

SERPINE1/PAI-1 (Blood Clots)

Gene Report

REPORT CATEGORY —



HEART & BLOOD
VESSELS

Sample Client

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omicsedge

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DISCLAIMER

This report does not diagnose this or any other health conditions. Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Personal information

NAME

Sample Client

SEX AT BIRTH

Male

HEIGHT

5ft 5" 165cm

WEIGHT

137lb 62kg



How this works

Our Wellness Reports analyze how your DNA influences your health.
We then use this analysis to give you personalized risk estimates and recommendations.



Similarly, our Trait Reports look at how your DNA influences your traits.



Your DNA is like an instruction manual — it contains a lot of information.
You can think of it as a blueprint for your body.

Genetic variants are parts of DNA that differ from person to person. Some can make you more vulnerable to certain health issues, while others may influence traits such as eye color.



We use artificial intelligence and machine learning to analyze all this information.
We then summarize your results as a risk score or display it on a gauge.

In total, we analyze up to 83 million genetic variants.

When we give a risk score, the risk icon tells you if you are at a higher or lower risk compared to other people:



Genotype color info:

AA

You don't have any risk alleles

AA

You have 1 risk allele

AA

You have 2 risk alleles

Your risk is also displayed as a percentile. This will tell you how your risks compare to our sample population. The lower your percentile number, the lower your risk. The "50th percentile" would be an average risk.

Similarly, the gauge tells you your relative risk score compared to our sample population, or it indicates a specific trait or haplotype you are more likely to have based on your genetic variants.

When applicable, we also list top evidence-based recommendations that may help lower your risk. The focus is on recommendations that may be of benefit to you, based on your genetics.

Our recommendations come in four categories: lifestyle, diet, supplements and drugs. The following icons tell you which category a recommendation falls into:



Our team of scientists also ranks each recommendation.
We rank based on impact and the strength of evidence in the medical literature.

Impact shows how strongly a recommendation will affect your health in a certain area.
Evidence is how much scientific support there is for the recommendation. Rankings are from 1 to 5 (low to high):



Impact

Impact scores range from 1-5. These scores reflect how much of an effect each recommendation can have. An impact score of 5 predicts the biggest effect.

When a recommendation affects something we can measure, we use those measurements to assign the impact score. For example, a recommendation that decreases cholesterol by 20% will have a higher impact score than one that decreases it by 5%.

Some recommendations affect things that we cannot directly measure, like stress or mood. For these, the impact score is based on how well they work relative to other recommendations and standard treatments. The best ones get the highest scores.

If there is a lot of research that shows a recommendation works especially well for your genotype, the impact score gets increased.

Recommendation Evidence

- 5 / 5
- Recommendations that are considered effective and generally recommended by experts and medical bodies.
-
- 4 / 5
- Recommendations that are considered likely effective and that have multiple independent meta-analyses and a great many studies supporting them.
-
- 3 / 5
- Recommendations that are considered possibly effective and have many studies supporting them
-
- 2 / 5
- Recommendations that have insufficient evidence, with two or several clinical trials supporting them, or many studies but with ambiguous results.
-
- 1 / 5
- Recommendations that have insufficient evidence, with a single clinical trial, or with many studies most of which didn't find support for the recommendation.
-
- 0 / 5
- No evidence in humans.

Genotype-specific Evidence

- High-quality
- Direct evidence that a recommendation helps more in people with your gene variant (many clinical trials, a few large clinical trials, or a meta-analysis).
-
- Medium-quality
- Direct evidence that a recommendation helps more in people with your gene variant (a few clinical trials or one large clinical trial).
-
- Low-quality
- Direct evidence that a recommendation helps more in people with your gene variant (a single clinical trial or more trials with inconsistent results).
-
- Indirect
- A recommendation may help more in people with your gene variant because it targets a specific gene or protein affected by your variant (e.g., MTHFR, dopamine).
-
- In theory
- A recommendation may help more in people with your gene variant because it targets a specific mechanism affected by your variant (e.g., inflammation, oxidative stress).

Some things to keep in mind:

- Genetics doesn’t play a considerable role in a condition or a trait.
- There is not enough research available to estimate a genetic predisposition.
- There are technical limitations to estimating or presenting a genetic predisposition.
- The topic is sensitive, and a genetic predisposition should only be estimated and presented by a healthcare professional.

Introduction

The [SERPINE1](#) gene encodes a protein called ‘*serpin family E member 1*’ or ‘*plasminogen activator inhibitor-1*’ (PAI-1). PAI-1 is the principal inhibitor of tissue plasminogen activator (tPA) and urokinase plasminogen activator (uPA), two proteins involved in the process of breaking down clots (*fibrinolysis*) [\[R\]](#).

During this process, the soluble protein plasminogen is transformed into its active form plasmin. This enzyme cleaves fibrin (the protein that forms blood clots) into degradation products such as the D-dimer. Both tPA and uPA activate this process by promoting plasmin production. In contrast, PAI-1 acts as an inhibitor of these proteins and allows the formation of blood clots [\[R\]](#).

Defects in this gene cause PAI-1 deficiency, a rare genetic disorder characterized by excessive bleeding and breakdown of blood clots. In contrast, variants resulting in higher PAI-1 levels may cause excessive blood clotting (*thrombophilia*) [\[R\]](#), [\[R\]](#).

SERPINE1

Genetics

Several *SERPINE1* polymorphisms increase the levels of PAI-1, resulting in an increased risk of blood clots and associated conditions such as ischemic stroke or myocardial infarction. Some of them include:

- The ‘D’ allele of [rs1799768](#), which acts as a proxy for another variant (4G/5G) and has been associated with myocardial infarction, ischemic stroke, asthma exacerbations, multiple organ dysfunction in critically ill patients, metabolic syndrome, sepsis, and cancer (particularly, endometrial) [[R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#)].
- The ‘A’ allele of [rs2227631](#), which is usually inherited with the previous one and has been associated with an increased risk of acute coronary heart disease, atherosclerosis, coronary syndrome, stroke, subarachnoid hemorrhage, PCOS, osteonecrosis of the femoral head, and breast cancer, as well as with worse outcomes in COVID-19 and Parkinson’s disease patients [[R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#), [R](#)].
- The ‘G’ allele of [rs7242](#), which has been associated with an increased risk of myocardial infarction and major depression disorder, worse outcomes in subarachnoid hemorrhage patients, and reduced risk of radiation-induced lung inflammation in lung cancer patients [[R](#), [R](#), [R](#), [R](#)].
- The ‘A’ allele of [rs6092](#), which has been associated with an increased risk of blood clots in COVID-19 patients, osteonecrosis in children with acute lymphoblastic leukemia, and oral premalignant disorders, decreased survival in colorectal cancer patients, and a decreased risk of type 2 diabetes [[R](#), [R](#), [R](#), [R](#), [R](#), [R](#)].
- The ‘T’ allele of [rs2227692](#), which has been associated with an increased risk of blood clots in COVID-19 patients and diffuse-type gastric cancer, but a decreased risk of recurrence or metastasis in patients with breast cancer [[R](#), [R](#), [R](#)].



TYPICAL ACTIVITY

Predisposed to typical SERPINE1 activity based on 4 genetic variants we looked at

Your top variants that most likely impact your genetic predisposition:

GENE	SNP	GENOTYPE
SERPINE1	rs1799768	TT
SERPINE1	rs2227631	GA
SERPINE1	rs6092	AG
NAT16	rs7242	TG
AP1S1	rs2227692	CC

The number of "risk" variants in this table doesn't necessarily reflect your overall result.