

9p21 (Heart Disease)

Gene Report

REPORT CATEGORY —



HEART & BLOOD
VESSELS

Sample Client

Report date: 15 January 2026

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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

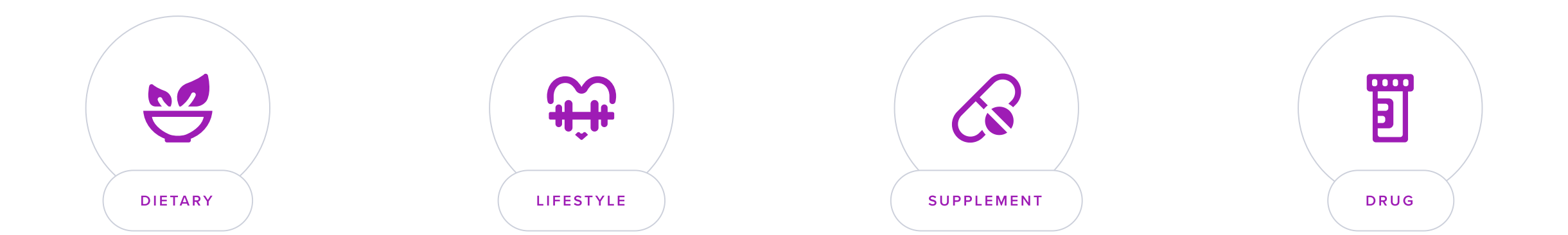
You have 2 risk alleles

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 9p21 locus (specifically the region near the [CDKN2A](#) and [CDKN2B](#) genes) has emerged as one of the most significant genetic loci associated with coronary artery disease (CAD) and other forms of heart disease. Multiple large-scale genome-wide association studies (GWAS) have linked single-nucleotide polymorphisms (SNPs) at this locus to increased risk of heart disease, particularly in populations of European descent [\[R\]](#), [\[R\]](#), [\[R\]](#)

The 9p21 region is located on chromosome 9 and contains genes related to cell cycle regulation, cell proliferation, and apoptosis—key processes involved in the inflammatory response that contributes to atherosclerosis and plaque formation. Importantly, it includes the *CDKN2B-AS* (*‘CDKN2B and CDKN2A antisense cis and trans regulatory RNA 1’*) gene, also known as *ANRIL* (*‘antisense non-coding RNA in the INK4 locus’*), which encodes a regulatory RNA molecule. Risk alleles at 9p21 have been shown to increase inflammation within arterial walls, impair vascular repair, and contribute to plaque rupture, which can ultimately lead to heart attacks and stroke [\[R\]](#), [\[R\]](#).

The 9p21 risk variants are believed to exert their effects through increased vascular inflammation and impaired cell cycle regulation in vascular smooth muscle cells (VSMCs) and endothelial cells. These processes lead to [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#):

- **Cellular proliferation:** Risk alleles can lead to excessive proliferation of VSMCs in the arterial wall, contributing to plaques that are prone to rupture.
- **Reduced apoptosis:** Reduced programmed cell death of damaged cells leads to plaque growth and increased plaque vulnerability.
- **Impaired endothelial function:** The risk alleles can disrupt the ability of endothelial cells to repair damaged blood vessels, leading to a loss of vascular integrity and an increased risk of atherosclerosis.

9p21 Genetics

One of the main variants in the 9p21 locus is [rs1333049](#), located near the *CDKN2B-AS* gene. Its minor ‘C’ allele has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Myocardial infarction [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Sudden cardiac death [\[R\]](#)
- Stroke [\[R\]](#)
- Myocardial infarction and cardiac death after acute coronary syndrome [\[R\]](#)
- Low 5-year survival in myocardial infarction patients [\[R\]](#)

Eating a Mediterranean diet may reduce the negative impact of this variant on myocardial infarction risk [\[R\]](#).

Another well-researched 9p21 variant is [rs10757278](#), also belonging to the *CDKN2B-AS* gene. Its minor ‘G’ allele has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Ischemic stroke [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Myocardial infarction [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Lower 5-year survival in myocardial infarction patients [\[R\]](#)
- Abdominal and intracranial aneurysm [\[R\]](#), [\[R\]](#)
- Acute coronary syndrome [\[R\]](#)
- Sudden cardiac death [\[R\]](#)
- Cancer (especially, breast and prostate) [\[R\]](#)

The ‘G’ allele of another 9p21 locus variant, [rs2383206](#), has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Ischemic stroke [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Myocardial infarction [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)

A study of over 8k participants found that a low adherence to a prudent diet rich in raw vegetables and fruits further increased the impact of this allele on heart disease risk [\[R\]](#).

Another variant linked to negative heart health outcomes is [rs2383207](#), whose ‘G’ allele has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Myocardial infarction [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Ischemic stroke [\[R\]](#), [\[R\]](#)



WORSE GENETICS

Predisposed to a worse 9p21 genetics based on 6 genetic variants we looked at

Your top variants that most likely impact your genetic predisposition:

GENE	SNP	GENOTYPE
CDKN2B	rs1333049	CC
CDKN2B	rs10757278	GG
CDKN2B	rs2383206	GG
CDKN2B	rs2383207	GG
CDKN2A	rs4977574	GG
CDKN2A	rs10757274	GG

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

Another minor variant at this locus, ‘G’ of [rs4977574](#), has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#)
- Myocardial infarction [\[R\]](#)
- Low 5-year survival in myocardial infarction patients [\[R\]](#)

Importantly, the negative impact of this variant on heart disease risk is further increased in people with a high intake of sugar-sweetened beverages [\[R\]](#).

Finally, the ‘G’ allele of [rs10757274](#) has been associated with an increased risk of:

- Coronary artery disease [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Myocardial infarction [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Sudden and arrhythmic cardiac death [\[R\]](#)
- Stroke [\[R\]](#)

Most of these variants are usually inherited together, meaning you will most likely carry all or none of them.