Tyrosinemia Type I

Tyrosinemia type I is a rare genetic disorder. It is characterized by high levels of the amino acid tyrosine that can lead to liver and kidney disease. A person must have two variants in the FAH gene in order to have tyrosinemia type I.

Overview Scientific Details

Jamie, 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 FAH gene.
- To identify carrier status for tyrosinemia type I.

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 subtypes of tyrosinemia.

Important Ethnicities

- This test is most relevant for people of **French Canadian** and **Finnish** descent.

You are likely not a carrier.

We ruled out the tested variants for tyrosinemia type I.

You still have a chance of being a carrier for tyrosinemia type I.

You may still have up to a **1 in 370 chance** of carrying a variant not covered by this test.

See Scientific Details

About Tyrosinemia Type I

Also known as: Fumarylacetoacetase Deficiency

When symptoms develop

Symptoms typically develop during infancy or in childhood.

Typical signs and symptoms

- High levels of tyrosine in the blood
- Liver and kidney problems
- Growth delay
- Episodes of pain, weakness, and mental distress
- Increased risk of liver cancer

Ethnicities most affected

This condition is most common in people of French Canadian, Finnish, Ashkenazi Jewish, European, Norwegian, and Turkish descent.

How it's treated

There is currently no known cure. Medication and a low protein diet may decrease liver and kidney damage. Liver transplantation is considered in some cases.

Read more at: Genetics Home Reference GeneReviews Orphanet

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

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Overview | Scientific Details

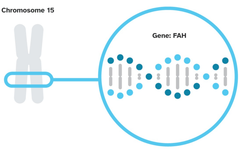
Tyrosinemia type I is caused by variants in the FAH gene.

FAH

The FAH gene contains instructions for making an enzyme called fumarylacetoacetate hydrolase. This enzyme breaks down the amino acid tyrosine, which is an important building block of many proteins. Certain variants in FAH prevent this function, leading to high levels of tyrosine byproducts.

Read more at Genetics Home Reference

Chromosome 15



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*			Additional Information
W262X **Gene:** FAH **Marker:** i5012862	**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 [10, 11, 13] | ClinVar
P261L **Gene:** FAH **Marker:** i5012861	**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 [3, 6] | ClinVar
IVS12+5G>A **Gene:** FAH **Marker:** i5012865	**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 [2, 3, 6, 7, 8, 9, 13] | ClinVar
IVS6-1G>T **Gene:** FAH **Marker:** i5012867	**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 [2, 3, 9] | 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 French Canadian, Ashkenazi Jewish, Finnish, European, Turkish, and Norwegian descent only.

- For people with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

French Canadian	1 in 289	[5]
Ashkenazi Jewish	1 in 149,999,999	[12]
Finnish	1 in 879	[12]
European	1 in 376	[12]
Turkish	1 in 219	[12]
Norwegian	1 in 249	[1]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Tyrosinemia Type I is indicated for the detection of four variants in the FAH gene. This test is intended to be used to determine carrier status for tyrosinemia type I in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of French Canadian and Finnish descent.

Special Considerations

- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

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.

French Canadian	98%	[7]
Ashkenazi Jewish	>99%	[12]
Finnish	86%	[7, 10, 11, 13]
European	69% (averaged across multiple countries)	[11]
Turkish	39%	[11]
Norwegian	42%	[4]

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.
- 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. **Angileri, Francesca et al. (2015). "Geographical and Ethnic Distribution of Mutations of the Fumarylacetoacetate Hydrolase Gene in Hereditary Tyrosinemia Type 1." JIMD Reports 19:43–58.**
2. **Arranz JA et al. (2002). "Splicing mutations, mainly IVS6-1(G>T), account for 70% of fumarylacetoacetate hydrolase (FAH) gene alterations, including 7 novel mutations, in a survey of 29 tyrosinemia type I patients." Hum Mutat. 20(3):180–8.**
3. **Bergman AJ et al. (1998). "Spectrum of mutations in the fumarylacetoacetate hydrolase gene of tyrosinemia type 1 patients in northwestern Europe and Mediterranean countries." Hum Mutat. 12(1):19–26.**
4. **Bliksrud YT et al. (2012). "Hereditary tyrosinaemia type I in Norway: Incidence and three novel small deletions in the fumarylacetoacetase gene." Scand J Clin Lab. Invest. 72(5):369–373.**
5. **De Braekeleer M et al. (1999). "Genetic epidemiology of hereditary tyrosinemia in Quebec and in Saguenay-Lac-St-Jean." Am J Hum Genet. 47(2):302–7.**
6. **Elpeleg ON et al. (2002). "Mutation analysis of the FAH gene in Israeli patients with tyrosinemia type I." Hum Mutat. 19(1):80–1.**
7. **Grompe M et al. (1994). "A single mutation of the fumarylacetoacetate hydrolase gene in French Canadians with hereditary tyrosinemia type I." N Engl J Med. 331(6):353–7.**
8. **Imtiaz F et al. (2011). "Identification of mutations causing hereditary tyrosinemia type I in patients of Middle Eastern origin." Mol Genet Metab. 104(4):688–90.**
9. **Ploos van Amstel JK et al. (1996). "Hereditary tyrosinemia type 1: novel missense, nonsense and splice consensus mutations in the human fumarylacetoacetate hydrolase gene; variability of the genotype-phenotype relationship." Hum Genet. 97(1):51–9.**
10. **Rootwelt H et al. (1994). "Novel splice, missense, and nonsense mutations in the fumarylacetoacetase gene causing tyrosinemia type 1." Am J Hum Genet. 55(4):653–8.**

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
March 2, 2018	Information specific to people of Norwegian descent was added. Customers who self-report having Norwegian ancestry may now see carrier frequency, carrier detection rate, and post-test carrier risk information specific to that ancestry.
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for one or more of the following genetic markers may see a genotype at these markers: i5012861, i5012865. This may also update the overall report result for these customers.
Oct. 21, 2015	Tyrosinemia Type I report created.