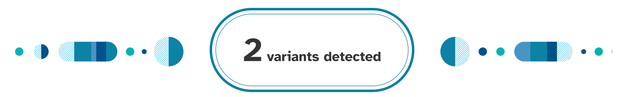
Hereditary Thrombophilia

Hereditary thrombophilia is a predisposition to developing harmful blood clots. These harmful blood clots most commonly form in the legs and can travel to the lungs. This test includes the two most common variants linked to hereditary thrombophilia.

Overview Scientific Details Frequently Asked Questions

Jamie, you have **both** of the genetic variants we tested.

People with these variants have an increased risk of developing harmful blood clots. Lifestyle, environment, and other factors can also affect your risk.



One variant detected in the F5 gene and one variant detected in the F2 gene.

How To Use This Test

This test does not diagnose hereditary thrombophilia 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.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

Intended Uses

- Tests for the **Factor V Leiden** variant in the F5 gene and the **Prothrombin G20210A** variant in the F2 gene linked to hereditary thrombophilia.

Limitations

- Does **not** test for all possible variants linked to hereditary thrombophilia.
- Does **not** test for variants in other genes linked to hereditary thrombophilia.

Ethnicity Considerations

- The variants included in this test are most common and best studied in people of **European** descent.
- These variants are also found in populations with European ancestry, like African Americans and Hispanics or Latinos.

You have an **increased risk** of developing harmful blood clots based on your genetic result.

It is important to discuss this result with a healthcare professional.

We detected the Factor V Leiden variant in the F5 gene and the Prothrombin G20210A variant in the F2 gene.

See Scientific Details

People with these variants are more likely to develop harmful blood clots than people without these variants.

These variants have been best studied in people of **European** descent. This doesn't mean you will definitely develop harmful blood clots. Other factors also influence your risk.

Since you share DNA with your family members, they may also be interested in this result.

One or both of your parents is expected to have at least one of these variants. In addition, each of your children likely has a 75% chance of having one or both variants. Your siblings may also have one or both variants.

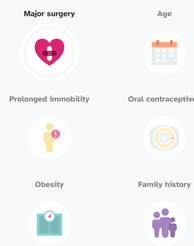
Lifestyle and other factors can also influence the chances of developing harmful blood clots.

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

Major surgery

Undergoing major surgery increases a person's risk of developing harmful blood clots.

See Scientific Details for more information



About Hereditary Thrombophilia

Hereditary thrombophilia is a predisposition to developing harmful blood clots. Harmful blood clots are more generally known as venous thromboembolism (VTE). When they form in the legs, the condition is known as deep vein thrombosis (DVT). When they travel to the lungs, the condition is known as pulmonary embolism (PE).

When it develops

Hereditary thrombophilia is genetic, but the risk of developing harmful blood clots increases with age and other factors.

Typical signs and symptoms of harmful blood clots

- Pain, tenderness, swelling, or redness in one or both legs
- Chest pain
- Difficulty breathing
- Hereditary thrombophilia may also be associated with recurrent late pregnancy loss in some people.

How common is the condition?

It is estimated that 1 in 29 people of European descent has a variant linked to hereditary thrombophilia. Around 1 in 1,999 people of European descent develops harmful blood clots each year. Some of these people have genetic variants and many others do not. Estimates are not as well known for people of other ethnicities.

How it's treated

Hereditary thrombophilia typically does not require any ongoing treatment. In some cases medications can be used to prevent harmful blood clots from forming. Medications and surgery can also be used to break up existing clots.

Read more at: Mayo Clinic MedlinePlus (Factor V Leiden) MedlinePlus (Prothrombin)

It is important to discuss this result with a healthcare professional.

If you have a personal or family history of harmful blood clots, consult with a healthcare professional.

Print report

If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help.

Learn more

See our Frequently Asked Questions for more information.

FAQs

Hereditary Thrombophilia

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Overview Scientific Details Frequently Asked Questions

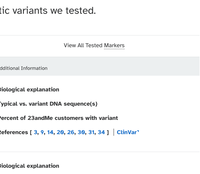
Hereditary thrombophilia is linked to variants in multiple genes.

This report includes one variant in the F5 gene and one variant in the F2 gene. This test does not cover variants in other genes linked to hereditary thrombophilia.

F5 F2

The F5 gene contains instructions for making a protein called coagulation factor V. When activated, factor V converts a protein called prothrombin into another protein called thrombin. Thrombin is important for forming blood clots. Certain variants in the F5 gene make the factor V protein harder to inactivate. This leads to more thrombin, which increases the chances of forming blood clots.

Read more at MedlinePlus*



You have both of the genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*			Additional Information
Factor V Leiden Gene: F5 Marker: rs6025	C Typical copy from one of your parents		T Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 9, 14, 26, 26, 30, 31, 34] | ClinVar*
Prothrombin G20210A Gene: F2 Marker: i3002432	A Variant copy from one of your parents		G Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [9, 12, 14, 24, 30, 34] | ClinVar*

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides risk estimates for venous thromboembolism (VTE), or the development of harmful blood clots, in people of European descent. Estimates for other ethnicities are not currently available.

Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

These risk estimates apply to a person experiencing a harmful blood clot for the first time, and do not apply if a person has developed one previously. A person who has already had harmful blood clots in the past may be at greater risk of developing another one, regardless of his or her genetic result.

Risk estimates are not available for people who have two copies of one tested variant and at least one copy of the other tested variant. An interpretation of "increased risk" is provided to people with these results because it is likely that their risk is at least as high as for people with two copies of a single variant only.

Consider talking to a healthcare professional if you have any concerns about your results.

References [20, 30]

One-year risk | Likelihood ratios | Odds ratios

Each year a number of people develop harmful blood clots. The one-year risk describes how many people on average are expected to develop a harmful blood clot per year.

Genotype	One-year risk
General population	1 in 1,999
One copy of Factor V Leiden variant	2 in 1,999
Two copies of Factor V Leiden variant	15 in 1,999
One copy of Prothrombin G20210A variant	Not available
Two copies of Prothrombin G20210A variant	Not available
One Factor V Leiden variant and one Prothrombin G20210A variant	Not available

Other Factors

Other factors besides the variants included in this test can influence your chances of developing harmful blood clots.

This is not a complete list of other factors.

People with multiple risk factors, including having one or more genetic variants, may have a higher risk of developing harmful blood clots.

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

Other Factors	References
Major surgery Major surgery (requiring general anesthesia and lasting at least 30 minutes) can greatly increase the chances of developing harmful blood clots. The risk depends on the type and location of the surgery, and if there are other risk factors. In some cases medication may be used to prevent harmful blood clots from forming.	[2, 8, 11, 15]
Age As people age their chances of developing harmful blood clots increase. This may be because as people age they may develop more risk factors for developing harmful blood clots.	[2, 16, 26]
Prolonged immobility Long periods of not moving can increase a person's risk of developing harmful blood clots. The risk increases the longer a person is immobile. For example, being confined to bed for an extended time due to illness or injury can increase the risk of harmful blood clots. Sitting for more than four hours at a time during long-distance travel can also slightly increase the risk.	[2, 5, 22, 28]
Oral contraceptives People who use oral contraceptives have a higher risk for harmful blood clots than people who don't use these medications.	[2, 10, 33]
Hormone replacement therapies People who use estrogen replacement therapy or testosterone therapy have a higher risk for harmful blood clots than people who don't use these hormonal therapies.	[2, 7, 13, 33]
Obesity Obesity (BMI ≥30) increases a person's chances of developing harmful blood clots.	[2, 23, 28]
Family history People with a family history of harmful blood clots have an increased risk of developing harmful blood clots themselves. The risk is even higher in families with more than one affected family member, and in families with members who had a harmful blood clot at an early age.	[4, 18, 34]
Pregnancy People who are pregnant have an increased risk for harmful blood clots. The risk remains elevated for the first three months after delivery.	[2, 21]
Cancer People with cancer may have an increased risk of developing harmful blood clots. This may be because cancer treatment, or cancer itself, increases the risk.	[2, 32]
Smoking Current smokers have a higher risk of developing harmful blood clots than non-smokers.	[6]

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for Hereditary Thrombophilia is indicated for reporting of the Factor V Leiden variant in the F5 gene, and the Prothrombin G20210A variant in the F2 gene. This report describes if a person has variants associated with a higher risk of developing harmful blood clots, but it does not describe a person's overall risk of developing harmful blood clots. This report is most relevant for people of European descent.

Special Considerations

- Testing for genetic variants associated with hereditary thrombophilia is recommended by ACMG and ACOG under certain circumstances. This test includes the two variants recommended for testing by ACMG and ACOG.

Test Performance Summary

Clinical Performance [17]

- The Factor V Leiden variant is expected to be responsible for around 14% of all VTE events in people of European descent.
- The Prothrombin G20210A variant is expected to be responsible for around 4% of all VTE events in people of European descent.
- Other variants in other genes not tested here may be more often associated with risk of developing harmful blood clots in people of other ethnicities.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. American College of Obstetricians and Gynecologists Committee on Practice Bulletins–Obstetrics. (2018). "ACOG Practice Bulletin No. 197: Inherited Thrombophilias in Pregnancy." Obstet Gynecol. 132(1):e18–e34. *
2. Anderson FA Jr et al. (2003). "Risk factors for venous thromboembolism." Circulation. 107(23 Suppl 1):I9-16. *
3. Bertina RM et al. (1994). "Mutation in blood coagulation factor V associated with resistance to activated protein C." Nature. 369(6475):64-7. *
4. Bezemer ID et al. (2009). "The value of family history as a risk indicator for venous thrombosis." Arch Intern Med. 169(6):610-5. *
5. Cannegieter SC et al. (2006). "Travel-related venous thrombosis: results from a large population-based case control study (MEGA study)." PLoS Med. 3(4):e307. *
6. Cheng YJ et al. (2013). "Current and former smoking and risk for venous thromboembolism: a systematic review and meta-analysis." PLoS Med. 10(9):e1001515. *
7. Cushman M et al. (2004). "Estrogen plus progestin and risk of venous thrombosis." JAMA. 292(13):1573-80. *
8. Ehrenforth S et al. (2004). "Impact of environmental and hereditary risk factors on the clinical manifestation of thrombophilia in homozygous carriers of factor V/G1691A." J Thromb Haemost. 2(3):430-6. *
9. Emmerich J et al. (2001). "Combined effect of factor V Leiden and prothrombin 20210A on the risk of venous thromboembolism--pooled analysis of 8 case-control studies including 2310 cases and 3204 controls. Study Group for Pooled-Analysis in Venous Thromboembolism." Thromb Haemost. 86(3):809-16. *
10. ESHRE Capri Workshop Group. (2013). "Venous thromboembolism in women: a specific reproductive health risk." Hum Reprod Update. 19(5):471-82. *

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
April 17, 2017	Hereditary Thrombophilia report created.

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Hereditary Thrombophilia

What is a harmful blood clot?

What does this test do?

What does this test **not** do?

The report says the variants included in this test are most common and best studied in people of **European** descent. What if I'm not of European descent or am only partly of European descent?

Where can I learn more about hereditary thrombophilia, support groups, and other resources?

My report says **two variants** were detected. What does this mean?

What does **increased risk** mean?

My report says **two variants** were detected. What are some things I could do?

How could my result affect my family?

Have more questions? **Check out our Customer Care Help Center.**