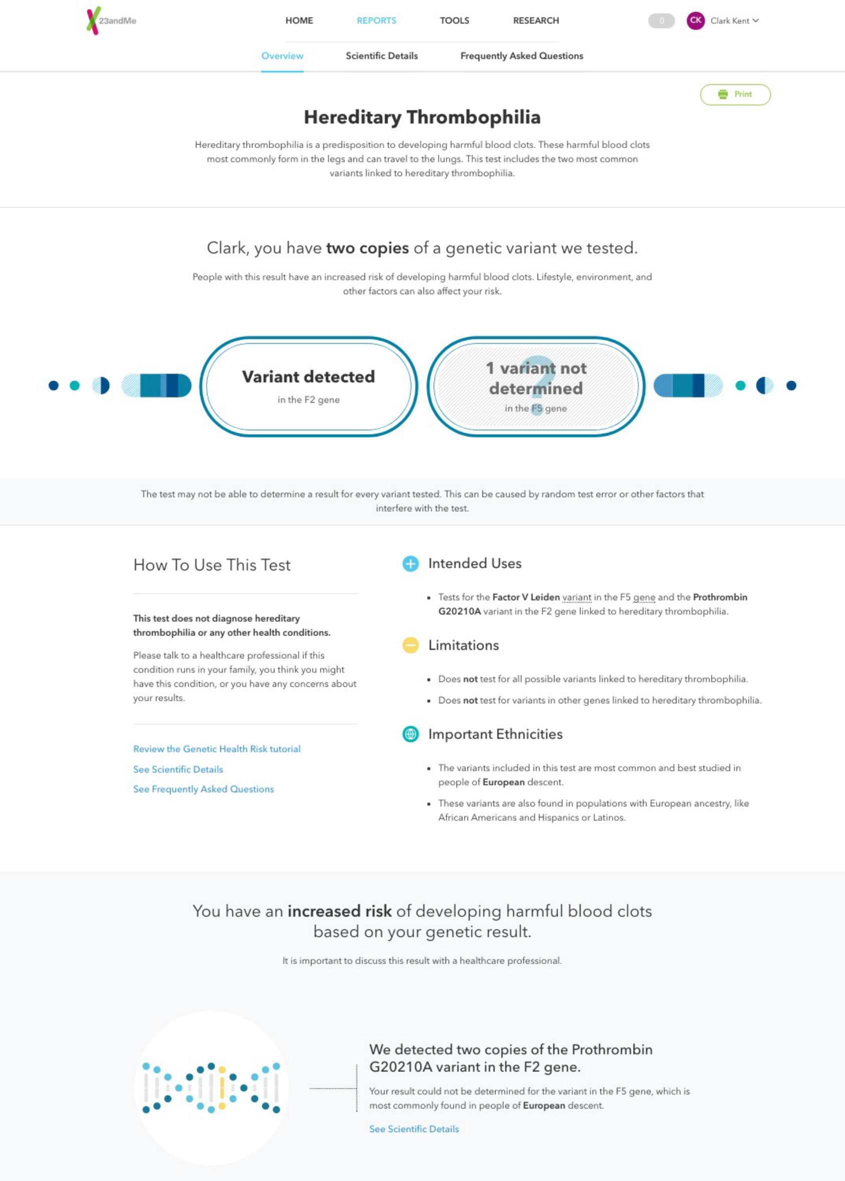


Hereditary Thrombophilia

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.

Clark, you have **two copies** of a genetic variant we tested.

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

Variant detected

in the F2 gene

1 variant not determined

in the F5 gene

The test may not be able to determine a result for every variant tested. This can be caused by random test error or other factors that interfere with the test.

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.

Important Ethnicities

- 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.

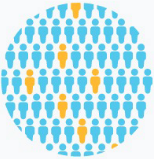
We detected two copies of the Prothrombin G20210A variant in the F2 gene.

Your result could not be determined for the variant in the F5 gene, which is most commonly found in people of **European** descent.

See Scientific Details

People with this result are more likely to develop harmful blood clots than people without this variant.

This variant has 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.

Both of your parents and each of your children likely have this variant. Your siblings likely have at least a 75% chance of having one or more copies of this variant.

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

Major surgery

Age

Prolonged immobility

Oral contraceptives

Obesity

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

How common is the condition?

It is estimated that 1 in 20 people of European descent has a variant linked to hereditary thrombophilia. Around 1 in 1,000 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

- Hereditary thrombophilia may also be associated with recurrent late pregnancy loss in some women.

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 Genetics Home Reference (Factor V Leiden) Genetics Home Reference (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

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Print

Hereditary Thrombophilia

Scientific Details

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.

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 Genetics Home Reference

Chromosome 1
Gene: F5

You have two copies of a genetic variant we tested. Your result for the other tested variant could not be determined.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*	Additional Information
Factor V Leiden **Gene:** F5 **Marker:** rs6025	**Not determined**	**Biological explanation** **Typical vs. variant DNA sequence(s)** **Percent of 23andMe customers with variant** **References** [2, 8, 11, 12, 18, 24, 28, 29] | ClinVar
Prothrombin G20210A **Gene:** F2 **Marker:** i3002432	**A** Variant copy from one of your parents **A** Variant copy from your other parent	**Biological explanation** **Typical vs. variant DNA sequence(s)** **Percent of 23andMe customers with variant** **References** [8, 10, 11, 12, 22, 28] | 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 [18, 28]

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 1,000
One copy of Factor V Leiden variant	2 in 1,000
Two copies of Factor V Leiden variant	15 in 1,000
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 for 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.	[1, 7, 9, 13]
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.	[1, 14, 24]
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.	[1, 3, 20, 26]
Oral contraceptives Women who use oral contraceptives or hormone replacement therapy have a higher risk for harmful blood clots than women who don't use these medications.	[1, 5, 6, 31]
Obesity Obesity (BMI ≥30) increases a person's chances of developing harmful blood clots.	[1, 21, 26]
Pregnancy Women have an increased risk of developing harmful blood clots during pregnancy and in the first three months after delivery.	[1, 19]
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.	[1, 30]
Smoking Current smokers have a higher risk of developing harmful blood clots than non-smokers.	[4]

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 under certain circumstances. This test includes the two variants recommended for testing by ACMG.

Test Performance Summary

Clinical Performance [15]

- 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.

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.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 404 samples. 404 out of 404 genotype results were correct. While unlikely, this test may provide false positive or false negative results.

References

1. Anderson FA Jr et al. (2003). "Risk factors for venous thromboembolism." Circulation. 107(23 Suppl 1):I9-16.
2. Bertina RM et al. (1994). "Mutation in blood coagulation factor V associated with resistance to activated protein C." Nature. 369(6475):64-7.
3. Cannegieter SC et al. (2006). "Travel-related venous thrombosis: results from a large population-based case control study (MEGA study)." PLoS Med. 3(8):e307.
4. 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.
5. Cushman M et al. (2004). "Estrogen plus progestin and risk of venous thrombosis." JAMA. 292(13):1573-80.
6. ESHRE Capri Workshop Group. (2013). "Venous thromboembolism in women: a specific reproductive health risk." Hum Reprod Update. 19(5):471-82.
7. 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.
8. 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.
9. Geerts WH et al. (2008). "Prevention of venous thromboembolism: American College of Chest Physicians Evidence-Based Clinical Practice Guidelines (8th Edition)." Chest. 133(6 Suppl):381S-453S.
10. Gehring NH et al. (2001). "Increased efficiency of mRNA 3' end formation: a new genetic mechanism contributing to hereditary thrombophilia." Nat Genet. 28(4):389-92.
11. Gohil R et al. (2009). "The genetics of venous thromboembolism. A meta-analysis involving approximately 120,000 cases and 180,000 controls." Thromb Haemost. 102(2):360-70.
12. Grody WW et al. (2001). "American College of Medical Genetics consensus statement on factor V Leiden mutation testing." Genet Med. 3(2):139-48.
13. Heit JA et al. (2000). "Risk factors for deep vein thrombosis and pulmonary embolism: a population-based case-control study." Arch Intern Med. 160(6):809-15.
14. Heit JA et al. (2005). "The incidence of venous thromboembolism among Factor V Leiden carriers: a community-based cohort study." J Thromb Haemost. 3(2):305-11.
15. Heit JA et al. (2011). "Genetic variation within the anticoagulant, procoagulant, fibrinolytic and innate immunity pathways as risk factors for venous thromboembolism." J Thromb Haemost. 9(6):1133-42.
16. Kujovich JL et al. (1993). "Factor V Leiden Thrombophilia"
17. Kujovich JL et al. (1993). "Prothrombin-Related Thrombophilia"
18. Kujovich JL et al. (2011). "Factor V Leiden Thrombophilia." Genet Med. 13(1):1-16.
19. Marik PE. (2010). "Venous thromboembolism in pregnancy." Clin Chest Med. 31(4):731-40.
20. Ocak G et al. (2013). "Risk of venous thrombosis in patients with major illnesses: results from the MEGA study." J Thromb Haemost. 11(1):116-23.
21. Pomp ER et al. (2007). "Risk of venous thrombosis: obesity and its joint effect with oral contraceptive use and prothrombotic mutations." Br J Haematol. 139(2):289-96.
22. Poort SR et al. (1996). "A common genetic variation in the 3'-untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increase in venous thrombosis." Blood. 88(10):3698-703.
23. Raskob GE et al. (2014). "Thrombosis: a major contributor to global disease burden." Arterioscler Thromb Vasc Biol. 34(11):2363-71.
24. Rosendaal FR et al. (1995). "High risk of thrombosis in patients homozygous for factor V Leiden (activated protein C resistance)" Blood. 85(6):1504-8.
25. Rosendaal FR et al. (2009). "Genetics of venous thrombosis." J Thromb Haemost. 7 Suppl 1:301-4.
26. Samama MM. (2000). "An epidemiologic study of risk factors for deep vein thrombosis in medical outpatients: the Sirius study." Arch Intern Med. 160(22):3415-20.
27. Sidney S et al. (2004). "Venous thromboembolic disease in users of low-estrogen combined estrogen-progestin oral contraceptives." Contraception. 70(1):3-10.
28. Simone B et al. (2013). "Risk of venous thromboembolism associated with single and combined effects of Factor V Leiden, Prothrombin 20210A and Methylenetethraydrofolate reductase C677T: a meta-analysis involving over 11,000 cases and 21,000 controls." Eur J Epidemiol. 28(8):621-47.
29. Sun X et al. (1994). "Blood coagulation factor Va abnormality associated with resistance to activated protein C in venous thrombophilia." Blood. 83(11):3120-5.

30. Walker AJ et al. (2013). "Incidence of venous thromboembolism in patients with cancer - a cohort study using linked United Kingdom databases." Eur J Cancer. 49(6):1404-13.

31. Wu O et al. (2005). "Oral contraceptives, hormone replacement therapy, thrombophilias and risk of venous thromboembolism: a systematic review. The Thrombosis: Risk and Economic Assessment of Thrombophilia Screening (TREATS) Study." Thromb Haemost. 94(1):17-25.

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

Frequently Asked Questions

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.

Hereditary Thrombophilia

What is a harmful blood clot?

A blood clot is a clump of blood that has become a somewhat solid mass. Blood clots usually form to stop bleeding after an injury. A harmful blood clot is a clot that forms improperly and can obstruct blood flow to vital tissues. When these clots 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).

Is this answer helpful?   Yes   No

What does this test do?

This test looks for two specific genetic variants linked to hereditary thrombophilia, one in the F5 gene called Factor V Leiden, and one in the F2 gene called Prothrombin G20210A.

People with the genetic variants included in this test are more likely to develop harmful blood clots. However, not everyone with these genetic variants will develop a harmful blood clot.

This test provides risk estimates for developing a harmful blood clot for the first time. A person who has developed a harmful blood clot in the past may have a higher risk of developing another harmful blood clot, regardless of whether he or she has a variant detected by this test.

This test does not include all possible genetic variants that may increase your risk of developing harmful blood clots.

Is this answer helpful?   Yes   No

What does this test not do?

This test does not diagnose hereditary thrombophilia. Only a healthcare professional can do that.

This test does not tell you if you have harmful blood clots or if you will definitely develop them in the future.

This test does not provide risk estimates for developing a harmful blood clot if you have had one in the past. A person who has developed a harmful blood clot in the past may have a higher risk of developing another harmful blood clot, regardless of whether he or she has a variant detected by this test.

This test does not include all possible genetic variants that may increase your risk of developing harmful blood clots.

This test does not provide a complete assessment of your risk of developing harmful blood clots.

Is this answer helpful? Yes No

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?

The tested variants are expected to be less common in people who are only partly of European descent, and are rare in people who are not of European descent.

The effect of these variants on a person's risk of developing harmful blood clots is well understood in people of European descent. If a person who is not of European descent has one or more of these genetic variants, he or she is still expected to have a higher risk of developing harmful blood clots, but the exact risk numbers are not well understood. See Scientific Details for more information.

There are other genetic variants linked to a higher risk of developing harmful blood clots that can be more common in people of non-European descent. These other variants are not included in this test. It is still possible for a person to have one of these other genetic variants, especially if they are not of European descent.

Is this answer helpful? Yes No

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

You can learn more about hereditary thrombophilia from the following resources:

- National Blood Clot Alliance
- North American Thrombosis Forum
- American Society of Hematology
- Centers for Disease Control and Prevention
- National Health Services (United Kingdom)

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 about genetic counseling.

Is this answer helpful? Yes No

My report says **two copies of a variant** called **Prothrombin G20210A** were detected but **one variant could not be determined**. What does this mean?

This means you have two copies of a genetic variant we tested. But we could not tell if you have the other tested genetic variant. This can be caused by random test error or other factors that interfere with the test.

People with this result have an increased risk of developing harmful blood clots. However, this does not mean you have developed or definitely will develop harmful blood clots. Other risk factors can also affect your risk of developing harmful blood clots. Learn more about other factors.

Is this answer helpful? Yes No

What does it mean if the result for a variant **could not be determined**?

This means we could not tell if you have or do not have the tested genetic variant. This can be caused by random test error or other factors that interfere with the test.

Is this answer helpful? Yes No

What does **increased risk** mean?

An "increased risk" means that, based on your genetic result for the test, your chances of developing harmful blood clots are higher than average.

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

Non-genetic factors may also influence your risk of developing harmful blood clots. Learn more about other factors.

Is this answer helpful? Yes No

My report says **two copies of a variant** called **Prothrombin G20210A** were detected. What are some things I could do?

This result is associated with an increased risk of developing harmful blood clots. Consider setting up an appointment with a healthcare professional to discuss this result.

Is this answer helpful? Yes No

How could my result affect my family?

Since you share DNA with your family members, they may also be interested in your result. If you are thinking about talking to family members about your results, see this article for a discussion of things to consider before having the conversation.

Because you have two copies of one variant, it is expected that:

- Each of your children will inherit this variant from you.
- Each of your parents has at least one copy of this variant.
- Each of your siblings has at least a 75% chance of having one or more copies of this variant.

Is this answer helpful? Yes No

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

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