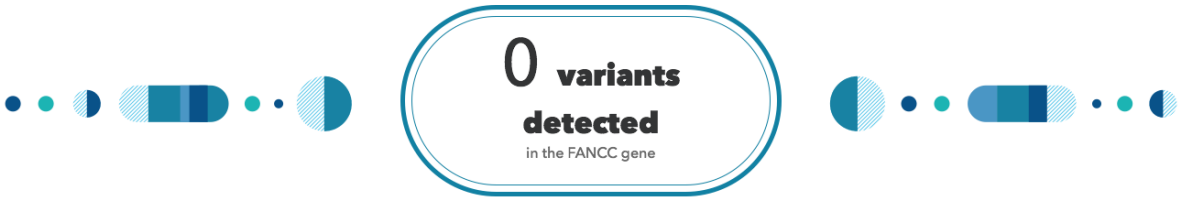
Fanconi Anemia Group C

Fanconi anemia group C is a rare genetic disorder. It is characterized by a decreased production of blood cells, birth defects, and an increased risk of infections and cancer. A person must have two variants in the FANCC gene in order to have this condition.

Erin, you **do not have the variants** we tested.

You could still have a variant not covered by this test.



How To Use This Test

This test does not diagnose any 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 Carrier Status tutorial

See Scientific Details

Intended Uses

- Tests for **multiple variants** in the FANCC gene.
- To identify carrier status for Fanconi anemia group C.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** other forms of Fanconi anemia.

Important Ethnicities

- This test is most relevant for people of **Ashkenazi Jewish** descent.

You are likely not a carrier.

This result is relevant for you because you have **Ashkenazi Jewish** ancestry.

We ruled out the most common variants for Fanconi anemia group C in people of Ashkenazi Jewish descent.

You still have a chance of being a carrier for Fanconi anemia group C.

You may still have up to a **1 in 88,000 chance** of carrying a variant not covered by this test.

See Scientific Details

About Fanconi Anemia Group C

When symptoms develop

Symptoms can develop anytime from birth to adulthood.

How it's treated

There is currently no known cure. Treatment focuses on increasing the number of blood cells, managing disabilities, and screening for cancer. Stem cell transplants may correct blood cell problems in some cases.

Typical signs and symptoms

- Skeletal and organ malformations at birth
- Increased risk of cancer
- Frequent infections
- Decreased blood cell production
- Very short height
- Areas of lighter or darker skin color

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish descent.

Read more at

Genetics Home Reference

GeneReviews

National Heart, Lung, and Blood Institute

Consider talking to a healthcare professional if you are concerned about your results.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

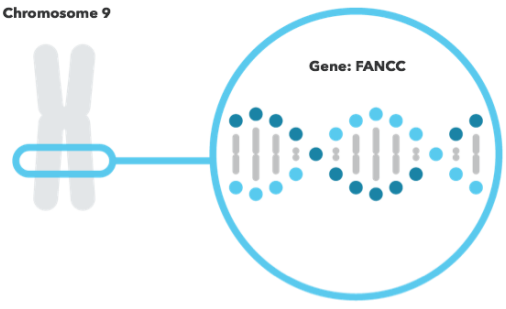
Learn more

Fanconi anemia group C is caused by variants in the FANCC gene.

FANCC

The FANCC gene contains instructions for making one part of a group of proteins called the Fanconi anemia core complex. This group of proteins plays an important role in the process of repairing damaged DNA. Certain variants in the FANCC gene result in a dysfunctional Fanconi anemia core complex.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	3

Marker Tested	Your Genotype*		Additional Information
IVS4+4A>T Gene: FANCC Marker: i4000336	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
R548X Gene: FANCC Marker: i4000412	**G** Typical 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** [4 , 6] | ClinVar
322delG Gene: FANCC Marker: i4000413	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [2 , 3 , 4 , 7] | 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 an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk** .

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of Ashkenazi Jewish descent only.

- For people of partial Ashkenazi Jewish descent, post-test carrier risk is less than that for those who are fully Ashkenazi Jewish. The exact post-test risk depends on how much Ashkenazi Jewish ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

Ashkenazi Jewish	1 in 88,000	[5]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Fanconi Anemia Group C is indicated for the detection of three variants in the FANCC gene. This test is intended to be used to determine carrier status for Fanconi anemia group C in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Ashkenazi Jewish descent.

Special Considerations

- Carrier testing for Fanconi anemia group C is recommended by ACMG for people of Ashkenazi Jewish descent considering having children. This test includes the one variant recommended for testing by ACMG.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Ashkenazi Jewish	>99%	[5]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 145 samples with known variant status. 145 out of 145 genotype results were correct. About 1 in 43,000 samples may receive a **Not Determined** result for one or more variants included in this test. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- 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. Alter BP et al. (1993). "Fanconi Anemia"
2. Donahue SL et al. (2004). "Intermediate DNA repair activity associated with the 322delG allele of the fanconi anemia complementation group C gene." J Mol Biol. 342(5):1443-55.
3. Gille JJ et al. (2012). "Diagnosis of Fanconi Anemia: Mutation Analysis by Multiplex Ligation-Dependent Probe Amplification and PCR-Based Sanger Sequencing." Anemia. 2012:603253.
4. Gillio AP et al. (1997). "Phenotypic consequences of mutations in the Fanconi anemia FAC gene: an International Fanconi Anemia Registry study." Blood. 90(1):105-10.
5. Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.
6. Huard CC et al. (2014). "The Fanconi anemia pathway has a dual function in Dickkopf-1 transcriptional repression." Proc Natl Acad Sci U S A. 111(6):2152-7.
7. Verlander PC et al. (1994). "Mutation analysis of the Fanconi anemia gene FACC." Am J Hum Genet. 54(4):595-601.