the number of days after transplantation, age and the use of calcium channel blockers (CCB) have a significant effect on the effects of tacrolimus.

COMMON DRUGS

Common drugs containing tacrolimus:

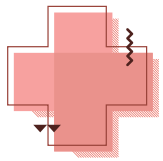
- Puleco
- Putzi

DRUG USAGE

Tacrolimus is commonly used for treatments as below:

- Organ transplantation
- Glomerulonephritis
- Graft versus host disease (GVHD)

Gene Name	Variant's Name	Your Genotype	Description
CYP3A5	CYP3A5*3	TC	Rs776746 is located in intron 3 and this mutation causes variability in CYP3A5 mRNA cleavage, resulting in a stop codon that leads to early termination of translation that produces an unstable truncated protein.



DRUG SENSITIVITY
COMMON DRUGS FOR CANCER

Drug Sensitivity

Fluorouracil

Fluorouracil is a cytotoxic antitumor drug with a wide anti-tumor spectrum and can be widely used in a variety of malignant tumors (such as digestive system malignant tumors, gynecological malignant tumors and others). DPYD, MTHFR and GSTP1 are the main related genes affecting the efficacy of fluorouracil. Almost all Asian population is non-carrier of the DPYD gene. This test only focuses on the effects of the MTHFR gene on fluorouracil.

Your result:

Normal Response

You have normal response to fluorouracil.

You have GG type of MTHFR, the use of fluorouracil is generally effective.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Local studies have found that patients with gastric cancer who receive adjuvant chemotherapy with fluorouracil regimen have a 50% of higher recurrence rate in patients with GG type of MTHFR than those with AA and AG types. Patients with AA and AG type of MTHFR have better progression-free survival than GG type.

A decrease in MTHFR enzymatic activity (AA/AG type) may result in hyperhomocysteinemia and an increased risk of cardiovascular disease. It is recommended that people with AA/AG type of MTHFR to take supplements like folic acid and B vitamins.

AA type of MTHFR accounted for 21% of the population while AG type accounted for 47% and GG type accounted for 32%.

COMMON DRUGS

Common drugs containing fluorouracil:

- Fluorouracil injection
- Compound fluorouracil oral solution

DRUG USAGE

Fluorouracil is commonly used for treatments as below:

- Malignant tumor

Gene Name	Variant's Name	Your Genotype	Description
MTHFR	677C>T	GG	The SNP locus rs1801133 on the MTHFR (methylenetetrahydrofolate reductase) gene carries a T (A) mutation and the substitution of proline with alanine (Ala222Val) results in a decrease in MTHFR enzymatic activity. The content of 5,10-methylenetetrahydrofolate increases and the combination with thymidylate synthase TS increases the cytotoxic effect of fluorouracil and enhances the chemotherapy effect. * A, T and C, G are complementary paired bases that are consistent with the Hg19 forward strand reference data where C is equivalent to G and T is equivalent to A.

Drug Sensitivity

Thiopurines

Thiopurines (including guanidine, azathioprine and thioguanine) are an alkaloid anti-metabolites commonly used in the treatment of acute leukemia, inflammatory bowel disease and other immune diseases. It can also prevent the rejection of transplanted organs. Methyltransferase (TPMT) activity in the body is inversely related to the level of active metabolites of such drugs. Carrier of TPMT gene variant possesses low enzyme activity leading to the accumulation of toxicity from active metabolites and an increased risk of myelosuppression (reduce number of white blood cells, platelets). In 2005, FDA has included the instruction of TPMT gene testing prior to starting therapy with guanidine and azathioprine.

Your result:

Good Response

Based on your genotype, your likelihood of experiencing side effect from thiopurines administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Mercaptopurine and thioguanine are mainly used to treat malignant diseases such as acute leukemia, chorionic epithelial cancer, melanoma and other malignant diseases. Azathioprine is also commonly used to treat non-malignant immune diseases such as rheumatoid arthritis, systemic lupus erythematosus, inflammatory bowel disease, organ transplant rejection and others.

Common adverse reactions are gastrointestinal reactions such as myelosuppression, nausea, vomiting, loss of appetite and liver damage.

The US survey of paediatricians showed that 61% of paediatric patients were tested for TPMT genes before prescription of drugs. Another survey from non-oncologists doctor in the UK showed that 47%-94% patients were tested TPMT gene tests before prescription of drugs.

COMMON DRUGS

Common drugs containing steroids:

- Mercaptopurine tablets
- Thioguanine tablets
- Azathioprine tablets

DRUG USAGE

Thiopurines is commonly used for treatments as below:

- Leukemia
- Choriocarcinoma
- Inflammatory bowel disease
- Kidney transplant

Thiopurines

Gene Name	Variant's Name	Your Genotype	Description
TPMT	TPMT*2	CC	Rs1800462 is a variant of C to G, which converts the alanine into proline at position 80. This results in a decrease in TPMT activity leading to a reduced metabolic drug clearance, increased concentration of active metabolite TGN and eventually increased the risk of myelosuppression.
TPMT	TPMT*3A	CC	Rs1800460 is a variant of C to T, which converts the alanine into threonine at position 154. This results in a decrease in TPMT enzymatic activity leading to a reduced metabolic drug clearance, increased concentration of active metabolite TGN and eventually increased the risk of myelosuppression. This locus has a strong linkage disequilibrium with rs1142345 and two alleles mutates simultaneously are defined as *3A.
TPMT	TPMT*3C	TT	Rs1142345 is a variant of T to C, which converts the tyrosine to cysteine at position 240. This results in a decrease in TPMT enzymatic activity leading to a reduced metabolic drug clearance, increased concentration of active metabolite TGN and eventually increased the risk of myelosuppression.

Drug Sensitivity

Capecitabine

Capecitabine is an anticancer drug that can be converted into 5-FU in the body to inhibit cell division and interfere the RNA and protein synthesis. This medicine is mainly used to treat rectal cancer, colon cancer, breast cancer and gastric cancer. DPYD and MTHFR are the main related genes affecting the efficacy of capecitabine. Since Asian population has almost no DPYD mutation gene that can loss enzyme activity, this test mainly discusses the effect of MTHFR gene on capecitabine.

Your result:

Normal Response

You have normal response to capecitabine, you have GG type of MTHFR.

The use of capecitabine is generally effective.

According to the relevant researches, the same genotype may have a higher recurrence rate when using a chemotherapy regimen containing this drug.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Reduced MTHFR enzyme activity (AA/AG type) may lead to hyperhomocysteinemia and an increased risk of cardiovascular disease. It is recommended that people with AA/AG type of MTHFR to take supplements like folic acid and B vitamins. AA type of MTHFR accounted for 21% of the population while AG type accounted for 47% and GG type accounted for 32%.

COMMON DRUGS

Common drugs containing capecitabine:

- Capecitabine tablets

DRUG USAGE

Capecitabine is commonly used for treatments as below:

- Colorectal cancer
- Breast cancer
- Gastric cancer

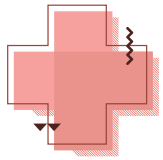
You need to pay attention to the use of the drug. Please communicate the following information to your doctor if:

- You are GG type of MTHFR.
- Your efficacy of capecitabine use is general. If you use a chemotherapy regimen containing this drug, the recurrence rate may be higher. It is recommended to inform doctor about your genotype.

Based on the above information, doctor may advise you:

- Perform other relevant tests to further determine your drug response
- Switch to other treatment options or replace with other drugs
- Do not make any changes and continue with the current medication plan

Gene Name	Variant's Name	Your Genotype	Description
MTHFR	g.11856378G>A	GG	The SNP locus rs1801133 on the MTHFR (methylenetetrahydrofolate reductase) gene carries the A mutation and the substitution of proline for alanine (Ala222Val) results in a decrease in MTHFR enzyme activity. Decreased enzyme activity caused an increase in intracellular 5,10-methylenetetrahydrofolate content, which combined with thymidylate synthase TS increased the cytotoxic effect of capecitabine and enhanced chemotherapy effect.



DRUG SENSITIVITY

OTHERS

Drug Sensitivity

Abacavir

Abacavir is a nucleoside analogue reverse transcriptase inhibitor and is commonly used in antiretroviral combination therapy for HIV infection. This drug is safe and the most serious adverse reaction is an allergic reaction. The HLA-B*57:01 gene is required to be detected before or after repeated use of abacavir according to each country's instructions. Due to the high risk of allergic reactions, abacavir is not recommended for patients with the HLA-B*57:01 allele.

Your result:

Good Response

Your are not the carrier of HLA-B*57:01 gene variant, your likelihood of experiencing side effect from abacavir administration is low.

Your use of abacavir is less likely to cause allergies.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Abacavir is mainly used to treat HIV infection. However, for AIDS patients carrying the HLA-B*57:01 variant gene that consume these drugs can lead to severe allergic reactions, including fever, rash, fatigue, nausea and vomiting. If the patient re-administers the drug after an allergic reaction, the allergic symptoms will be more serious and even life-threatening than when the drug is first administered.

In the western population, carrier of HLA-B*57:01 gene variant is relatively common, with a frequency of 6-7%. Up to 20% of the population in southwest Asia is a carrier. However, the frequency of carriers in East Asian and African populations is relatively low (<1%).

DRUG USAGE

Abacavir is commonly used for treatments as below:

- AIDS
- HIV infection

Gene Name	Variant's Name	Your Genotype	Description
HLA-B	HLA-B*57:01	TT	Rs2395029 is labelled as SNP of HLA-B*57:01. The positive predictive value for carrying the *57:01 allelic variation when carrying the G variant is about 50%, indicating that 50% of the individual carrying the G variant carries the *57:01 variation. However, the sensitivity, specificity and negative predictive value of this site were both higher than 99%.

Drug Sensitivity

Celecoxib

Celecoxib is a non-steroidal anti-inflammatory drug, which exerts antipyretic, analgesic and anti-inflammatory effects by specifically inhibiting cyclooxygenase-2. It is mainly used to relieve osteoarthritis, rheumatoid arthritis, ankylosing spondylitis, acute pain and can also prevent colorectal adenomas. Celecoxib is mainly metabolized by the CYP2C9 enzyme in the liver. Patients carrying the CYP2C9 mutation possess low enzyme activity that may cause celecoxib to accumulate in the body leading to an increased risk of adverse drug reactions.

Your result:

Good Response

Based on your genotype, your likelihood of experiencing side effect from celecoxib administration is low.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Celecoxib belongs to non-steroidal anti-inflammatory drugs (NSAIDs). These drugs mainly play a role in inhibiting the activity of cyclooxygenase COX. Non-specific anti-inflammatory drugs (such as ibuprofen, naproxen and others) inhibit both COX-1 and COX-2. Inhibition of COX-2 mainly produce analgesia, anti-inflammatory and other pharmacological effects while inhibition of COX-1 can cause adverse reactions such as gastrointestinal bleeding. Celecoxib belongs to the new generation of non-steroidal NSAIDs, which can specifically inhibit COX-2 and have a lower risk of adverse reactions.

Celecoxib was the first drug to justify the importance of genetic information. In the instructions of Celebrex, it is indicated that for patients with CYP2C9 *3/*3 genotype (homozygous variation), the initial dose should be halved when prescription; for juvenile arthritis patients, it is recommeded to change drug.

COMMON DRUGS

Common drugs containing celecoxib:

- Celebrex
- Celecoxib capsule

DRUG USAGE

Celecoxib is commonly used for treatments as below:

- Osteoarthritis
- Rheumatoid Arthritis
- Acute pain
- Ankylosing spondylitis

Gene Name	Variant's Name	Your Genotype	Description
CYP2C9	CYP2C9*3	AA	Rs1057910 is a variant of A to C, which converts the lysine into leucine at position 359. This missense mutation leads to a decrease in the activity of the liver CYP2C9 enzyme and a decrease in the metabolic clearance rate of the drug. Therefore, carrier of this variant should reduce the dose.

Drug Sensitivity

Sildenafil

Sildenafil (brand name: Viagra) is a highly selective phosphodiesterase 5 inhibitor that relaxes smooth muscle by enhancing the NO-cGMP pathway. Currently, it is not only used to treat male erectile dysfunction, but also for pulmonary hypertension. GNB3 is the main gene affecting the drug efficacy.

Your result:

Low Response

You have low reponse to sildenafil.

Your GNB3 genotype is CT or TT, according to the relevant researches, 50% of people who have the same genotype may not show effective response with sildenafil.

However, there is inadequate information to judge a drug response, so use it as a reference only.

Please consult a professional doctor or pharmacist before making any medication changes.

ABOUT

Erectile dysfunction (ED) is a common disease in adult males. The prevalence of ED in men aged 40-70 in the United States is 52%. At present, there are mainly three phosphodiesterase 5 (PDE5) inhibitors listed in the local market: sildenafil, vardenafil and tadalafil. The main adverse reactions of these drugs are hypotension, arrhythmia, transient visual loss or decreased vision, anxiety, seizures and others, which can lead to sudden cardiac death. In the Asian population, the GNB3 genotype distribution ratio is approximately 27% of CC type, 49.6% of CT type and 23.3% of TT type.

COMMON DRUGS

Common drugs containing sildenafil:

- Sildenafil citrate tablets
- Viagra

DRUG USAGE

Sildenafil is commonly used for treatments as below:

- Erectile dysfunction
- Pulmonary hypertension

Gene Name	Variant's Name	Your Genotype	Description
GNB3	C825T	TC	The variation of rs5443 from C to T results in the deletion of nucleotides at 498-620 region in exon 9 of GNB3 gene, which is characterized by a truncated but functional splice variant GNB3s with an enhanced signal transduction. When sildenafil is used, it is easier to exert its effects.

REFERENCES

ACOG Committee on Genetics. (2009). "ACOG Committee Opinion No. 442: Preconception and prenatal carrier screening for genetic diseases in individuals of Eastern European Jewish descent." Obstet Gynecol. 114(4):950-3.

ACOG Committee on Genetics. (2017). "ACOG Committee Opinion No. 691: Carrier screening for genetic conditions. Obstet Gynecol 2017;129:e41–55.

ACOG Committee on Genetics. (2017). "ACOG Committee Opinion No. 691: Carrier screening for genetic conditions. Obstet Gynecol. 129(3):e41–55.

Adams D et al. (2003). "Free Sialic Acid Storage Disorders." [Updated 2013 Jun 06].

Adato A et al. (2002). "USH3A transcripts encode clarin-1, a four-transmembrane-domain protein with a possible role in sensory synapses." Eur J Hum Genet. 10(6):339-50.

Ahmed M et al. (2001). "The beta-globin genotype E121Q/W15X (cd121GAA-->CAA/cd15TGG-->TGA) underlines Hb d/beta-(0) thalassaemia marked by domination of haemoglobin D." Ann Hematol. 80(11):629-33.

Akin H et al. (2010). "MEFV mutations in patients with Familial Mediterranean Fever from the Aegean region of Turkey." Mol Biol Rep. 37(1):93-8.

Akli S et al. (1993). "A null allele frequent in non-Jewish Tay-Sachs patients." Hum Genet. 90(6):614-20.

Akyol M, et al (2007). The common non-synonymous variant G38S of the KCNE1-(minK)-gene is not associated to QT interval in Central European Caucasians: results from the KORA study. Eur Heart J

Alagramam KN et al. (2011). "Mutations in protocadherin 15 and cadherin 23 affect tip links and mechanotransduction in mammalian sensory hair cells." PLoS One. 6(4):e19183.

Albert Min-Shan Ko , et al (2013). ALPK1 genetic regulation and risk in relation to gout. International Journal of Epidemiology

Alexey V. Polonikova, et al (2015). Alterederythrocytemembrane protein composition mirrors pleiotropic effects of hypertension susceptibility genes and disease pathogenesis. Journal of Hypertension.

Allen KJ et al., (2008) . Iron-overload-related Disease in HFE Hereditary Hemochromatosis . N Engl J Med

Amato A et al. (2014). "Carrier screening for inherited haemoglobin disorders among secondary school students and young adults in Latium, Italy." J Community Genet. 5(3):265-8.

American College of Obstetricians and Gynecologists Committee on Genetics. (2011). "ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis." Obstet Gynecol. 117(4):1028-31.

Amselem S et al. (1988). "Determination of the spectrum of beta-thalassemia genes in Spain by use of dot-blot analysis of amplified beta-globin DNA." Am J Hum Genet. 43(1):95-100.

Anderson S et al. (2012). "Medium chain acyl-CoA dehydrogenase deficiency detected among Hispanics by New Jersey newborn screening." Am J Med Genet A. 158A(9):2100-5.

Andreotti G et al. (2013). "Biochemical phenotype of a common disease-causing mutation and a possible therapeutic approach for the phosphomannomutase 2-associated disorder of glycosylation." Mol Genet Genomic Med. 1(1):32-44.

Andresen BS et al. (2001). "Medium-chain acyl-CoA dehydrogenase (MCAD) mutations identified by MS/MS-based prospective screening of newborns differ from those observed in patients with clinical symptoms: identification and characterization of a new, prevalent mutation that results in mild MCAD deficiency." Am J Hum Genet. 68(6):1408-18.

Andrew D. Johnson, et al (2010). Association of hypertension drug target genes with blood pressure and hypertension in 86,588 individuals. Hypertension.

Angileri, Francesca et al. (2015). "Geographical and Ethnic Distribution of Mutations of the Fumarylacetoacetate Hydrolase Gene in Hereditary Tyrosinemia Type 1." JIMD Reports 19:43–58.

Antonarakis SE et al. (1984). "beta-Thalassemia in American Blacks: novel mutations in the "TATA" box and an acceptor splice site." Proc Natl Acad Sci U S A. 81(4):1154-8.

Arnold GL et al. (2010). "Lack of genotype-phenotype correlations and outcome in MCAD deficiency diagnosed by newborn screening in New York State." Mol Genet Metab. 99(3):263-8.

Arranz JA et al. (2002). "Splicing mutations, mainly IVS6-1(G>T), account for 70% of fumarylacetoacetate hydrolase (FAH) gene alterations, including 7 novel mutations, in a survey of 29 tyrosinemia type I patients." Hum Mutat. 20(3):180-8.

Aula N et al. (2000). "The spectrum of SLC17A5-gene mutations resulting in free sialic acid-storage diseases indicates some genotype-phenotype correlation." Am J Hum Genet. 67(4):832-40.

Aula N et al. (2002). "Unraveling the molecular pathogenesis of free sialic acid storage disorders: altered targeting of mutant sialin." Mol Genet Metab. 77(1-2):99-107.

Aulehla-Scholz C et al. (1990). "Molecular basis of beta-thalassemia in Turkey: detection of rare mutations by direct sequencing." Hum Genet. 84(2):195-7.

Avramova Z et al. (1988). "Metabolic behaviour of a stable DNA-protein complex." Int J Biochem. 20(1):61-5.

Ayesh SK et al. (2005). "Genetic screening of familial Mediterranean fever mutations in the Palestinian population." Saudi Med J. 26(5):732-7.

Bacon BR et al., (2011) . Diagnosis and management of hemochromatosis: 2011 practice guideline by the American Association for the Study of Liver Diseases . Hematology

Bali DS et al. (2006). "Glycogen Storage Disease Type I." [Updated 2016 Aug 25].

Bali DS et al. (2006). "Glycogen Storage Disease Type I." [Updated 2016 Aug 25].

Baojun Wang, et al (2010). Association of DNA Polymorphisms Within the CYP11B2/CYP11B1 Locus and Postoperative Hypertension Risk in the Patients With Aldosterone-producing Adenomas. Basic and Translational Science.

REFERENCES

Bardelli T et al. (2002). "Two novel genetic lesions and a common BH4-responsive mutation of the PAH gene in Italian patients with hyperphenylalaninemia." Mol Genet Metab. 77(3):260-6.

Bartoli M et al. (2008). "Mannosidase I inhibition rescues the human alpha-sarcoglycan R77C recurrent mutation." Hum Mol Genet. 17(9):1214-21.

Bellizzi JJ 3rd et al. (2000). "The crystal structure of palmitoyl protein thioesterase 1 and the molecular basis of infantile neuronal ceroid lipofuscinosis." Proc Natl Acad Sci U S A. 97(9):4573-8.

Ben-Chetrit E et al. (2000). "The E148Q mutation in the MEFV gene: is it a disease-causing mutation or a sequence variant?" Hum Mutat. 15(4):385-6.

Bender MA et al. (2003). "Sickle Cell Disease." [Updated 2017 Aug 17].

Ben-Yosef T et al. (2003). "A mutation of PCDH15 among Ashkenazi Jews with the type 1 Usher syndrome." N Engl J Med. 348(17):1664-70.

Bergman AJ et al. (1998). "Spectrum of mutations in the fumarylacetoacetate hydrolase gene of tyrosinemia type 1 patients in northwestern Europe and Mediterranean countries." Hum Mutat. 12(1):19-26.

Bergmann C et al. (2003). "Spectrum of mutations in the gene for autosomal recessive polycystic kidney disease (ARPKD/PKHD1)." J Am Soc Nephrol. 14(1):76-89.

Bergmann C et al. (2005). "Clinical consequences of PKHD1 mutations in 164 patients with autosomal-recessive polycystic kidney disease (ARPKD)." Kidney Int. 67(3):829-48.

Beutler E et al. (1991). "Identification of the second common Jewish Gaucher disease mutation makes possible population-based screening for the heterozygous state." Proc Natl Acad Sci U S A. 88(23):10544-7.

Beutler E et al. (1992). "Mutations in Jewish patients with Gaucher disease." Blood. 79(7):1662-6.

Bierut LJ, et al (2010). A genome-wide association study of alcohol dependence. Proc Natl Acad Sci U S A.

Bik-Multanowski M et al. (2013). "Molecular genetics of PKU in Poland and potential impact of mutations on BH4 responsiveness." Acta Biochim Pol. 60(4):613-6.

BIOPKU Database

Bjursell C et al. (2000). "PMM2 mutation spectrum, including 10 novel mutations, in a large CDG type 1A family material with a focus on Scandinavian families." Hum Mutat. 16(5):395-400.

Bliksrud YT et al. (2012). "Hereditary tyrosinaemia type I in Norway: Incidence and three novel small deletions in the fumarylacetoacetase gene." Scand J Clin Lab. Invest. 72(5):369–373.

Bo Xi, et al (2010). Polymorphism near the ATP2B1 gene is associated with hypertension risk in East Asians: A meta-analysis involving 15 909cases and 18 529 controls. Blood Pressure.

Bobadilla JL et al. (2002). "Cystic fibrosis: a worldwide analysis of CFTR mutations--correlation with incidence data and application to screening." Hum Mutat. 19(6):575-606.

Boito CA et al. (2005). "Clinical and molecular characterization of patients with limb-girdle muscular dystrophy type 2I." Arch Neurol. 62(12):1894-9.

Boles DJ et al. (1995). "The molecular basis of HEXA mRNA deficiency caused by the most common Tay-Sachs disease mutation." Am J Hum Genet. 56(3):716-24.

Bonnet C et al. (2012). "Usher syndrome (sensorineural deafness and retinitis pigmentosa): pathogenesis, molecular diagnosis and therapeutic approaches." Curr Opin Neurol. 25(1):42-9.

Bonyadi M et al. (2009). "MEFV mutations in Iranian Azeri Turkish patients with familial Mediterranean fever." Clin Genet. 76(5):477-80.

Boonyawat B et al. (2014). "Molecular analysis of beta-globin gene mutations among Thai beta-thalassemia children: results from a single center study." Appl Clin Genet. 7:253-8.

Borck G et al. (2003). "Mutations in the PDS gene in German families with Pendred's syndrome: V138F is a founder mutation." J Clin Endocrinol Metab. 88(6):2916-21.

Bourteel H et al. (2009). "Clinical and mutational spectrum of limb-girdle muscular dystrophy type 2I in 11 French patients." J Neurol Neurosurg Psychiatry. 80(12):1405-8.

Brassier A et al. (2013). "Dihydrolipoamide dehydrogenase deficiency: a still overlooked cause of recurrent acute liver failure and Reye-like syndrome." Mol Genet Metab. 109(1):28-32.

Braverman N et al. (1997). "Human PEX7 encodes the peroxisomal PTS2 receptor and is responsible for rhizomelic chondrodysplasia punctata." Nat Genet. 15(4):369-76.

Braverman N et al. (2002). "Mutation analysis of PEX7 in 60 probands with rhizomelic chondrodysplasia punctata and functional correlations of genotype with phenotype." Hum Mutat. 20(4):284-97.

Braverman NE et al. (2001). "Rhizomelic Chondrodysplasia Punctata Type 1." [Updated 2012 Sep 13].

Brites P et al. (1998). "Molecular basis of rhizomelic chondrodysplasia punctata type I: high frequency of the Leu-292 stop mutation in 38 patients." J Inherit Metab Dis. 21(3):306-8.

Brockington M et al. (2001). "Mutations in the fukutin-related protein gene (FKRP) identify limb girdle muscular dystrophy 2I as a milder allelic variant of congenital muscular dystrophy MDC1C." Hum Mol Genet. 10(25):2851-9.

Brownstein Z et al. (2004). "The R245X mutation of PCDH15 in Ashkenazi Jewish children diagnosed with nonsyndromic hearing loss foreshadows retinitis pigmentosa." Pediatr Res. 55(6):995-1000.

Buch S, et al (2007). A genome-wide association scan identifies the hepatic cholesterol transporter ABCG8 as a susceptibility factor for human gallstone disease. Nat. Genet.

Busslinger M et al. (1981). "Beta + thalassemia: aberrant splicing results from a single point mutation in an intron." Cell. 27(2 Pt 1):289-98.

Canatan D et al. (2006). "Hemoglobinopathy control program in Turkey." Community Genet. 9(2):124-6.

REFERENCES

Carrié A et al. (1997). "Mutational diversity and hot spots in the alpha-sarcoglycan gene in autosomal recessive muscular dystrophy (LGMD2D)." J Med Genet. 34(6):470-5.

Cartier EA et al. (2001). "Defective trafficking and function of KATP channels caused by a sulfonylurea receptor 1 mutation associated with persistent hyperinsulinemic hypoglycemia of infancy." Proc Natl Acad Sci U S A. 98(5):2882-7.

Cazeneuve C et al. (1999). "MEFV-Gene analysis in armenian patients with Familial Mediterranean fever: diagnostic value and unfavorable renal prognosis of the M694V homozygous genotype-genetic and therapeutic implications." Am J Hum Genet. 65(1):88-97.

Cerreto M et al. (2011). "Natural phenylalanine hydroxylase variants that confer a mild phenotype affect the enzyme's conformational stability and oligomerization equilibrium." Biochim Biophys Acta. 1812(11):1435-45.

Ceylan GG et al. (2012). "Frequency of alterations in the MEFV gene and clinical signs in familial Mediterranean fever in Central Anatolia, Turkey." Genet Mol Res. 11(2):1185-94.

Chan DK et al. (2014). "GJB2-associated hearing loss: systematic review of worldwide prevalence, genotype, and auditory phenotype." Laryngoscope. 124(2):E34-53.

Chang KH, et al (2013). Association between PARK16 and Parkinson's disease in the Han Chinese population: a meta-analysis. Neurobiol Aging

Chang XL, et al (2011). Association of GWAS loci with PD in China. Am J Med Genet B Neuropsychiatr Genet

Chehab FF et al. (1987). "The molecular basis of beta-thalassemia in Lebanon: application to prenatal diagnosis." Blood. 69(4):1141-5.

Chen H, et al (2010). Ethnicity-based subgroup meta-analysis of the association of LOXL1 polymorphisms with glaucoma. Mol Vis.

Chen LY et al. (2000). "Structural requirements for the stability and microsomal transport activity of the human glucose 6-phosphate transporter." J Biol Chem. 275(44):34280-6.

Cheng TC et al. (1984). "beta-Thalassemia in Chinese: use of in vivo RNA analysis and oligonucleotide hybridization in systematic characterization of molecular defects." Proc Natl Acad Sci U S A. 81(9):2821-5.

Cheng X, et al (2011). The Same Chromosome 9p21.3 Locus Is Associated With Type 2 Diabetes and Coronary Artery Disease in a Chinese Han Population. Diabetes.

Chibo Liu, et al (2010) . Pooled Analyses of the Associations of Polymorphisms in the GRK4 and EMILIN1 Genes with Hypertension Risk. International Journal of Medical Sciences.

Cho MH, et al (2010). Variants in FAM13A are associated with chronic obstructive pulmonary disease. Nat Genet.

Choi BY et al. (2009). "Hypo-functional SLC26A4 variants associated with nonsyndromic hearing loss and enlargement of the vestibular aqueduct: genotype-phenotype correlation or coincidental polymorphisms?" Hum Mutat. 30(4):599-608.

Chou JY et al. (2002). "Type I glycogen storage diseases: disorders of the glucose-6-phosphatase complex." Curr Mol Med. 2(2):121-43.

Chou JY et al. (2008). "Mutations in the glucose-6-phosphatase-alpha (G6PC) gene that cause type Ia glycogen storage disease." Hum Mutat. 29(7):921-30.

Christopher T Johansen, et al (2010). Excess of rare variants in genes identified by genome-wide association study of hypertriglyceridemia. Nature Genetics

Chrzanowska KH et al. (2012). "Nijmegen breakage syndrome (NBS)." Orphanet J Rare Dis. 7:13.

Chu X, et al (2011). A genome-wide association study identifies two new risk loci for Graves' disease. Nat Genet.

Coffee EM et al. (2010). "Increased prevalence of mutant null alleles that cause hereditary fructose intolerance in the American population." J Inherit Metab Dis. 33(1):33-42.

Coffee EM et al. (2010). "Mutations in the promoter region of the aldolase B gene that cause hereditary fructose intolerance." J Inherit Metab Dis. 33(6):715-25.

Cohen M et al. (2007). "The changing face of Usher syndrome: clinical implications." Int J Audiol. 46(2):82-93.

Cohen M et al. (2007). "The changing face of Usher syndrome: clinical implications." Int J Audiol. 46(2):82-93.

Cohn ES et al. (1999). "Clinical phenotype and mutations in connexin 26 (DFNB1/GJB2), the most common cause of childhood hearing loss." Am J Med Genet. 89(3):130-6.

Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.

Cottrell CE et al. (2012). "Maternal uniparental disomy of chromosome 4 in a patient with limb-girdle muscular dystrophy 2E confirmed by SNP array technology." Clin Genet. 81(6):578-83.

Coyle B et al. (1998). "Molecular analysis of the PDS gene in Pendred syndrome." Hum Mol Genet. 7(7):1105-12.

Cross NC et al. (1990). "A new aldolase B variant, N334K, is a common cause of hereditary fructose intolerance in Yugoslavia." Nucleic Acids Res. 18(7):1925.

D'Andrea P et al. (2002). "Hearing loss: frequency and functional studies of the most common connexin26 alleles." Biochem Biophys Res Commun. 296(3):685-91.

Daniel F, et al (2010). Association of Variants at UMOD with Chronic Kidney Disease and Kidney Stones—Role of Age and Comorbid Diseases. PLoS Genet.

Darbari DS et al. (2013). "Severe painful vaso-occlusive crises and mortality in a contemporary adult sickle cell anemia cohort study." PLoS One. 8(11):e79923.

Das AK et al. (1998). "Molecular genetics of palmitoyl-protein thioesterase deficiency in the U.S." J Clin Invest. 102(2):361-70.

REFERENCES

Das AK et al. (2001). "Biochemical analysis of mutations in palmitoyl-protein thioesterase causing infantile and late-onset forms of neuronal ceroid lipofuscinosis." Hum Mol Genet. 10(13):1431-9.

David Celorrio, et al (2016). Influence of Dopaminergic System Genetic Variation and Lifestyle Factors on Excessive Alcohol Consumption. Alcohol and Alcoholism

Dazzo C et al. (1990). "Molecular evidence for compound heterozygosity in hereditary fructose intolerance." Am J Hum Genet. 46(6):1194-9.

De Braekeleer M et al. (1990). "Genetic epidemiology of hereditary tyrosinemia in Quebec and in Saguenay-Lac-St-Jean." Am J Hum Genet. 47(2):302-7.

De Braekeleer M et al. (1993). "Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Saguenay in northeastern Quebec." Genet Epidemiol. 10(1):17-25.

De Laurenzi V et al. (1997). "Sjögren-Larsson syndrome is caused by a common mutation in northern European and Swedish patients." J Invest Dermatol. 109(1):79-83.

Debray FG et al. (2011). "LRPPRC mutations cause a phenotypically distinct form of Leigh syndrome with cytochrome c oxidase deficiency." J Med Genet. 48(3):183-9.

Dechamethakun S, et al (2014). Associations between the CDKN2A/B, ADTRP and PDGFD polymorphisms and the development of coronary atherosclerosis in Japanese patients. J Atheroscler Thromb.

Dehghan A et al. (2008) . "Association of three genetic loci with uric acid concentration and risk of gout: a genome-wide association study." 372(9654):1953-61. Lancet

Denny JC, et al (2011). Variants Near FOXE1 Are Associated with Hypothyroidism and Other Thyroid Conditions: Using Electronic Medical Records for Genome- and Phenome-wide Studies. Am J Hum Genet.

Desviat LR et al. (2004). "Tetrahydrobiopterin responsiveness: results of the BH4 loading test in 31 Spanish PKU patients and correlation with their genotype." Mol Genet Metab. 83(1-2):157-62.

Dichgans M, et al (2014). Shared Genetic Susceptibility to Ischemic Stroke and Coronary Artery Disease. Stroke

Djordjevic M et al. (2013). "Molecular Genetics and Genotype-Based Estimation of BH4-Responsiveness in Serbian PKU Patients: Spotlight on Phenotypic Implications of p.L48S." JIMD Rep. 9:49-58.

Dobrowolski SF et al. (2011). "Molecular genetics and impact of residual in vitro phenylalanine hydroxylase activity on tetrahydrobiopterin responsiveness in Turkish PKU population." Mol Genet Metab. 102(2):116-21.

Dodé C et al. (2000). "Mutations in the MEFV gene in a large series of patients with a clinical diagnosis of familial Mediterranean fever." Am J Med Genet. 92(4):241-6.

Donahue SL et al. (2004). "Intermediate DNA repair activity associated with the 322delG allele of the fanconi anemia complementation group C gene." J Mol Biol. 342(5):1443-55.

Dong J et al. (2001). "Nonradioactive detection of the common Connexin 26 167delT and 35delG mutations and frequencies among Ashkenazi Jews." Mol Genet Metab. 73(2):160-3.

Dong L, et al (2011) Correlation of complement factor H gene polymorphisms with exudative age-related macular degeneration in a Chinese cohort. Neurosci Lett.

Draviam RA et al. (2006). "Alpha-sarcoglycan is recycled from the plasma membrane in the absence of sarcoglycan complex assembly." Traffic. 7(7):793-810.

Dubois PC, et al(2010). Multiple common variants for celiac disease influencing immune gene expression. Nat Genet.

Duclos F et al. (1998). "Beta-sarcoglycan: genomic analysis and identification of a novel missense mutation in the LGMD2E Amish isolate." Neuromuscul Disord. 8(1):30-8.

Dunne MJ et al. (2004). "Hyperinsulinism in infancy: from basic science to clinical disease." Physiol Rev. 84(1):239-75.

Dupré N et al. (2003). "Hereditary motor and sensory neuropathy with agenesis of the corpus callosum." Ann Neurol. 54(1):9-18.

Dupré N et al. (2006). "Hereditary ataxia, spastic paraparesis and neuropathy in the French-Canadian population." Can J Neurol Sci. 33(2):149-57.

Dupré N et al. (2006). "Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum". [Updated 2014 June 12].

Dzikiewicz-Krawczyk A et al. (2012). "Impact of heterozygous c.657-661del, p.I171V and p.R215W mutations in NBN on nibrin functions." Mutagenesis. 27(3):337-43.

Ebberink MS et al. (2011). "Genetic classification and mutational spectrum of more than 600 patients with a Zellweger syndrome spectrum disorder." Hum Mutat. 32(1):59-69.

Edelmann L et al. (2001). "Maple syrup urine disease: identification and carrier-frequency determination of a novel founder mutation in the Ashkenazi Jewish population." Am J Hum Genet. 69(4):863-8.

Ekstein J et al. (2004). "Mutation frequencies for glycogen storage disease Ia in the Ashkenazi Jewish population." Am J Med Genet A. 129A(2):162-4.

Elena V. Kazakova, et al (2015). Association between UBE2E2 variant rs7612463 and type 2 diabetes mellitus in a Chinese Han Population. Acta Biochim Pol

Ellinor PT, et al (2010). Common variants in KCNN3 are associated with lone atrial fibrillation. Nat Genet.

Ellis NA et al. (1998). "The Ashkenazic Jewish Bloom syndrome mutation blmAsh is present in non-Jewish Americans of Spanish ancestry." Am J Hum Genet. 63(6):1685-93.

Elpeleg ON et al. (2002). "Mutation analysis of the FAH gene in Israeli patients with tyrosinemia type I." Hum Mutat. 19(1):80-1.

Elstein D et al. (2005). "Phenotypic heterogeneity in patients with Gaucher disease and the N370S/V394L genotype." Genet Test. 9(1):26-9.

Engert JC et al. (2000). "ARSACS, a spastic ataxia common in northeastern Québec, is caused by mutations in a new gene encoding an 11.5-kb ORF." Nat Genet. 24(2):120-5.

REFERENCES

Erikson A et al. (2002). "Free sialic acid storage (Salla) disease in Sweden." Acta Paediatr. 91(12):1324-7.

Eriksson N, et al (2012). Novel Associations for Hypothyroidism Include Known Autoimmune Risk Loci. PLoS ONE.

Esposito G et al. (2002). "Structural and functional analysis of aldolase B mutants related to hereditary fructose intolerance." FEBS Lett. 531(2):152-6.

Fabry ME et al. (1981). "Some aspects of the pathophysiology of homozygous Hb CC erythrocytes." J Clin Invest. 67(5):1284-91.

Fan Wang, et al (2011). Genome-wide association identifies a susceptibility locus for coronary artery disease in the Chinese Han population. Nature Genetics.

Faraco J, et al (2013). ImmunoChip study implicates antigen presentation to T cells in narcolepsy. PLoS Genet

Farrer LA, et al (1997). Effects of age, sex, and ethnicity on the association between apolipoprotein E genotype and Alzheimer disease. A meta-analysis. APOE and Alzheimer Disease Meta Analysis Consortium. JAMA

Fattoum S et al. (1993). "HbC/beta-thalassemia association. Eleven cases observed in Tunisia." Ann Pediatr (Paris). 40(1):45-8.

Faustino P et al. (1992). "Novel promoter and splice junction defects add to the genetic, clinical or geographic heterogeneity of beta-thalassaemia in the Portuguese population." Hum Genet. 89(5):573-6.

Fei Zhu, et al(2013). Association study confirmed susceptibility loci with keloid in the Chinese Han population. PLoS One.

Feigenbaum A et al. (2004). "Canavan disease: carrier-frequency determination in the Ashkenazi Jewish population and development of a novel molecular diagnostic assay." Am J Med Genet A. 124A(2):142-7.

Fellman V et al. (2008). "Screening of BCS1L mutations in severe neonatal disorders suspicious for mitochondrial cause." J Hum Genet. 53(6):554-8.

Fellman V. (2002). "The GRACILE syndrome, a neonatal lethal metabolic disorder with iron overload." Blood Cells Mol Dis. 29(3):444-50.

Ferdinandusse S et al. (2006). "Clinical and biochemical spectrum of D-bifunctional protein deficiency." Ann Neurol. 59(1):92-104.

Ferdinandusse S et al. (2006). "Mutational spectrum of D-bifunctional protein deficiency and structure-based genotype-phenotype analysis." Am J Hum Genet. 78(1):112-24.

Ferreira, et al (2008). Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. Nat. Genet.

Fields RR et al. (2002). "Usher syndrome type III: revised genomic structure of the USH3 gene and identification of novel mutations." Am J Hum Genet. 71(3):607-17.

Fingerlin TE, et al (2013). Genome-wide association study identifies multiple susceptibility loci for pulmonary fibrosis. Nat Genet.

Flaschker N et al. (2007). "Description of the mutations in 15 subjects with variant forms of maple syrup urine disease." J Inherit Metab Dis. 30(6):903-9.

Fletcher LM et al., (2002) . Excess Alcohol Greatly Increases the Prevalence of Cirrhosis in Hereditary Hemochromatosis . Gastroenterology

Floeth M et al. (1999). "Digenic junctional epidermolysis bullosa: mutations in COL17A1 and LAMB3 genes." Am J Hum Genet. 65(6):1530-7.

Forrest SM et al. (1991). "Mutation detection in phenylketonuria by using chemical cleavage of mismatch: importance of using probes from both normal and patient samples." Am J Hum Genet. 49(1):175-83.

Fumihiko Takeuchi , et al (2013). Genetic Impact on Uric Acid Concentration and Hyperuricemia in the Japanese Population. J Atheroscler Thromb

Fuu-Jen Tsai, et al (2010). A Genome-Wide Association Study Identifies Susceptibility Variants for Type 2 Diabetes in Han Chinese. PLoS Genetics

Gallego CJ et al., (2015) . Penetrance of Hemochromatosis in HFE Genotypes Resulting in p.Cys282Tyr and p.[Cys282Tyr];[His63Asp] in the eMERGE Network . Am J Hum Genet

Gånemo A et al. (2009). "Sjögren-larsson syndrome: a study of clinical symptoms and dermatological treatment in 34 Swedish patients." Acta Derm Venereol. 89(1):68-73.

Garcia-Gonzalez MA et al. (2007). "Genetic interaction studies link autosomal dominant and recessive polycystic kidney disease in a common pathway." Hum Mol Genet. 16(16):1940-50.

Garewal G et al. (2005). "Nucleotide -88 (C-T) promoter mutation is a common beta-thalassemia mutation in the Jat Sikhs of Punjab, India." Am J Hematol. 79(4):252-6.

German J et al. (2007). "Syndrome-causing mutations of the BLM gene in persons in the Bloom's Syndrome Registry." Hum Mutat. 28(8):743-53.

Gersting SW et al. (2008). "Loss of function in phenylketonuria is caused by impaired molecular motions and conformational instability." Am J Hum Genet. 83(1):5-17.

Gharavi AG, et al (2011). Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nat Genet.

Giambona A et al. (2015). "Incidence of haemoglobinopathies in Sicily: the impact of screening and prenatal diagnosis." Int J Clin Pract. 69(10):1129-38.

Gille JJ et al. (2012). "Diagnosis of Fanconi Anemia: Mutation Analysis by Multiplex Ligation-Dependent Probe Amplification and PCR-Based Sanger Sequencing." Anemia. 2012:603253.

Gillio AP et al. (1997). "Phenotypic consequences of mutations in the Fanconi anemia FAC gene: an International Fanconi Anemia Registry study." Blood. 90(1):105-10.

REFERENCES

Gjetting T et al. (2001). "In vitro expression of 34 naturally occurring mutant variants of phenylalanine hydroxylase: correlation with metabolic phenotypes and susceptibility toward protein aggregation." Mol Genet Metab. 72(2):132-43.

Glaser B et al. (2003). "Familial Hyperinsulinism." [Updated 2013 Jan 24]

Glaser B et al. (2011). "ABCC8 mutation allele frequency in the Ashkenazi Jewish population and risk of focal hyperinsulinemic hypoglycemia." Genet Med. 13(10):891-4.

Gong LL, et al (2012). Association of tissue plasminogen activator and plasminogen activator inhibitor polymorphism with myocardial infarction: a meta-analysis. Thromb Res

Gonzalez Trevino O et al. (2001). "Clinical and molecular analysis of three Mexican families with Pendred's syndrome." Eur J Endocrinol. 144(6):585-93.

Gonzalez-Redondo JM et al. (1988). "Clinical and genetic heterogeneity in black patients with homozygous beta-thalassemia from the southeastern United States." Blood. 72(3):1007-14.

Grace ME et al. (1994). "Analysis of human acid beta-glucosidase by site-directed mutagenesis and heterologous expression." J Biol Chem. 269(3):2283-91.

Gray GR et al. (1995). "Hb Lulu Island (alpha 2 beta 2 107[G9]Gly-->Asp)-beta zero- thalassemia (codon 15; TGG-->TAG), a form of thalassemia intermedia." Am J Hematol. 50(1):26-9.

Gregersen N et al. (1993). "Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency: the prevalent mutation G985 (K304E) is subject to a strong founder effect from northwestern Europe." Hum Hered. 43(6):342-50.

Gregory, et al (2007). Interleukin 7 receptor alpha chain (IL7R) shows allelic and functional association with multiple sclerosis. Nat Genet.

Grompe M et al. (1994). "A single mutation of the fumarylacetoacetate hydrolase gene in French Canadians with hereditary tyrosinemia type I." N Engl J Med. 331(6):353-7.

Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.

Gudbjartsson DF, et al (2010). Association of variants at UMOD with chronic kidney disease and kidney stones-role of age and comorbid diseases. PLoS Genet.

Gudbjartsson DF, et al (2012). Variants conferring risk of atrial fibrillation on chromosome 4q25. Nature

Gudmar Thorleifsson, et al (2009). Sequence variants in the CLDN14 gene associate with kidney stones and bone mineral density. Nat Genet.

Gunay-Aygun M et al. (2010). "PKHD1 sequence variations in 78 children and adults with autosomal recessive polycystic kidney disease and congenital hepatic fibrosis." Mol Genet Metab. 99(2):160-73.

H. S. , et al (2011). Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. Circ Cardiovasc Genet.

Hackman P et al. (2005). "Enrichment of the R77C alpha-sarcoglycan gene mutation in Finnish LGMD2D patients." Muscle Nerve. 31(2):199-204.

Han F, et al(2012). TCRA, P2RY11, and CPT1B/CHKB associations in Chinese narcolepsy. Sleep Med.

Han S et al. (2016). "Deafness gene mutations in newborns in Beijing." Acta Otolaryngol. 136(5):475-9.

Han SH et al. (2008). "Carrier frequency of GJB2 (connexin-26) mutations causing inherited deafness in the Korean population." J Hum Genet. 53(11-12):1022-8.

Han X, et al (2014). Association between phosphatase related gene variants and coronary artery disease: case-control study and meta-analysis. Int J Mol Sci.

Hao, et al (2008) Two Polymorphisms in NEDD4L Gene and Essential Hypertension in Chinese Hans ¾A Population-Based Case-Control Study. Clinical and Experimental Hypertension.

Harold D, et al (2009) Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. Nat Genet.

HbVar Database, Penn State University.

Healy et al. (2008) . "Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study." 7(7):583-90. Lancet Neurol

Hechtman P et al. (1990). "More than one mutant allele causes infantile Tay-Sachs disease in French-Canadians." Am J Hum Genet. 47(5):815-22.

Herrera W et al. (2008). "Retinal disease in Usher syndrome III caused by mutations in the clarin-1 gene." Invest Ophthalmol Vis Sci. 49(6):2651-60.

Hiebler S et al. (2014). "The Pex1-G844D mouse: a model for mild human Zellweger spectrum disorder." Mol Genet Metab. 111(4):522-32.

Hirokawa M, et al(2014). A genome-wide association study identifies PLCL2 and AP3D1-DOT1L-SF3A2 as new susceptibility loci for myocardial infarction in Japanese. Eur J Hum Genet.

Hirota T, et al (2012). Genome-wide association study identifies eight new susceptibility loci for atopic dermatitis in the Japanese population. Nat Genet.

Hirschfield GM, et al (2009). Primary biliary cirrhosis associated with HLA, IL12A, and IL12RB2 variants. N Engl J Med.

Hirschfield GM, et al (2010). Variants at IRF5-TNPO3, 17q12-21 and MMEL1 are associated with primary biliary cirrhosis. Nat Genet.

Holmberg V et al. (2000). "Phenotype-genotype correlation in eight patients with Finnish variant late infantile NCL (CLN5)." Neurology. 55(4):579-81.

Hoshino H et al. (2014). "Canavan disease: clinical features and recent advances in research." Pediatr Int. 56(4):477-83.

REFERENCES

Hou DC et al. (1999). "Glycogen storage disease type Ib: structural and mutational analysis of the microsomal glucose-6-phosphate transporter gene." Am J Med Genet. 86(3):253-7.

Howard HC et al. (2002). "The K-Cl cotransporter KCC3 is mutant in a severe peripheral neuropathy associated with agenesis of the corpus callosum." Nat Genet. 32(3):384-92.

Hsu HW et al. (2008). "Spectrum of medium-chain acyl-CoA dehydrogenase deficiency detected by newborn screening." Pediatrics. 121(5):e1108-14.

Hu C et al. (2009) Variations in KCNQ1 are associated with type 2 diabetes and beta cell function in a Chinese population. Diabetologia 52(7):1322-5

Huang S et al. (1986). "The same "TATA" box beta-thalassemia mutation in Chinese and US blacks: another example of independent origins of mutation." Hum Genet. 74(2):162-4.

Huang S et al. (2011). "Extremely discrepant mutation spectrum of SLC26A4 between Chinese patients with isolated Mondini deformity and enlarged vestibular aqueduct." J Transl Med. 9:167.

Huard CC et al. (2014). "The Fanconi anemia pathway has a dual function in Dickkopf-1 transcriptional repression." Proc Natl Acad Sci U S A. 111(6):2152-7.

Huopio H et al. (2002). "Acute insulin response tests for the differential diagnosis of congenital hyperinsulinism." J Clin Endocrinol Metab. 87(10):4502-7.

Hwa HL et al. (2003). "Mutation spectrum of the connexin 26 (GJB2) gene in Taiwanese patients with prelingual deafness." Genet Med. 5(3):161-5.

Ikram MA, et al (2009). Genomewide association studies of stroke. N Engl J Med.

Imtiaz F et al. (2011). "Identification of mutations causing hereditary tyrosinemia type I in patients of Middle Eastern origin." Mol Genet Metab. 104(4):688-90.

International Stroke Genetics Consortium (ISGC), Wellcome Trust Case Control Consortium 2 (WTCCC2), et al (2012). Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke. Nat Genet.

Iron Disorder Institute www.irondisorders.org/diet/

Isosomppi J et al. (2002). "Lysosomal localization of the neuronal ceroid lipofuscinosis CLN5 protein." Hum Mol Genet. 11(8):885-91.

Jagell S et al. (1981). "Sjögren-Larsson syndrome in Sweden. A clinical, genetic and epidemiological study." Clin Genet. 19(4):233-56.

James M, et al (2010). Genetic variants of STAT4 associated with rheumatoid arthritis in persons of Asian and European ancestry do not replicate in African-Americans. Ann Rheum Di.

Janecke AR et al. (2000). "Mutation analysis in glycogen storage disease type 1 non-a." Hum Genet. 107(3):285-9.

Jank JM et al. (2014). "The domain-specific and temperature-dependent protein misfolding phenotype of variant medium-chain acyl-CoA dehydrogenase." PLoS One. 9(4):e93852.

Jarjour RA. (2010). "Familial Mediterranean fever in Syrian patients: MEFV gene mutations and genotype-phenotype correlation." Mol Biol Rep. 37(1):1-5.

Jeong-Hyun Kim , et al (2010). A new association between polymorphisms of the SLC6A7 gene in the chromosome 5q31–32 region and asthma. Journal of Human Genetics.

Jiang H, et al (2009) Analyses of single nucleotide polymorphisms and haplotype linkage of LOC387715 and the HTRA1 gene in exudative age-related macular degeneration in a Chinese cohort. Retina.

Jiang L, et al(2014). Novel risk loci for rheumatoid arthritis in Han Chinese and congruence with risk variants in Europeans. Arthritis & Rheumatology

Jin Y, et al (2010). Variant of TYR and autoimmunity susceptibility loci in generalized vitiligo. N Engl J Med.

Joensuu T et al. (2001). "Mutations in a novel gene with transmembrane domains underlie Usher syndrome type 3." Am J Hum Genet. 69(4):673-84.

Jorge Esparza-Gordillo, et al (2009). A common variant on chromosome 11q13 is associated with atopic dermatitis Nat Genet.

Jun Tian, et al (2012) Association of Genetic Polymorphisms and Age-related Macular Degeneration in Chinese population. Invest Ophthalmol Vis Sci.

Jungang Xie, et al (2015). Gene susceptibility identification in a longitudinal study confirms new loci in the development of chronic obstructive pulmonary disease and influences lung function decline. Respiratory Research.

Kääb et al. (2009) . "Large scale replication and meta-analysis of variants on chromosome 4q25 associated with atrial fibrillation." 30(7):813-9. Eur. Heart J.

Kaback MM et al. (1999). "Hexosaminidase A Deficiency." [Updated 2011 Aug 11].

Kang PB et al. (2007). "LGMD2I in a North American population." BMC Musculoskelet Disord. 8:115.

Karacić I et al. (2009). "Genotype-predicted tetrahydrobiopterin (BH4)-responsiveness and molecular genetics in Croatian patients with phenylalanine hydroxylase (PAH) deficiency." Mol Genet Metab. 97(3):165-71.

Karen A Hunt, et al (2008). Newly identified genetic risk variants for celiac disease related to the immune response. Nat Genet

Kathiresan, S et al (2009). Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. NAT GET

Kaul R et al. (1994). "Canavan disease: mutations among Jewish and non-Jewish patients." Am J Hum Genet. 55(1):34-41.

Kaul R et al. (1996). "Identification and expression of eight novel mutations among non-Jewish patients with Canavan disease." Am J Hum Genet. 59(1):95-102.

REFERENCES

Keller MA et al. (2014). "A gatekeeper helix determines the substrate specificity of Sjögren-Larsson Syndrome enzyme fatty aldehyde dehydrogenase." Nat Commun. 5:4439.

Kennedy S et al. (2010). "The first three years of screening for medium chain acyl-CoA dehydrogenase deficiency (MCADD) by newborn screening ontario." BMC Pediatr. 10:82.

Kenneson A et al. (2002). "GJB2 (connexin 26) variants and nonsyndromic sensorineural hearing loss: a HuGE review." Genet Med. 4(4):258-74.

Kikkawa YS et al. (2008). "Development of outer hair cells in Ames waltzer mice: mutation in protocadherin 15 affects development of cuticular plate and associated structures." Anat Rec (Hoboken). 291(2):224-32.

Kiryluk K, et al (2014). Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nat Genet.

Kishnani PS et al. (2014). "Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics." Genet Med. 16(11):e1.

Kivirikko S et al. (1996). "Mutational hotspots in the LAMB3 gene in the lethal (Herlitz) type of junctional epidermolysis bullosa." Hum Mol Genet. 5(2):231-7.

Kjaergaard S et al. (1998). "Absence of homozygosity for predominant mutations in PMM2 in Danish patients with carbohydrate-deficient glycoprotein syndrome type 1." Eur J Hum Genet. 6(4):331-6.

Kjaergaard S et al. (2001). "Congenital disorder of glycosylation type Ia (CDG-Ia): phenotypic spectrum of the R141H/F119L genotype." Arch Dis Child. 85(3):236-9.

Kleier S et al. (2000). "Clinical presentation and mutation identification in the NBS1 gene in a boy with Nijmegen breakage syndrome." Clin Genet. 57(5):384-7.

Klein et al. (2007) . "Deciphering the role of heterozygous mutations in genes associated with parkinsonism." 6(7):652-62. Lancet Neurol

Kleta R et al. (2003). "Biochemical and molecular analyses of infantile free sialic acid storage disease in North American children." Am J Med Genet A. 120A(1):28-33.

Kornum BR, et al(2011). Common variants in P2RY11 are associated with narcolepsy. Nat Genet.

Kotarsky H et al. (2010). "Characterization of complex III deficiency and liver dysfunction in GRACILE syndrome caused by a BCS1L mutation." Mitochondrion. 10(5):497-509.

Kotarsky H et al. (2012). "Metabolite profiles reveal energy failure and impaired beta-oxidation in liver of mice with complex III deficiency due to a BCS1L mutation." PLoS One. 7(7):e41156.

Köttgen A et al. (2013) . "Genome-wide association analyses identify 18 new loci associated with serum urate concentrations." 45(2):145-54. Nat. Genet.

Köttgen A, et al (2009). Multiple loci associated with indices of renal function and chronic kidney disease. Nat. Genet.

Kou I, et al (2013). Genetic variants in GPR126 are associated with adolescent idiopathic scoliosis. Nat Genet.

Kriegshäuser G et al. (2018). "Clinical and genetic heterogeneity in a large cohort of Armenian patients with late-onset familial Mediterranean fever." Genet Med.

Krüger L et al. (2007). "Cancer incidence in Nijmegen breakage syndrome is modulated by the amount of a variant NBS protein." Carcinogenesis. 28(1):107-11.

Kumar S et al. (2007). "Case report of HbC/beta(0)-thalassemia from India." Int J Lab Hematol. 29(5):381-5.

Kure S et al. (1998). "Molecular analysis of glycogen storage disease type Ib: identification of a prevalent mutation among Japanese patients and assignment of a putative glucose-6-phosphate translocase gene to chromosome 11." Biochem Biophys Res Commun. 248(2):426-31.

Kuwano R et al. (2006) Dynamin-binding protein gene on chromosome 10q is associated with late-onset Alzheimer's disease Hum. Mol. Genet. 15(13):2170-82

Kyrri AR et al. (2013). "The changing epidemiology of β-thalassemia in the Greek-Cypriot population." Hemoglobin. 37(5):435-43.

Lai K et al. (2017). "The prevalence of thalassemia in mainland China: evidence from epidemiological surveys." Sci Rep. 7(1):920.

Laimer M et al. (2010). "Herlitz junctional epidermolysis bullosa." Dermatol Clin. 28(1):55-60.

Landels EC et al. (1992). "Beta-hexosaminidase splice site mutation has a high frequency among non-Jewish Tay-Sachs disease carriers from the British Isles." J Med Genet. 29(8):563-7.

Lau LI, et al (2006) Association of the Y402H polymorphism in complement factor H gene and neovascular age-related macular degeneration in Chinese patients. Invest Ophthalmol Vis Sci.

lavinia Paternoster, et al (2011). Meta-analysis of genome-wide association studies identifies three new risk loci for atopic dermatitis Nat Genet.

Lee DH et al. (2004). "The molecular basis of phenylketonuria in Koreans." J Hum Genet. 49(11):617-21.

Lee YW et al. (2008). "Mutation analysis of PAH gene and characterization of a recurrent deletion mutation in Korean patients with phenylketonuria." Exp Mol Med. 40(5):533-40.

Lei KJ et al. (1993). "Mutations in the glucose-6-phosphatase gene that cause glycogen storage disease type 1a." Science. 262(5133):580-3.

Lei KJ et al. (1995). "Genetic basis of glycogen storage disease type 1a: prevalent mutations at the glucose-6-phosphatase locus." Am J Hum Genet. 57(4):766-71.

Lentz J et al. (1999). "Usher Syndrome Type I." [Updated 2016 May 19].

Lerer I et al. (2000). "Contribution of connexin 26 mutations to nonsyndromic deafness in Ashkenazi patients and the variable phenotypic effect of the mutation 167delT." Am J Med Genet. 95(1):53-6.

REFERENCES

Leuzzi V et al. (2006). "The spectrum of phenylalanine variations under tetrahydrobiopterin load in subjects affected by phenylalanine hydroxylase deficiency." J Inherit Metab Dis. 29(1):38-46.

Levéen P et al. (2011). "The GRACILE mutation introduced into Bcs1l causes postnatal complex III deficiency: a viable mouse model for mitochondrial hepatopathy." Hepatology. 53(2):437-47.

Li D, Zhao H, et al (2011). Strong protective effect of the aldehyde dehydrogenase gene (ALDH2) 504lys (*2) allele against alcoholism and alcohol-induced medical diseases in Asians. Hum Genet

Li Jia Chen, et al (2006) Association of complement factor H polymorphisms with exudative age-related macular degeneration. Molecular Vision.

Li N et al. (2015). "Molecular characterisation of phenylketonuria in a Chinese mainland population using next-generation sequencing." Sci Rep. 5:15769.

Li Su, et al (2008). Association of GWAS-Supported Variants rs2200733 and rs6843082 on Chromosome 4q25 with Ischemic Stroke in the Southern Chinese Han Population. J Mol Neurosci.

Liang Y et al. (2014). "The mutation spectrum of the phenylalanine hydroxylase (PAH) gene and associated haplotypes reveal ethnic heterogeneity in the Taiwanese population." J Hum Genet. 59(3):145-52.

Liao J, et al (2014). Meta-analysis of genome-wide association studies in multiethnic Asians identifies two loci for age-related nuclear cataract. Hum Mol Genet

Lie-Injo LE et al. (1989). "Beta-thalassemia mutations in Indonesia and their linkage to beta haplotypes." Am J Hum Genet. 45(6):971-5.

Lijuan Wang, et al (2014). Association between single-nucleotide polymorphisms in six hypertensive candidate genes and hypertension among northern Han Chinese individuals. Hypertension Research.

Lim LE et al. (1995). "Beta-sarcoglycan: characterization and role in limb-girdle muscular dystrophy linked to 4q12." Nat Genet. 11(3):257-65.

Lin LI et al. (1991). "The spectrum of beta-thalassemia mutations in Taiwan: identification of a novel frameshift mutation." Am J Hum Genet. 48(4):809-12.

Liu JZ et al. (1989). "Studies of beta-thalassemia mutations in families living in three provinces in southern China." Hemoglobin. 13(6):585-95.

Liu M, et al(2014). Apolipoprotein E gene polymorphism and Alzheimer's disease in Chinese population: a meta-analysis. Sci Rep.

Liu X, et al (2009). The functional variant rs1048990 in PSMA6 is associated with susceptibility to myocardial infarction in a Chinese population. Atherosclerosis

Liu XZ et al. (2002). "The prevalence of connexin 26 (GJB2) mutations in the Chinese population." Hum Genet. 111(4-5):394-7.

Liu Y, et al (2014). Genetic polymorphisms in ZFHX3 are associated with atrial fibrillation in a Chinese Han population. PLoS One.

Liu YH, et al (2014). Gene polymorphisms associated with susceptibility to coronary artery disease in Han Chinese people. Genet Mol Res.

Longinotti M et al. (1994). "A 12-year preventive program for beta-thalassemia in Northern Sardinia." Clin Genet. 46(3):238-43.

López-Bigas N et al. (2002). "Erratum: Identification of five new mutations of PDS/SLC26A4 in Mediterranean families with hearing impairment." Hum Mutat. 20(1):77-8.

Louise V Wain, et al (2011). Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics.

Lövkvist H1, et al (2008). Ischaemic stroke in hypertensive patients is associated with variations in the PDE4D genome region. European Journal of Human Genetics.

Lu PJ et al. (2010). "Mutations alter secretion of fukutin-related protein." Biochim Biophys Acta. 1802(2):253-8.

Lu WH, et al (2015). Association between aldosterone synthase (CYP11B2) -344C/T polymorphism and atrial fibrillation among Han and Kazak residents of the Xinjiang region. Int J Clin Exp Med.

Lyly A et al. (2009). "Novel interactions of CLN5 support molecular networking between Neuronal Ceroid Lipofuscinosis proteins." BMC Cell Biol. 10:83.

M. G. Heckmana, et al (2013). Genetic variants associated with myocardial infarction in the PSMA6 gene and Chr9p21 are also associated with ischaemic stroke. European Journal of Neurology.

Maier EM et al. (2005). "Population spectrum of ACADM genotypes correlated to biochemical phenotypes in newborn screening for medium-chain acyl-CoA dehydrogenase deficiency." Hum Mutat. 25(5):443-52.

Makhoul NJ et al. (2005). "Genetic heterogeneity of Beta thalassemia in Lebanon reflects historic and recent population migration." Ann Hum Genet. 69(Pt 1):55-66.

Martin DC et al. (2007). "Evaluation of the risk for Tay-Sachs disease in individuals of French Canadian ancestry living in new England." Clin Chem. 53(3):392-8.

Martin RA et al. (2003). "Sialic acid storage disease of the Salla phenotype in American monozygous twin female sibs." Am J Med Genet A. 120A(1):23-7.

Marvit J et al. (1987). "GT to AT transition at a splice donor site causes skipping of the preceding exon in phenylketonuria." Nucleic Acids Res. 15(14):5613-28.

Masmoudi S et al. (2000). "Pendred syndrome: phenotypic variability in two families carrying the same PDS missense mutation." Am J Med Genet. 90(1):38-44.

Matalon R et al. (1999). "Canavan Disease." [Updated 2011 Aug 11].

Matern D et al. (2000). "Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency." [Updated 2015 Mar 5].

Matsubara Y et al. (1990). "Identification of a common mutation in patients with medium-chain acyl-CoA dehydrogenase deficiency." Biochem Biophys Res Commun. 171(1):498-505.

REFERENCES

Matsuo H et al. (2013) . "Common dysfunctional variants in ABCG2 are a major cause of early-onset gout." 3:2014. Sci Rep

Matsuoka R, et al (2015). Association of six genetic variants with myocardial infarction. Int J Mol Med

Matthijs G et al. (2000). "Mutations in PMM2 that cause congenital disorders of glycosylation, type Ia (CDG-Ia)." Hum Mutat. 16(5):386-94.

Maurer MH et al. (2010). "High prevalence of the NBN gene mutation c.657-661del5 in Southeast Germany." J Appl Genet. 51(2):211-4.

Maxwell MA et al. (1999). "A common PEX1 frameshift mutation in patients with disorders of peroxisome biogenesis correlates with the severe Zellweger syndrome phenotype." Hum Genet. 105(1-2):38-44.

McDowell GA et al. (1992). "The presence of two different infantile Tay-Sachs disease mutations in a Cajun population." Am J Hum Genet. 51(5):1071-7.

Mega, Jessica L. Stitziel, Nathan O. Smith, J. Gustav, et al (2015). Genetic risk, coronary heart disease events, and the clinical benefit of statin therapy: an analysis of primary and secondary prevention trials. The Lancet.

Mehta PA et al. (2002). "Fanconi Anemia." [Updated 2017 Feb 23].

Melanie Kolz , et al (2009). Meta-Analysis of 28,141 Individuals Identifies Common Variants within Five New Loci That Influence Uric Acid Concentrations. PLoS Genetics

Melis D et al. (2005). "Genotype/phenotype correlation in glycogen storage disease type 1b: a multicentre study and review of the literature." Eur J Pediatr. 164(8):501-8.

Mercier J et al. (2001). "Rapid detection of the sacsin mutations causing autosomal recessive spastic ataxia of Charlevoix-Saguenay." Genet Test. 5(3):255-9.

Miller JN et al. (2015). "The novel Cln1(R151X) mouse model of infantile neuronal ceroid lipofuscinosis (INCL) for testing nonsense suppression therapy." Hum Mol Genet. 24(1):185-96.

Mitchell P, et al (1995) Prevalence of Age--related Maculopathy in Australia The Blue Mountains Eye Study. Ophthalmology.

Mitchison HM et al. (1998). "Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits." Hum Mol Genet. 7(2):291-7.

Miyagawa M et al. (2014). "Mutation spectrum and genotype-phenotype correlation of hearing loss patients caused by SLC26A4 mutations in the Japanese: a large cohort study." J Hum Genet. 59(5):262-8.

Miyake Y, et al (2012). SNCA polymorphisms, smoking, and sporadic Parkinson's disease in Japanese. Parkinsonism Relat Disord

Miyake, et al (2008). Association of TCF7L2 polymorphisms with susceptibility to type 2 diabetes in 4,087 Japanese subjects. J Hum Genet

Modell B et al. (2008). "Global epidemiology of haemoglobin disorders and derived service indicators." Bull World Health Organ. 86(6):480-7.

Moffatt MF , et al (2007). Genetic variants regulating ORMDL3 expression contribute to the risk of childhood asthma. Nature.

Moharir A et al. (2013). "The role of N-glycosylation in folding, trafficking, and functionality of lysosomal protein CLN5." PLoS One. 8(9):e74299.

Mole SE et al. (2001). "Neuronal Ceroid-Lipofuscinoses." [Updated 2013 Aug 1].

Mole SE et al. (2001). "Neuronal Ceroid-Lipofuscinoses." [Updated 2013 Aug 1].

Mole SE et al. (2005). "Correlations between genotype, ultrastructural morphology and clinical phenotype in the neuronal ceroid lipofuscinoses." Neurogenetics. 6(3):107-26.

Monaghan KG et al. (2008). "Technical standards and guidelines for reproductive screening in the Ashkenazi Jewish population." Genet Med. 10(1):57-72.

Monaghan KG et al. (2008). "Technical standards and guidelines for reproductive screening in the Ashkenazi Jewish population." Genet Med. 10(1):57-72.

Montfort M et al. (2004). "Functional analysis of 13 GBA mutant alleles identified in Gaucher disease patients: Pathogenic changes and "modifier" polymorphisms." Hum Mutat. 23(6):567-75.

Mootha VK et al. (2003). "Identification of a gene causing human cytochrome c oxidase deficiency by integrative genomics." Proc Natl Acad Sci U S A. 100(2):605-10.

Morell RJ et al. (1998). "Mutations in the connexin 26 gene (GJB2) among Ashkenazi Jews with nonsyndromic recessive deafness." N Engl J Med. 339(21):1500-5.

Morin C et al. (1993). "Clinical, metabolic, and genetic aspects of cytochrome C oxidase deficiency in Saguenay-Lac-Saint-Jean." Am J Hum Genet. 53(2):488-96.

Morin P et al. (2004). "Functional characterization of wild-type and mutant human sialin." EMBO J. 23(23):4560-70.

Motley AM et al. (2002). "Mutational spectrum in the PEX7 gene and functional analysis of mutant alleles in 78 patients with rhizomelic chondrodysplasia punctata type 1." Am J Hum Genet. 70(3):612-24.

Mühle C et al. (2005). "Novel and recurrent mutations in the laminin-5 genes causing lethal junctional epidermolysis bullosa: molecular basis and clinical course of Herlitz disease." Hum Genet. 116(1-2):33-42.

Mühleisen TW, et al (2014). Genome-wide association study reveals two new risk loci for bipolar disorder. Nat Commun.

Myouzen K, et al(2012). Functional variants in NFKBIE and RTKN2 involved in activation of the NF-κ B pathway are associated with rheumatoid arthritis in Japanese. PLoS Genetics

Nakagawa S et al. (2012). "Platelet hexosaminidase a enzyme assay effectively detects carriers missed by targeted DNA mutation analysis." JIMD Rep. 6:1-6.

REFERENCES

Nakano A et al. (2000). "Herlitz junctional epidermolysis bullosa: novel and recurrent mutations in the LAMB3 gene and the population carrier frequency." J Invest Dermatol. 115(3):493-8.

Nakashima M, et al (2010). A genome-wide association study identifies four susceptibility loci for keloid in the Japanese population. Nat Genet.

Namba A et al. (2001). "Genetic features of hearing loss associated with ear anomalies: PDS and EYA1 mutation analysis." J Hum Genet. 46(9):518-21.

Nellis MM et al. (2003). "Relationship of causative genetic mutations in maple syrup urine disease with their clinical expression." Mol Genet Metab. 80(1-2):189-95.

Ness SL et al. (2003). "Genetic homogeneity and phenotypic variability among Ashkenazi Jews with Usher syndrome type III." J Med Genet. 40(10):767-72.

Nestorowicz A et al. (1996). "Mutations in the sulonylurea receptor gene are associated with familial hyperinsulinism in Ashkenazi Jews." Hum Mol Genet. 5(11):1813-22.

Neu-Yilik G et al. (2011). "Mechanism of escape from nonsense-mediated mRNA decay of human beta-globin transcripts with nonsense mutations in the first exon." RNA. 17(5):843-54.

Ober C, et al (2008). Effect of variation in CHI3L1 on serum YKL-40 level, risk of asthma, and lung function. N Engl J Med.

Ohara T et al. (2012) Association study of susceptibility genes for late-onset Alzheimer's disease in the Japanese population. Psychiatr. Genet.

Okada Y, et al (2012). Meta-analysis identifies nine new loci associated with rheumatoid arthritis in the Japanese population. Nat Genet.

Okamoto K, et al (2011). Common variation in GPC5 is associated with acquired nephrotic syndrome. Nat Genet.

Okano Y et al. (1991). "Molecular basis of phenotypic heterogeneity in phenylketonuria." N Engl J Med. 324(18):1232-8.

Oner R et al. (1990). "Beta-thalassemia in Turkey." Hemoglobin. 14(1):1-13.

Ong T et al. (2001). "Cystic Fibrosis and Congenital Absence of the Vas Deferens." [Updated 2017 Feb 2].

O'Reilly L et al. (2004). "The Y42H mutation in medium-chain acyl-CoA dehydrogenase, which is prevalent in babies identified by MS/MS-based newborn screening, is temperature sensitive." Eur J Biochem. 271(20):4053-63.

Origa R et al. (2000). "Beta-Thalassemia." [Updated 2015 May 14].

Otonkoski T et al. (1999). "A point mutation inactivating the sulfonylurea receptor causes the severe form of persistent hyperinsulinemic hypoglycemia of infancy in Finland." Diabetes. 48(2):408-15.

Ozdemir O et al. (2011). "Prevalence of known mutations in the MEFV gene in a population screening with high rate of carriers." Mol Biol Rep. 38(5):3195-200.

Padeh S et al. (2003). "Clinical and diagnostic value of genetic testing in 216 Israeli children with Familial Mediterranean fever." J Rheumatol. 30(1):185-90.

Padmanabhan S, et al (2010). Genome-Wide Association Study of Blood Pressure Extremes Identifies Variant near UMOD Associated with Hypertension. PLoS Genet.

Park HY, et al (2015). Utilizing Genetic Predisposition Score in Predicting Risk of Type 2 Diabetes Mellitus Incidence: A Community-based Cohort Study on Middle-aged Koreans. J Korean Med Sci.

Pasa S et al. (2008). "Familial Mediterranean fever gene mutations in the Southeastern region of Turkey and their phenotypical features." Amyloid. 15(1):49-53.

Pasmooij AM et al. (2007). "Revertant mosaicism in junctional epidermolysis bullosa due to multiple correcting second-site mutations in LAMB3." J Clin Invest. 117(5):1240-8.

Pastores GM et al. (2000). "Gaucher Disease." [Updated 2015 Feb 26].

Pászty C et al. (1997). "Transgenic knockout mice with exclusively human sickle hemoglobin and sickle cell disease." Science. 278(5339):876-8.

Patrick T Ellinor, et al (2012). Meta-analysis identifies six new susceptibility loci for atrial fibrillation Nat Genet.

Pawloski JR et al. (2005). "Impaired vasodilation by red blood cells in sickle cell disease." Proc Natl Acad Sci U S A. 102(7):2531-6.

Pegoraro E et al. (2000). "Limb-Girdle Muscular Dystrophy Overview." [Updated 2012 Aug 30].

Peng WH, et al (2009). Chromosome 9p21 polymorphism is associated with myocardial infarction but not with clinical outcome in Han Chinese. Clin Chem Lab Med.

Pey AL et al. (2003). "Phenylketonuria: genotype-phenotype correlations based on expression analysis of structural and functional mutations in PAH." Hum Mutat. 21(4):370-8.

Pfendner EG et al. (2008). "Junctional Epidermolysis Bullosa." [Updated 2014 Jan 2].

Philipp S. Wild, Tanja Zeller, Arne Schillert, et al (2011). A Genome-Wide Association Study Identifies LIPA as a Susceptibility Gene for Coronary Artery Disease. Circulation Cardiovascular Genetics.

Ploos van Amstel JK et al. (1996). "Hereditary tyrosinemia type 1: novel missense, nonsense and splice consensus mutations in the human fumarylacetoacetate hydrolase gene; variability of the genotype-phenotype relationship." Hum Genet. 97(1):51-9.

Posteraro P et al. (2004). "Laminin-5 mutational analysis in an Italian cohort of patients with junctional epidermolysis bullosa." J Invest Dermatol. 123(4):639-48.

Pourová R et al. (2010). "Spectrum and frequency of SLC26A4 mutations among Czech patients with early hearing loss with and without Enlarged Vestibular Aqueduct (EVA)." Ann Hum Genet. 74(4):299-307.

Powars DR et al. (2005). "Outcome of sickle cell anemia: a 4-decade observational study of 1056 patients." Medicine (Baltimore). 84(6):363-76.

REFERENCES

Preuss M et al. (2010) Design of the Coronary ARtery DIsease Genome-Wide Replication And Meta-Analysis (CARDIoGRAM) Study: A Genome-wide association meta-analysis involving more than 22 000 cases and 60 000 controls. Circ Cardiovasc Genet

Puckett RL et al. (2010). "Maple syrup urine disease: further evidence that newborn screening may fail to identify variant forms." Mol Genet Metab. 100(2):136-42.

Pulkkinen L et al. (1994). "A homozygous nonsense mutation in the beta 3 chain gene of laminin 5 (LAMB3) in Herlitz junctional epidermolysis bullosa." Genomics. 24(2):357-60.

Pulkkinen L et al. (1997). "Predominance of the recurrent mutation R635X in the LAMB3 gene in European patients with Herlitz junctional epidermolysis bullosa has implications for mutation detection strategy." J Invest Dermatol. 109(2):232-7.

Qin YM et al. (2000). "Human peroxisomal multifunctional enzyme type 2. Site-directed mutagenesis studies show the importance of two protic residues for 2-enoyl-CoA hydratase 2 activity." J Biol Chem. 275(7):4965-72.

Quan C , et al (2010). Genome-wide association study for vitiligo identifies susceptibility loci at 6q27 and the MHC. Nat Genet.

Quinonez SC et al. (2014). "Dihydrolipoamide Dehydrogenase Deficiency."

Ramasamy A, et al (2011). A genome-wide meta-analysis of genetic variants associated with allergic rhinitis and grass sensitization and their interaction with birth order. J Allergy Clin Immunol

Rebibo-Sabbah A et al. (2007). "In vitro and ex vivo suppression by aminoglycosides of PCDH15 nonsense mutations underlying type 1 Usher syndrome." Hum Genet. 122(3-4):373-81.

Regier DS et al. (2000). "Phenylalanine Hydroxylase Deficiency." [Updated 2017 Jan 5].

Rellos P et al. (2000). "Expression, purification, and characterization of natural mutants of human aldolase B. Role of quaternary structure in catalysis." J Biol Chem. 275(2):1145-51.

Resnick IB et al. (2002). "Nijmegen breakage syndrome: clinical characteristics and mutation analysis in eight unrelated Russian families." J Pediatr. 140(3):355-61.

Rhead WJ. (2006). "Newborn screening for medium-chain acyl-CoA dehydrogenase deficiency: a global perspective." J Inherit Metab Dis. 29(2-3):370-7.

Rivera A, et al (2005) Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. Hum Mol Genet.

Rivera I et al. (2011). "Phenylalanine hydroxylase deficiency: molecular epidemiology and predictable BH4-responsiveness in South Portugal PKU patients." Mol Genet Metab. 104 Suppl:S86-92.

Rizzo WB et al. (1999). "The molecular basis of Sjögren-Larsson syndrome: mutation analysis of the fatty aldehyde dehydrogenase gene." Am J Hum Genet. 65(6):1547-60.

Rizzo WB et al. (2008). "Abnormal fatty alcohol metabolism in cultured keratinocytes from patients with Sjögren-Larsson syndrome." J Lipid Res. 49(2):410-9.

Romano A et al. (2013). "Comparative analysis and functional mapping of SACS mutations reveal novel insights into sacsin repeated architecture." Hum Mutat. 34(3):525-37.

Rootwelt H et al. (1994). "Novel splice, missense, and nonsense mutations in the fumarylacetoacetase gene causing tyrosinemia type 1." Am J Hum Genet. 55(4):653-8.

Rootwelt H et al. (1996). "Fumarylacetoacetase mutations in tyrosinaemia type I." Hum Mutat. 7(3):239-43.

Rossetti S et al. (2003). "A complete mutation screen of PKHD1 in autosomal-recessive polycystic kidney disease (ARPKD) pedigrees." Kidney Int. 64(2):391-403.

Rui Li , et al (2015). A meta-analysis of the associations between the Q141K and Q126X ABCG2 gene variants and gout risk. Int J Clin Exp Pathol

Rund D et al. (1991). "Evolution of a genetic disease in an ethnic isolate: beta-thalassemia in the Jews of Kurdistan." Proc Natl Acad Sci U S A. 88(1):310-4.

Sabbagh AS et al. (2008). "MEFV gene mutations spectrum among Lebanese patients referred for Familial Mediterranean Fever work-up: experience of a major tertiary care center." Mol Biol Rep. 35(3):447-51.

Salehzadeh F. (2015). "Familial Mediterranean Fever in Iran: A Report from FMF Registration Center." Int J Rheumatol. 2015:912137.

Salin-Cantegrel A et al. (2011). "Transit defect of potassium-chloride Co-transporter 3 is a major pathogenic mechanism in hereditary motor and sensory neuropathy with agenesis of the corpus callosum." J Biol Chem. 286(32):28456-65.

Samani NJ, et al (2007). Genomewide association analysis of coronary artery disease. N Engl J Med.

Sánchez-Gutiérrez JC et al. (2002). "Molecular analysis of the aldolase B gene in patients with hereditary fructose intolerance from Spain." J Med Genet. 39(9):e56.

Sansaricq C et al. (2006). "Biochemical and molecular diagnosis of lipoamide dehydrogenase deficiency in a North American Ashkenazi Jewish family." J Inherit Metab Dis. 29(1):203-4.

Santamaria R et al. (1996). "Molecular basis of hereditary fructose intolerance in Italy: identification of two novel mutations in the aldolase B gene." J Med Genet. 33(9):786-8.

Santer R et al. (2000). "Molecular analysis in glycogen storage disease 1 non-A: DHPLC detection of the highly prevalent exon 8 mutations of the G6PT1 gene in German patients." Hum Mutat. 16(2):177.

Santer R et al. (2005). "The spectrum of aldolase B (ALDOB) mutations and the prevalence of hereditary fructose intolerance in Central Europe." Hum Mutat. 25(6):594.

Santos LL et al. (2010). "Variations in genotype-phenotype correlations in phenylketonuria patients." Genet Mol Res. 9(1):1-8.

Sanz MM et al. (2006). "Bloom's Syndrome." [Updated 2016 Apr 7].

REFERENCES

Sarwar N, Sandhu MS, Ricketts SL, Butterworth AS, et al (2010). Triglyceride-mediated pathways and coronary disease:collaborative analysis of 101 studies. The Lancet.

Sasarman F et al. (2010). "LRPPRC and SLIRP interact in a ribonucleoprotein complex that regulates posttranscriptional gene expression in mitochondria." Mol Biol Cell. 21(8):1315-23.

Sasarman F et al. (2015). "Tissue-specific responses to the LRPPRC founder mutation in French Canadian Leigh Syndrome." Hum Mol Genet. 24(2):480-91.

Sato E et al. (2001). "Phenotypes associated with replacement of His by Arg in the Pendred syndrome gene." Eur J Endocrinol. 145(6):697-703.

Savukoski M et al. (1998). "CLN5, a novel gene encoding a putative transmembrane protein mutated in Finnish variant late infantile neuronal ceroid lipofuscinosis." Nat Genet. 19(3):286-8.

Schapira (2006) . "The importance of LRRK2 mutations in Parkinson disease." 63(9):1225-8. Arch Neurol

Schiffmann R et al. (2005). "Mucolipidosis IV." [Updated 2015 Jul 30].

Schmidt S, et al (2006) Cigarette smoking strongly modifies the association of LOC387715 and age-related macular degeneration. Am J Hum Genet.

Schollen E et al. (2000). "Lack of Hardy-Weinberg equilibrium for the most prevalent PMM2 mutation in CDG-Ia (congenital disorders of glycosylation type Ia)." Eur J Hum Genet. 8(5):367-71.

Schulze, et al (2008). Two variants in Ankyrin 3 (ANK3) are independent genetic risk factors for bipolar disorder. Mol Psychiatry

Scott DA et al. (2000). "Functional differences of the PDS gene product are associated with phenotypic variation in patients with Pendred syndrome and non-syndromic hearing loss (DFNB4)." Hum Mol Genet. 9(11):1709-15.

Scott SA et al. (2010). "Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases." Hum Mutat. 31(11):1240-50.

Seaver LH et al. (2009). "ACMG practice guideline: genetic evaluation of short stature." Genet Med. 11(6):465-70.

Sebastio G et al. (1991). "Aldolase B mutations in Italian families affected by hereditary fructose intolerance." J Med Genet. 28(4):241-3.

Seydewitz HH et al. (2000). "Molecular genetic analysis of 40 patients with glycogen storage disease type Ia: 100% mutation detection rate and 5 novel mutations." Hum Mutat. 15(1):115-6.

Shaag A et al. (1995). "The molecular basis of canavan (aspartoacylase deficiency) disease in European non-Jewish patients." Am J Hum Genet. 57(3):572-80.

Shaag A et al. (1999). "Molecular basis of lipoamide dehydrogenase deficiency in Ashkenazi Jews." Am J Med Genet. 82(2):177-82.

Sharp AM et al. (2005). "Comprehensive genomic analysis of PKHD1 mutations in ARPKD cohorts." J Med Genet. 42(4):336-49.

Shekarabi M et al. (2012). "Loss of neuronal potassium/chloride cotransporter 3 (KCC3) is responsible for the degenerative phenotype in a conditional mouse model of hereditary motor and sensory neuropathy associated with agenesis of the corpus callosum." J Neurosci. 32(11):3865-76.

Shohat M et al. (2000). "Familial Mediterranean Fever." [Updated 2016 Dec 15].

Shohat M et al. (2003). "Familial Dysautonomia." [Updated 2014 Dec 18].

Sistermans EA et al. (2000). "Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population." Eur J Hum Genet. 8(7):557-60.

Sleat DE et al. (2016). "Analysis of large-scale whole exome sequencing data to determine the prevalence of genetically-distinct forms of neuronal ceroid lipofuscinosis." Gene. 593(2):284-91.

Smith EH et al. (2010). "Allelic diversity in MCAD deficiency: the biochemical classification of 54 variants identified during 5 years of ACADM sequencing." Mol Genet Metab. 100(3):241-50.

Smith RJH (1998). "Pendred Syndrome/Nonsyndromic Enlarged Vestibular Aqueduct." [Updated 2017 Oct 19].

Smith RJH et al. (1998). "Nonsyndromic Hearing Loss and Deafness, DFNB1." [Updated 2016 Aug 18].

Sniderman King L et al. (2006). "Tyrosinemia Type I." [Updated 2017 May 25].

Song F et al. (2005). "Phenylketonuria mutations in Northern China." Mol Genet Metab. 86 Suppl 1:S107-18.

So-Young Bang, et al (2010). Research article Peptidyl arginine deiminase type IV (PADI4) haplotypes interact with shared epitope regardless of anti-cyclic citrullinated peptide antibody or erosive joint status in rheumatoid arthritis: a case control study. Arthritis Research & Therapy.

Sozuoz A et al. (1988). "Beta thalassaemia mutations in Turkish Cypriots." J Med Genet. 25(11):766-8.

Spaapen LJ et al. (2003). "Tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency, state of the art." Mol Genet Metab. 78(2):93-9.

Sparks SE et al. (2005). "PMM2-CDG (CDG-Ia)." [Updated 2015 Oct 29].

Sreekumar G. Pillai, et al (2009). A Genome-Wide Association Study in Chronic Obstructive Pulmonary Disease (COPD): Identification of Two Major Susceptibility Loci. PLoS Genetics.

Srinivasan BS et al. (2010). "A universal carrier test for the long tail of Mendelian disease." Reprod Biomed Online. 21(4):537-51.

Stefansson, et al (2007). A genetic risk factor for periodic limb movements in sleep. N Engl J Med.

Steinberg S et al. (2004). "The PEX Gene Screen: molecular diagnosis of peroxisome biogenesis disorders in the Zellweger syndrome spectrum." Mol Genet Metab. 83(3):252-63.

REFERENCES

Steinberg SJ et al. (2003). "Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum." [Updated 2012 May 10].

Stender S, et al (2011). Sterol transporter adenosine triphosphate-binding cassette transporter G8, gallstones, and biliary cancer in 62,000 individuals from the general population. Hepatology

St-Louis M et al. (1994). "Identification of a stop mutation in five Finnish patients suffering from hereditary tyrosinemia type I." Hum Mol Genet. 3(1):69-72.

Stopa JD et al. (2011). "Stabilization of the predominant disease-causing aldolase variant (A149P) with zwitterionic osmolytes." Biochemistry. 50(5):663-71.

Strauss KA et al. (2005). "Genome-wide SNP arrays as a diagnostic tool: clinical description, genetic mapping, and molecular characterization of Salla disease in an Old Order Mennonite population." Am J Med Genet A. 138A(3):262-7.

Strauss KA et al. (2006). "Maple Syrup Urine Disease." [Updated 2013 May 9].

Stroppiano M et al. (1999). "Mutations in the glucose-6-phosphatase gene of 53 Italian patients with glycogen storage disease type Ia." J Inherit Metab Dis. 22(1):43-9.

Su S, et al (2013). The associations between single nucleotide polymorphisms of DNA repair genes, DNA damage, and age-related cataract: Jiangsu Eye Study. Invest Ophthalmol Vis Sci.

Sulem P et al. (2011) . "Identification of low-frequency variants associated with gout and serum uric acid levels." 43(11):1127-30. Nat. Genet.

Sun LD, et al (2011). Genome-wide association study identifies two new susceptibility loci for atopic dermatitis in the Chinese Han population. Nat Genet.

Sveen ML et al. (2006). "High prevalence and phenotype-genotype correlations of limb girdle muscular dystrophy type 2I in Denmark." Ann Neurol. 59(5):808-15.

Svensson E et al. (1993). "Relation between genotype and phenotype in Swedish phenylketonuria and hyperphenylalaninemia patients." Eur J Pediatr. 152(2):132-9.

Sweeney WE et al. (2001). "Polycystic Kidney Disease, Autosomal Recessive." [Updated 2016 Sep 15].

Takahashi Y, et al (2011). A genome-wide association study identifies common variants near LBX1 associated with adolescent idiopathic scoliosis. Nat Genet.

Tamagnini GP et al. (1993). "Beta-thalassemia mutations in the Portuguese; high frequencies of two alleles in restricted populations." Hemoglobin. 17(1):31-40.

Tan EK, et al (2010). Multiple LRRK2 variants modulate risk of Parkinson disease: a Chinese multicenter study. Hum Mutat

Tanaka K et al. (1997). "A survey of the newborn populations in Belgium, Germany, Poland, Czech Republic, Hungary, Bulgaria, Spain, Turkey, and Japan for the G985 variant allele with haplotype analysis at the medium chain Acyl-CoA dehydrogenase gene locus: clinical and evolutionary consideration." Pediatr Res. 41(2):201-9.

Tang W et al. (2015). "Spectrum of α-thalassemia and β-thalassemia mutations in the Guilin Region of southern China." Clin Biochem. 48(16-17):1068-72.

Taniguchi M et al. (2015). "Carrier frequency of the GJB2 mutations that cause hereditary hearing loss in the Japanese population." J Hum Genet.

Taylor JP et al. (2002). "Mutations of the PDS gene, encoding pendrin, are associated with protein mislocalization and loss of iodide efflux: implications for thyroid dysfunction in Pendred syndrome." J Clin Endocrinol Metab. 87(4):1778-84.

Tchernitchko D et al. (2003). "The E148Q MEFV allele is not implicated in the development of familial Mediterranean fever." Hum Mutat. 22(4):339-40.

The International Multiple Sclerosis Genetics Consortium (2007). Risk Alleles for Multiple Sclerosis Identified by a Genomewide Study. Nat Genet.

Theodoridou S et al. (2008). "Carrier screening and prenatal diagnosis of hemoglobinopathies. A study of indigenous and immigrant couples in northern Greece, over the last 5 years." Hemoglobin. 32(5):434-9.

Thies W et al. (2013) Alzheimer's disease facts and figures. Alzheimers Dement

Thomas PM et al. (1995). "Mutations in the sulfonylurea receptor gene in familial persistent hyperinsulinemic hypoglycemia of infancy." Science. 268(5209):426-9.

Thorleifsson, et al (2007). Common sequence variants in the LOXL1 gene confer susceptibility to exfoliation glaucoma. Science

Tian G et al. (2009). "Clarin-1, encoded by the Usher Syndrome III causative gene, forms a membranous microdomain: possible role of clarin-1 in organizing the actin cytoskeleton." J Biol Chem. 284(28):18980-93.

Tianxiao Li , et al (2016). Lack of association between RAD50-IL13 polymorphisms and pediatric asthma susceptibility in Northeastern Han Chinese. J Asthma.

Torralba MA et al. (2002). "High prevalence of the 55-bp deletion (c.1263del55) in exon 9 of the glucocerebrosidase gene causing misdiagnosis (for homozygous N370S (c.1226A > G) mutation) in Spanish Gaucher disease patients." Blood Cells Mol Dis. 29(1):35-40.

Touitou I. (2001). "The spectrum of Familial Mediterranean Fever (FMF) mutations." Eur J Hum Genet. 9(7):473-83.

Treisman R et al. (1983). "Specific transcription and RNA splicing defects in five cloned beta-thalassaemia genes." Nature. 302(5909):591-6.

Tsuchida S et al. (2012). "Hydratase activities of green fluorescent protein tagged human multifunctional enzyme type 2 hydratase domain and its variants." J Oleo Sci. 61(8):443-50.

Tsukada K et al. (2015). "Ethnic-specific spectrum of GJB2 and SLC26A4 mutations: their origin and a literature review." Ann Otol Rhinol Laryngol. 124 Suppl 1:61S-76S.

Tuo J, et al (2008) The HtrA1 promoter polymorphism, smoking, and age-related macular degeneration in multiple case-control samples. Ophthalmology.

REFERENCES

Turhan A et al. (2002). "Primary role for adherent leukocytes in sickle cell vascular occlusion: a new paradigm." Proc Natl Acad Sci U S A. 99(5):3047-51.

Tyfield LA et al. (1995). "Discordant phenylketonuria phenotypes in one family: the relationship between genotype and clinical outcome is a function of multiple effects." J Med Genet. 32(11):867-70.

Urabe Y, et al (2012). A genome-wide association study of nephrolithiasis in the Japanese population identifies novel susceptible Loci at 5q35.3, 7p14.3, and 13q14.1. PLoS Genet.

Van Grunsven EG et al. (1998). "Peroxisomal D-hydroxyacyl-CoA dehydrogenase deficiency: resolution of the enzyme defect and its molecular basis in bifunctional protein deficiency." Proc Natl Acad Sci U S A. 95(5):2128-33.

Van Grunsven EG et al. (1999). "Enoyl-CoA hydratase deficiency: identification of a new type of D-bifunctional protein deficiency." Hum Mol Genet. 8(8):1509-16.

Van Grunsven EG et al. (1999). "Peroxisomal bifunctional protein deficiency revisited: resolution of its true enzymatic and molecular basis." Am J Hum Genet. 64(1):99-107.

Van Hauwe P et al. (1998). "Two frequent missense mutations in Pendred syndrome." Hum Mol Genet. 7(7):1099-104.

van Heel, et al (2007). A genome-wide association study for celiac disease identifies risk variants in the region harboring IL2 and IL21. Nat Genet

Vanderver A et al. (2014). "Leukodystrophy Overview."

Varki R et al. (2006). "Epidermolysis bullosa. I. Molecular genetics of the junctional and hemidesmosomal variants." J Med Genet. 43(8):641-52.

Varon R et al. (1999). "Nijmegen Breakage Syndrome." [Updated 2017 Feb 2].

Varon R et al. (2000). "Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations." Eur J Hum Genet. 8(11):900-2.

Vega AI et al. (2011). "Expression analysis revealing destabilizing mutations in phosphomannomutase 2 deficiency (PMM2-CDG): expression analysis of PMM2-CDG mutations." J Inherit Metab Dis. 34(4):929-39.

Verlander PC et al. (1994). "Mutation analysis of the Fanconi anemia gene FACC." Am J Hum Genet. 54(4):595-601.

Vermeer S et al. (2003). "ARSACS." [Updated 2012 Oct 11].

Vesa J et al. (1995). "Mutations in the palmitoyl protein thioesterase gene causing infantile neuronal ceroid lipofuscinosis." Nature. 376(6541):584-7.

Vichinsky EP et al. (2005). "Changes in the epidemiology of thalassemia in North America: a new minority disease." Pediatrics. 116(6):e818-25.

Vidaud M et al. (1989). "A 5′ splice-region G----C mutation in exon 1 of the human beta-globin gene inhibits pre-mRNA splicing: a mechanism for beta+-thalassemia." Proc Natl Acad Sci U S A. 86(3):1041-5.

Villard E, et al (2011). A genome-wide association study identifies two loci associated with heart failure due to dilated cardiomyopathy. Eur Heart J.

Visapää I et al. (2002). "GRACILE syndrome, a lethal metabolic disorder with iron overload, is caused by a point mutation in BCS1L." Am J Hum Genet. 71(4):863-76.

Vithana EN, et al(2012). Genome-wide association analyses identify three new susceptibility loci for primary angle closure glaucoma. Nat Genet.

Vockley J et al. (2014). "Phenylalanine hydroxylase deficiency: diagnosis and management guideline." Genet Med. 16(2):188-200.

Waddell L et al. (2006). "Medium-chain acyl-CoA dehydrogenase deficiency: genotype-biochemical phenotype correlations." Mol Genet Metab. 87(1):32-9.

Waliany S et al. (2000). "Identification of three novel mutations of the palmitoyl-protein thioesterase-1 (PPT1) gene in children with neuronal ceroid-lipofuscinosis." Hum Mutat. 15(2):206-7.

Walter C et al. (2001). "Disorders of peroxisome biogenesis due to mutations in PEX1: phenotypes and PEX1 protein levels." Am J Hum Genet. 69(1):35-48.

Walter MC et al. (2004). "FKRP (826C>A) frequently causes limb-girdle muscular dystrophy in German patients." J Med Genet. 41(4):e50.

Wang AZ, et al (2011). Association of SNP rs17465637 on chromosome 1q41 and rs599839 on 1p13.3 with myocardial infarction in an American caucasian population. Ann Hum Genet.

Wang QJ et al. (2007). "A distinct spectrum of SLC26A4 mutations in patients with enlarged vestibular aqueduct in China." Clin Genet. 72(3):245-54.

WANG Wei , et al (2011). Polymorphism on chromosome 9p21.3 contributes to early-onset and severity of coronary artery disease in non-diabetic and type 2 diabetic patients. Chinese medical journal

Ward CJ et al. (2003). "Cellular and subcellular localization of the ARPKD protein; fibrocystin is expressed on primary cilia." Hum Mol Genet. 12(20):2703-10.

Wasserstein MP et al. (2006). "Acid Sphingomyelinase Deficiency." [Updated 2015 Jun 18].

Waters PJ et al. (2000). "Characterization of phenylketonuria missense substitutions, distant from the phenylalanine hydroxylase active site, illustrates a paradigm for mechanism and potential modulation of phenotype." Mol Genet Metab. 69(2):101-10.

Watson MS et al. (2004). "Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel." Genet Med. 6(5):387-91.

Winkelmann, et al (2007). Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nat Genet.

REFERENCES

Woodward OM et al. (2009) . "Identification of a urate transporter, ABCG2, with a common functional polymorphism causing gout." 106(25):10338-42. Proc. Natl. Acad. Sci. U.S.A.

Wreden CC et al. (2005). "Varied mechanisms underlie the free sialic acid storage disorders." J Biol Chem. 280(2):1408-16.

Wynn RM et al. (2001). "Biochemical basis of type lB (E1beta) mutations in maple syrup urine disease. A prevalent allele in patients from the Druze kindred in Israel." J Biol Chem. 276(39):36550-6.

Xian-Fa Tang, et al(2012). Association analyses identify three susceptibility Loci for vitiligo in the Chinese Han population. J Invest Dermatol.

Xiang Ren, et al (2010). Identification of NPPA variants associated with atrial fibrillation in a Chinese GeneID population. European Society of Cardiology

Xiangdong Liu et al.(2010) Genome-wide meta-analyses identify three loci associated with primary biliary cirrhosis Nat. Genet.

Xie F, et al (2011). Replication of Putative Susceptibility Loci from Genome- Wide Association Studies Associated with Coronary Atherosclerosis in Chinese Han Population. PLoS One.

Xingnan Li , et al (2010). Genome-wide association study of asthma identifies RAD50-IL13 and HLA-DR/DQ regions. J Allergy Clin Immunol.

Xiong F et al. (2010). "Molecular epidemiological survey of haemoglobinopathies in the Guangxi Zhuang Autonomous Region of southern China." Clin Genet. 78(2):139–48.

Xiong HY et al. (2015). "RNA splicing. The human splicing code reveals new insights into the genetic determinants of disease." Science. 347(6218):1254806.

Xu F et al. (2004). "The role of the LRPPRC (leucine-rich pentatricopeptide repeat cassette) gene in cytochrome oxidase assembly: mutation causes lowered levels of COX (cytochrome c oxidase) I and COX III mRNA." Biochem J. 382(Pt 1):331-6.

Xu F et al. (2012). "LRPPRC mutation suppresses cytochrome oxidase activity by altering mitochondrial RNA transcript stability in a mouse model." Biochem J. 441(1):275-83.

Yan Xu, et al (2011). A polymorphism in the ABCG1 promoter is functionally associated with coronary artery disease in a Chinese Han population. Nature Genetics.

Yan Yun, et al (2014). The SNP rs1883832 in CD40 Gene and Risk of Atherosclerosis in Chinese Population: A Meta-Analysis. PLOS one.

Yann Le Strat, et al (2008). The 3¢ Part of the Dopamine Transporter Gene DAT1/SLC6A3 Is Associated With Withdrawal Seizures in Patients With Alcohol Dependence. Alcohol Clin Exp Res

Ye H, et al (2014) Prevalence of Age-Related Macular Degeneration in an Elderly Urban Chinese Population in China: The Jiangning Eye Study. Invest Ophthalmol Vis Sci.

Yi Huang, et al (2013). Relationship between chemokine (C–X–C motif) ligand 12 gene variant (rs1746048) and coronary heart disease: Case–control study and meta-analysis. Gene.

Yigit S et al. (2008). "MEFV mutations in patients with familial Mediterranean fever in the Black Sea region of Turkey: Samsun experience [corrected]." J Rheumatol. 35(1):106-13.

Yiping Ba, et al (2011). Association of EPHX2 polymorphism G860A with hypertension in middle ages. Journal of Chinese Physician.

Yokota I et al. (1991). "Molecular survey of a prevalent mutation, 985A-to-G transition, and identification of five infrequent mutations in the medium-chain Acyl-CoA dehydrogenase (MCAD) gene in 55 patients with MCAD deficiency." Am J Hum Genet. 49(6):1280-91.

Yokota I et al. (1992). "Impaired tetramer assembly of variant medium-chain acyl-coenzyme A dehydrogenase with a glutamate or aspartate substitution for lysine 304 causing instability of the protein." J Biol Chem. 267(36):26004-10.

Yongqin Wang, et al (2014). Genetic Variants Associated with Myocardial Infarction and the Risk Factors in Chinese Population. PLOS one

Yoon JS et al. (2008). "Heterogeneity in the processing defect of SLC26A4 mutants." J Med Genet. 45(7):411-9.

Yuen WY et al. (2011). "Herlitz junctional epidermolysis bullosa: diagnostic features, mutational profile, incidence and population carrier frequency in the Netherlands." Br J Dermatol. 165(6):1314-22.

Yule Xu, et al (2008) Association of CFH, LOC387715, and HTRA1 polymorphisms with exudative age-related macular degeneration in a northern Chinese population. Molecular Vision.

Zare-Karizi Sh et al. (2011). "Mutation spectrum of phenylketonuria in Iranian population." Mol Genet Metab. 102(1):29-32.

Zekanowski C et al. (1997). "Molecular basis of mild hyperphenylalaninaemia in Poland." J Med Genet. 34(12):1035-6.

Zelante L et al. (1997). "Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans." Hum Mol Genet. 6(9):1605-9.

Zeng BJ et al. (2002). "Identification and characterization of novel mutations of the aspartoacylase gene in non-Jewish patients with Canavan disease." J Inherit Metab Dis. 25(7):557-70.

Zhang XJ, et al (2009). Psoriasis genome-wide association study identifies susceptibility variants within LCE gene cluster at 1q21. Nat Genet.

Zhao SX, et al (2013). Robust evidence for five new Graves' disease risk loci from a staged genome-wide association analysis. Hum Mol Genet.

Zschocke J et al. (2001). "Molecular and functional characterisation of mild MCAD deficiency." Hum Genet. 108(5):404-8.

Zschocke J. (2003). "Phenylketonuria mutations in Europe." Hum Mutat. 21(4):345-56.

Zurflüh MR et al. (2008). "Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency." Hum Mutat. 29(1):167-75.