



Personal Genetic Scan™ Report

Introduction

The Personal Genetic Scan Report:

Congratulations! Your Personal Genetic Scan Report is here, and you are eager to know what it says. Before you proceed further and go through the many pages of this report, please make sure that you have read the first pages of this report carefully so that you can understand your report better.

The Personal Genetic Scan report is intended to give you an overall summary of your genetic health and provide insights into how your genes may affect your health today and in the future. This report provides information about single gene conditions, rare and complex conditions, cancer markers, memory related conditions, drug responses as well as traits and wellness topics. This report is intended to be a general summary to give you a starting point to learn about your genetic makeup and health. While the personal genetic scan looks at a great number of markers, this specific report does not include every single marker tested. Not all the markers tested represent useful data at the present time but as research evolves and our understanding of how our genes work improves, more of the markers tested will have known functions. Results for all the markers tested for your sample can be found in your Raw Data Report which you can download from your account.

Scope:

This report is intended to be used as a tool to better understand your health and wellness and discover possible predispositions to genetic conditions. We strongly recommend that you read the Disclaimer section at the end of this report. In brief, what we provide is information about your genome that should be considered information of a non-specific nature and should never be used as a substitute for the advice of a healthcare professional or a genetic counselor. GenoTypica does not, under any circumstances, provide medical advice, professional diagnosis, opinions, treatment options, treatments or other health services to the users of our services or products. Nothing we provide should be construed as an attempt to offer or render a medical opinion or otherwise engage in the practice of medicine.

This report is not a diagnosis of any kind. Know that having a variant for a disease does not necessarily mean you will develop the disease. Additionally, having multiple variants for a specific disease does not indicate an increased risk of developing a disease. We do not test for every variant associated with a disease risk. Many factors contribute to the chance of developing a disease or condition such as lifestyle, environment, and family history. Together these factors, including genetic variations, play a part in influencing your health. The Personal Genetic Scan Report is just one tool to use to better understand your genetic health. Please note that this report is provided to you as described and defined in the Terms and Conditions, the Privacy policy, and research agreement that you agreed to when you submitted a sample for DNA analysis.

Organization of the Report:

The Personal Genetic Scan Report is organized into 7 sections. Each section contains results for a group of markers related to a specific topic. These sections are:

- Single Gene Conditions
- Rare Conditions
- Complex Conditions
- Cancer
- Traits and Wellness
- Memory Related
- Drug Interactions

Each section includes a description of the condition and for many, the inheritance pattern. This information is provided to help you better understand how having a certain variant could affect your health or well-being. Click on any condition name to learn more about it. The information is provided from publicly available databases.

Understanding Your Report:

To fully understand this report, there are several key terms you must know. This section defines these key terms and illustrates where they appear in your report.

Gene:

Genes are specific segments of DNA known to be responsible for controlling all the processes in your body. Each gene has its own specific role in the human body. Each gene has a specific arrangement of letters (called bases, in technical literature) and they are A, T, G and C, referred to as the DNA sequence of that gene. Variations or changes in any base (A, T, G or C) in a gene can disrupt the processes controlled by the gene and could result in a disease or condition.

Gene Affected:

The gene where the variant is located. Click on the link attached to each gene name to learn more about the role this gene plays in the development of a disease.

Name	Result	Gene Affected	Variant ID	Genotype
Charlevoix-Saguenay spastic ataxia	Variant Absent	SACS	rs281865117	II
	Variant Absent	SACS	rs281865118	GG
	Variant Absent	SACS	rs281865120	GG

Variant:

A genetic variant is a specific and known change in a person's DNA sequence. This change is often one base changing to another base at a specific position. Instead of the expected base, a different base or variant is found. Most variants are benign and do not affect a person's health, but others are known to cause or be associated with a specific disease.

Variant ID:

This is a unique identification number assigned to each variant. Clicking on the variant ID will take you to a page containing more in-depth information on this variant and its location on the affected gene.

Name	Result	Gene Affected	Variant ID	Genotype
Charlevoix-Saguenay spastic ataxia	Variant Absent	SACS	rs281865117	II
	Variant Absent	SACS	rs281865118	GG
	Variant Absent	SACS	rs281865120	GG

Genotype:

This indicates what version of a variant is present in your DNA. In other words, what exactly is the base or letter at a particular position. If the variant is absent, the genotype will read Match Reference (the genotype most commonly present in the normal population) or the expected base at that position. The match reference genotype is indicated with a green or black letter in this report and the report pages have a green background and borders. When the genotype is indicated with an orange color, one copy of the variant was detected. When the genotype is indicated with the red color, two copies of the variant were detected.

Name	Result	Gene Affected	Variant ID	Genotype
Charlevoix-Saguenay spastic ataxia	Variant Absent	SACS	rs281865117	II
	Variant Absent	SACS	rs281865118	GG
	Variant Absent	SACS	rs281865120	GG

Each disease section includes a description of the disease and its inheritance pattern. This information is provided to help you better understand how having a certain variant could affect your health. Click on any disease name to learn more about the disease.

Reading your Results:

There are 3 possible results for each variant tested. Each result is listed and explained below. When reading your report, refer to this section to better understand what the report might indicate so you can decide on further evaluation with the help of a counselor or other health professional who can better explain the condition that might be of concern to you.

Variant Absent:

This means we were not able to detect a variant of a specific marker associated with the listed disease or condition. In other words, you have the reference at that position. It also means that no familial history or potential pre-disposition is indicated. Based on current understanding, you are unlikely to be affected by this condition.

Carrier Detected:

This means you have 1 variant copy of a specific marker associated with the listed condition. One copy of your gene has the variant and the other copy does not have the variant (or is the reference base) at that position. This means that a potential pre-disposition or a potential familial history is indicated for the condition. You can also potentially pass this variant on to your offspring. Developing or showing symptoms of the disease depends on the specific inheritance pattern of the disease (dominant or recessive) as well as how strongly this marker is associated with the disease and other factors that contribute to the condition. Therefore, consulting with a genetic counselor/healthcare provider is wise to better understand your specific situation, particularly if there are concerns.

Variant Detected:

This means you have 2 variant copies of a specific marker associated with the listed condition. Both copies of your gene have the variant and therefore, this variant can be passed on to your offspring. This also means that a potential pre-disposition or a potential familial history is indicated for the condition. Developing or showing symptoms of the disease depends on the specific inheritance pattern of the disease (dominant or recessive) as well as how strongly this specific marker is associated with the disease and other factors that contribute to the condition. Keep in mind that this is a marker associated with a condition and not all markers associated with a condition affect everyone (as other complex factors are involved) in the same way. We strongly recommend that you consult with a genetic counselor/healthcare provider to evaluate your specific situation.

Things to Remember:

We hope you find this report useful and educational in your journey to better understand your genetic, health, and wellness. When reading your Personal Genetic Report please remember the following:

- This report is not a diagnosis for any disease or condition listed or a recommendation for treatment or advice regarding your health or health condition(s). Please read the Disclaimer section at the end of the report.
- Having one variant or multiple variants for a disease does not necessarily mean you will get the disease.
- This report does not contain information for all the markers tested. We do not test for every variant associated with each disease or trait listed.

If you have concerns, please get in touch with your healthcare provider or genetic counselor.

Also, talking to friends or others who are a part of advocacy groups associated with specific conditions can provide information and support. The National Organization for Rare Disorders website maintains a list of advocacy groups and they can be found here: [Rare Disease](#)

Also, a genetic counselor can help in many ways. The role of a genetic counselor is explained here: [Genetic Counselor Information](#)

To find a genetic counselor near you, please go to this site: [Find a Genetic Counselor](#)

Also, information about DNA testing and Genome Analysis was provided to you on the GenoTypica website. [DNA TestingGenome Analysis](#)

Single Gene Conditions

This personal genetic scan report provides information on genetic variants associated with single gene diseases or conditions. These are diseases or conditions that occur due to changes in one specific gene.

Report Limitations:

This report does not include all possible variants for a disease. This report does not diagnose any diseases or conditions listed. This is not a diagnostic test for any of the listed disease or condition and should not be substituted for a formal test ordered by a healthcare provider. Other factors contribute to your chances of developing all listed diseases and conditions, including lifestyle, environment, and family history. You should contact a health care professional/genetic counselor before making any decisions that could affect your health.

Summary of Results:

Variant Detected:

[Familial hypercholesterolemia 1](#)

[Wilson disease](#)

Carrier Detected:

[Thrombophilia due to factor V Leiden](#)

Charlevoix-Saguenay spastic ataxia

Autosomal recessive spastic ataxia of Charlevoix-Saguenay, more commonly known as ARSACS, is a condition affecting muscle movement. People with ARSACS typically have abnormal tensing of the muscles (spasticity), problems with balance and coordination (cerebellar ataxia), and reduced sensation and weakness in the arms and legs (peripheral neuropathy).

Additional muscle problems that can occur in ARSACS include muscle wasting (amyotrophy), involuntary eye movements (nystagmus), and difficulty swallowing (dysphagia) and speaking (dysarthria). Other features of ARSACS involve high-arched feet (pes cavus), a spine that curves to the side (scoliosis), yellow streaks of fatty tissue in the light-sensitive tissue at the back of the eye (hypermyelination of the retina), urinary tract problems, intellectual disability, hearing loss, and recurrent seizures (epilepsy).

An unsteady walking style (gait) is the first symptom of ARSACS. Walking problems usually begin between the ages of 12 months and 18 months, as toddlers are learning to walk. These movement problems worsen over time, with increased spasticity and ataxia of the arms and legs. In some cases spasticity goes away, but this apparent improvement is thought to be due to the wasting away (atrophy) of nerves in the arms and legs. Most affected individuals require wheelchair assistance by the time they are in their thirties or forties.

While this condition was named after the area in which it was first seen, the Charlevoix-Saguenay region of Quebec, Canada, ARSACS has been identified in individuals worldwide.

Inheritance:

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.

Name	Result	Gene Affected	Variant ID	Genotype
Charlevoix-Saguenay spastic ataxia	Variant Absent	SACS	rs281865117	II
	Variant Absent	SACS	rs281865118	GG
	Variant Absent	SACS	rs281865120	GG

Sample Report - Remaining Conditions will be displayed on Actual Report

Rare Conditions

When a disease or condition affects a small portion of the population, it is considered rare. Genetics is increasingly being used to inform our understanding of uncommon conditions and their underlying biological basis. Variants associated with the following rare conditions were examined as part of your personal genetic scan.

Report Limitations:

This report does not include all possible variants for a disease. This report does not diagnose any diseases or conditions listed. This is not a diagnostic test for any of the listed disease or condition and should not be substituted for a formal test ordered by a healthcare provider. Other factors contribute to your chances of developing all listed diseases and conditions, including lifestyle, environment, and family history. You should contact a health care professional/genetic counselor before making any decisions that could affect your health.

Summary of Results:

Variant Detected:

[Stargardt disease 1](#)

[Propionic acidemia](#)

[Von Hippel-Lindau syndrome](#)

[Leigh syndrome](#)

[Duchenne muscular dystrophy](#)

[Sandhoff disease](#)

Carrier Detected:

[Von Hippel-Lindau syndrome](#)

[Gilbert's syndrome](#)

Familial cold autoinflammatory syndrome 2

Familial cold autoinflammatory syndrome type 2 is a condition that causes episodes of fever, skin rash, and joint pain. These episodes can be triggered by exposure to cold temperatures, or they may arise without warning, and they can last a few hours to several days. These episodes typically begin in childhood and persist throughout life.

Episodes typically occur after an hour or more of cold exposure in affected individuals who are sensitive to cold; however only a few minutes of cold exposure is required in some individuals.

In people with familial cold autoinflammatory syndrome type 2, the most common symptom that occurs during an episode is a fever. Other common features are an itchy rash and joint and muscle pain.

Additional features of familial cold autoinflammatory syndrome type 2 include abdominal pain, diarrhea, headache, and nausea. Some affected individuals develop hearing loss (sensorineural deafness) due to chronic inflammation.

Inheritance:

In most cases, familial cold autoinflammatory syndrome type 2 is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most affected individuals have one parent with the condition. In some families, individuals with an NLRP12 gene variant may develop familial cold autoinflammatory syndrome type 2 but others with the variant do not, which is a situation known as reduced penetrance. In rare cases, familial cold autoinflammatory syndrome type 2 is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have variants. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Name	Result	Gene Affected	Variant ID	Genotype
Familial cold autoinflammatory syndrome 2	Variant Absent	NLRP12	rs104895564	GG
	Variant Absent	NLRP12	rs145171629	GG

Sample Report - Remaining Conditions will be displayed on Actual Report

Complex Conditions

Complex conditions imply that many genes are involved in such conditions. Your susceptibility is most often the result of the combination of alterations in multiple genes across the genome and therefore the analysis is also complex and subjective. Each genetic variation contributes to the overall risk you have in developing one of the complex conditions below. However, the proportion that each examined variant influences your risk in developing a disease will vary. Some variants are known to contribute more hereditary risk than other variants for a particular condition. There are also non-genetic factors, such as diet, lifestyle and the environment, that will also influence whether you develop a particular condition.

Report Limitations:

This report does not include all possible variants for a disease. This report does not diagnose any diseases or conditions listed. This is not a diagnostic test for any of the listed disease or condition and should not be substituted for a formal test ordered by a healthcare provider. Other factors contribute to your chances of developing all listed diseases and conditions, including lifestyle, environment, and family history. You should contact a health care professional/genetic counselor before making any decisions that could affect your health.

Summary of Results:

Variant Detected:

[Gout](#)

[Grave's Disease](#)

Carrier Detected:

[Age Related Macular Degeneration](#)

[Asthma](#)

[Coronary Artery Disease](#)

[Pompe Disease](#)

[Hemochromatosis \(HFE\)](#)

Gout

Gout is a type of arthritis, which is a group of related disorders caused by episodes of abnormal inflammation in the joints. People with gout have high levels of a substance called urate in the blood (hyperuricemia). Gout develops when hyperuricemia leads to the formation of urate crystals in joints, triggering an inflammatory response from the immune system.

In people with gout, the first episode of inflammation (called a flare) usually affects the big toe or other joints in the foot or ankle. If urate levels remain high, flares can recur, affecting additional joints throughout the body. The time between flares varies among affected individuals; however, most people who experience multiple flares have their second one within a year of their first.

Flares usually begin at night and can last several days. It is unclear what causes a flare to stop; the body likely turns off the inflammation response after a certain period of time. During a flare, individuals can experience throbbing or burning pain, swelling, warmth, redness, and difficulty moving the affected joint. Fevers may occur, after which the skin over the affected joint can begin to peel. Without treatment, people with gout can experience frequent flares and joint pain and damage, which can limit mobility and decrease quality of life.

In about 15 percent of people with gout, urate accumulates in the kidneys and forms kidney stones. As the condition worsens, urate crystals can also be deposited under the skin or in other soft tissue, forming a nodule called a tophus (plural: tophi). These tophi often form in the hands, elbows, or feet. Tophi do not typically cause pain, but they can become inflamed, infected, or ooze fluid. Depending on their location, tophi can interfere with movements such as walking or gripping objects.

Many people with gout also have other health conditions. Most affected individuals have high blood pressure (hypertension), chronic kidney disease, or obesity. Some also have diabetes, heart disease, or a history of stroke. It is unclear whether gout is the cause of a person's increased risk for these conditions, or whether the conditions cause the development of gout, or whether both of these situations occur to influence disease.

Inheritance:

The inheritance pattern of gout is unclear because many genetic and environmental factors appear to be involved. However, having a close relative with gout likely increases a person's risk of developing the condition.

Name	Result	Gene Affected	Variant ID	Genotype
Gout	Variant Detected	SLC2A9	rs6449213	TT
	Variant Absent	ABCG2	rs72552713	GG
	Variant Absent	SLC2A9	rs16890979	CC
	Variant Absent	ABCG2	rs2231142	GG
	Variant Absent	SLC2A9	rs737267	GG

Sample Report - Remaining Conditions will be displayed on Actual Report

Cancer

Cancer is the uncontrolled growth and multiplication of cells in your body. Cancer is very complex, and studies have demonstrated that it results from a combination of genetic and non-genetic factors. While most cancer develops because of random, or sporadic, alterations in your DNA (called somatic mutations), there are hereditary factors (germline mutations) that can pre-dispose towards certain cancers. Because of hereditary pre-disposition, similar or related cancers may be present in multiple family members. If you are concerned that you could be at risk for any inherited cancer, we strongly recommend that you speak with a genetic counselor. As part of your personal genetic scan, your DNA was examined for the following variants associated with various cancer conditions and it may include reported somatic variations as well as germline variations.

Report Limitations:

This report does not include all possible variants for a disease. This report does not diagnose any diseases or conditions listed. This is not a diagnostic test for any of the listed disease or condition and should not be substituted for a formal test ordered by a healthcare provider. Other factors contribute to your chances of developing all listed diseases and conditions, including lifestyle, environment, and family history. You should contact a health care professional/genetic counselor before making any decisions that could affect your health.

Summary of Results:

Variant Detected:

[Melanoma](#)

Carrier Detected:

[Colorectal Cancer](#)

Colorectal Cancer

The colon is the longest part of the large intestine which also absorbs some nutrients and water as food passes through the digestive system. The rectum is the lower part of the large intestine where stool is stored until it leaves the body. Colorectal cancer is cancer that develops in the tissues of the colon or rectum. Colon cancer refers to cancer that begins in the colon and rectal cancer refers to cancer that begins in the rectum. The term colorectal cancer is also used to refer to cancer of either of these parts.

Changes in DNA, also known as mutations or variants, can lead to colorectal cancer. Certain variants are inherited but some variants could develop over an individual's lifetime with no family history of colorectal cancer. Other factors that could affect the risk could be lifestyle, environment, age (risk increases with age), presence of polyps (growths) which could turn cancerous over time, familial adenomatous polyposis (FAP), Lynch syndrome (hereditary non-polyposis colorectal cancer), chronic ulcerative colitis or Crohn disease for 8 years or more, alcoholic use, smoking, and obesity.

Symptoms may not always be obvious, especially in the initial stages but could include Diarrhea, Constipation, other changes in bowel movement that is out of the ordinary, blood in stool, weight loss and fatigue. Experts recommend that individuals start screening for colorectal cancer at the age of 45.

Inheritance:

Inherited via both autosomal dominant and autosomal recessive meaning having just one copy has an increased risk.

Name	Result	Gene Affected	Variant ID	Genotype
Colorectal Cancer	Carrier Detected	AURKA	rs2273535	AT
	Variant Absent	MUTYH	rs36053993	CC
	Variant Absent	TCF7L2	rs7903146	CC
	Variant Absent	SMAD7	rs4939827	CC

Sample Report - Remaining Conditions will be displayed on Actual Report

Traits and Wellness

Differences in your DNA are known to influence response to everyday activities such as sleep, diet, and exercise. There are a variety of genetic variations that can influence your senses and physical traits. Every person has unique features and characteristics that are determined by the interaction of their DNA, environment, and lifestyle. Since genetics play a role in the inheritance of traits, different family members may share similar traits with you.

Your genome has been examined for the traits described below. A few variants may influence one trait or hundreds of variants may contribute to how a trait is expressed. The variants that have been examined are listed.

The information contained in your genome controls each chemical reaction and process in your body. Certain genetic variants may influence the metabolism or absorption of vitamins, drugs and nutrients. These activities are related to a healthy lifestyle and may inform your choices about dairy consumption, types of exercise, and sleep routine. The environment in which we live and our lifestyle, known as non-genetic factors, also contribute to our wellness and should be taken into consideration.

Tendinopathies in lower extremities (legs)

Tendinopathies in lower extremities are degenerative conditions of the tendon causing pain and disability. They are common in sport injuries due to overuse of tendons but can also affect non-athletes.

Studies have identified a number of markers associated with the risk of tendinopathies and ligament ruptures in the lower extremities. These markers are located on the genes COL1A1, GDF5, and MMP3.

The GDF5 gene encodes a protein involved in regulating the development of numerous tissue and cell types including cartilage and joints. The variant T causes a decrease in GDF5 activity which could lead to a decrease cartilage amount.

Gene	Variant ID	Genotype	Summary
GDF5	rs143383	AA	possible increased risk of suffering tendinopathies

References:

[Reference 1](#)

[Reference 2](#)

[Reference 3](#)

[Reference 4](#)

Sample Report - Remaining Conditions will be displayed on Actual Report

Memory Related

More work is continually being done to examine the exact cause of many memory related conditions like Alzheimer's disease. Your genes are known to play a role in the progression and development of many such conditions. Some genetic variations that have been associated with late onset Alzheimer's disease, which typically develops after age 65, have been evaluated. Characteristics of Alzheimer's disease are memory loss, cognitive decline, as well as changes in behavior and personality. Lifestyle and environmental factors are known to also influence the development and progression of Alzheimer's disease. You may share genetic risk factors for Alzheimer's disease with family members. We strongly recommend that it is important to discuss your questions or concerns regarding Alzheimer's disease with a healthcare professional/genetic counselor.

Report Limitations:

This report does not include all possible variants for a disease. This report does not diagnose any diseases or conditions listed. This is not a diagnostic test for any of the listed disease or condition and should not be substituted for a formal test ordered by a healthcare provider. Other factors contribute to your chances of developing all listed diseases and conditions, including lifestyle, environment, and family history. You should contact a health care professional/genetic counselor before making any decisions that could affect your health.

TREM2 Memory

Two studies have demonstrated that variations in the TREM2 gene are associated with Alzheimer's disease. One variation, or change, that occurs in this gene is thought to result in an altered immune state in the brain regions affected by Alzheimer's disease. The CT and TT genotypes (rs75932628) may increase the risk for developing Alzheimer's disease.

Gene	Variant ID	Genotype	Summary
TREM2	rs75932628	CC	normal risk of developing Alzheimer's disease

References:

[Reference 1](#)

[Reference 2](#)

[Reference 3](#)

[Reference 4](#)

Sample Report - Remaining Conditions will be displayed on Actual Report

Drug Interactions

An individual's response to medication varies greatly from one person to another. Many studies and genomics data available suggest that an individual's genetic makeup influences the response to different medications and dosages. This is because genes may affect how an individual metabolizes medication just like it metabolizes vitamins or other supplements we may take. Similarly, with the same medication, some individuals may experience adverse side effects while others may not be responsive to that medication at all. It is important to know that your response to medication will also depend on age, lifestyle, drug-drug interactions, etc. What we are reporting here is informational and helpful for you to perhaps understand why some drugs may not have worked for you in the past. There could be other reasons behind the effects you have noticed and therefore, it may not be genetic or connected to any of the genes reported here. What we are providing should be considered purely educational and informative.

This is not a diagnostic test and should not be considered as medical advice in any way, and any medication changes should only be made after first consulting with a physician or healthcare provider.

Warfarin metabolism and dosage response

Warfarin is an anticoagulant drug (blood thinner) that is prescribed to prevent blood clots for treating individuals with heart valve disease, irregular heartbeat, a history of heart attack, stroke, or a prior blood clot in the deep veins of the arms or legs. Some individuals show a complete resistance to the drug warfarin regardless of how high the dose is, these individuals will be at risk for developing blood clots if they remain on the average warfarin dose for treating blood clots. Individuals with warfarin sensitivity take longer time to metabolize warfarin which means the medicine remains in their body longer compared to an average person, these individuals will be at risk for abnormal bleeding if they take the average dose (or more) of warfarin.

Warfarin Resistance:

Some individuals show a complete resistance to the drug warfarin regardless of how high the dose is, these individuals will be at risk for developing blood clots if they remain on the average warfarin dose for treating blood clots. Some individuals exhibit incomplete resistance to the drug and for these individuals, a higher dose of warfarin can be beneficial.

Warfarin Sensitivity:

Individuals with a low tolerance for the drug warfarin have a sensitivity to the drug. Individuals with warfarin sensitivity take longer time to metabolize warfarin which means the medicine remains longer in their body compared to an average person. An individual with warfarin sensitivity will be at risk for abnormal bleeding if they take the average dose (or more) of warfarin.

CYP2C9

The CYP2C9 enzyme helps to break down compounds like steroid hormones and fatty acids. CYP2C9 enzyme also plays a major role in breaking down the drug warfarin and assists in metabolizing other Non-Steroidal Anti-Inflammatory Drugs (NSAID) such as ibuprofen.

The CC and AC genotypes (rs1057910) both indicate that an individual may be sensitive to warfarin and may need less of the drug to be effective, while the AA genotype typically indicates an average response to warfarin.

Gene	Variant ID	Genotype	Summary
CYP2C9	rs1057910	AA	average warfarin metabolism - normal dosage

CYP2C9

The CYP2C9 enzyme helps to break down compounds like steroid hormones and fatty acids. CYP2C9 enzyme also plays a major role in breaking down the drug warfarin and assists in metabolizing other Non-Steroidal Anti-Inflammatory Drugs (NSAID) such as ibuprofen.

For the rs1799853 variant, CC genotype typically indicates an average response to warfarin, while both the CT and TT genotypes both indicate that an individual may be sensitive to warfarin and may need less of the drug to be effective in preventing blood clots.

Gene	Variant ID	Genotype	Summary
CYP2C9	rs1799853	TC	~20% reduction in warfarin metabolism; some NSAID risk

VKORC1

The VKORC1 gene provides instructions for making a vitamin K epoxide reductase enzyme. The VKORC1 enzyme converts one form of vitamin K into another form which helps in activating blood clotting proteins.

For the rs9923231(CC) genotype typically indicates an average response to warfarin, while both the CT and TT genotypes both indicate that an individual may be sensitive to warfarin and may need less of the drug to be effective in preventing blood clots.

Gene	Variant ID	Genotype	Summary
VKORC1	rs9923231	CC	average warfarin metabolism - normal dosage

VKORC1

The VKORC1 gene provides instructions for making a vitamin K epoxide reductase enzyme. The VKORC1 enzyme converts one form of vitamin K into another form which helps in activating blood clotting proteins.

Gene	Variant ID	Genotype	Summary
VKORC1	rs8050894	CC	warfarin sensitive - lower dosage

References:

[Reference 1](#)

[Reference 2](#)

Sample Report - Remaining Conditions will be displayed on Actual Report

Citations

Descriptions Provided by:

MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US); [updated Jun 24; cited 2020 Jul 1]. Available from: <https://medlineplus.gov/>.

2001 Sherry et al. paper: Sherry, S.T., Ward, M.H., Kholodov, M., Baker, J., Phan, L., Smigielski, E.M., Sirotkin, K. (2001). dbSNP: the NCBI database of genetic variation. *Nucleic Acid Research*, 29: 308-311.

Raw Data

The results for every marker successfully tested are included in your Raw Data Report. While we do not provide a detailed summary of all the tested markers, there are many third-party sites that may be able to generate one for you. You can upload your raw data file to a third-party if you so desire.

Not all the markers tested represent useful data at the present time but as research evolves and our understanding of how our genes work improves, more of the markers tested will have known functions. Results for all the markers tested for your sample can be found in your Raw Data Report which you can download from your account. As research continues for human genetics and diseases, the association with these markers may be discovered, and you will already have your data ready to upload for an updated analysis.

Additionally, the raw data includes a complete set of markers related to ancestry. While we do not provide an ancestry report, there are many available resources (some free) where you can upload your raw data file and receive a detailed ancestry report. We may provide this service in the future.

Version Information

As more information becomes available the generated reports may change to include newly discovered conditions.

Your report was generated on 2023-08-03T11:29:34.488671

Disclaimer

Disclaimer: GenoTypica is a direct to consumer (DTC) company which provides genome (DNA) analysis services. Unlike others, we are focused on providing a personal genetic scan report or information regarding your genetic makeup that may have some influence on your current and/or future health and wellbeing. Please note that the goods and services we provide are not subject to review by the FDA or any other Government agency. GenoTypica is not a genetic testing service and should not be used for any medical or clinical purpose, including diagnosis and treatment of diseases. What we provide is information about your genome, should be considered information of a non-specific nature, and should never be used as a substitute for the advice of a healthcare professional. GenoTypica does not, under any circumstances, provide medical advice, professional diagnosis, opinions, treatment options or treatments or other health services to the users of our services or products. We strongly discourage anyone from “self-diagnosing” based on the information gathered from the PG Scan report or any electronic report we provide including, but not limited to, any attempt to use or adopt any data or information gained through our website or links to other sites provided, as a substitute for a consultation with a doctor or a genetic counselor. Nothing we provide should be construed as an attempt to offer or render a medical opinion or otherwise engage in the practice of medicine. WE DO NOT GIVE MEDICAL ADVICE AND NOTHING HEREIN CREATES A DOCTOR-PATIENT RELATIONSHIP.

Medical, genetic and health related information changes constantly. As a result, the information provided through the profile or any report provided by GenoTypica through any electronic means, may not be current, complete or exhaustive, nor should you rely on any information found online or through any data or communication from us as a recommendation of a course of treatment for you or any other individual. Reliance on any information provided by GenoTypica or through its Website, or any linked websites, is done solely at your own risk. GenoTypica, as a company, and individuals acting on behalf of GenoTypica do not recommend or endorse any specific tests, products, procedures, opinions or any information that may be provided on our website or on the linked websites. If you have any health concerns, or if you have questions regarding your overall health, you should always consult with your doctor or another health care professional.

Never disregard or delay medical advice or treatment as a result of something you have read on the GenoTypica Website, report, or downloaded data.

If you reside in the United States, and you have a medical emergency, please call 9-1-1 immediately. If you live outside the United States, and you have a medical emergency, please call your local emergency services immediately.