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.

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

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

0 variants detected in the FAH gene

---

### 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 149,000,000 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.

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.

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, and Turkish descent.

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

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

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
You have no variants detected by this test.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>W262X</td>
<td>G</td>
<td>Typical copy from one of your parents</td>
</tr>
<tr>
<td>Gene: FAH</td>
<td></td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Marker: ISO12862</td>
<td></td>
<td>Typical variant DNA sequence(s)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [8, 9, 11]</td>
</tr>
</tbody>
</table>

| P361L        | C              | Typical copy from one of your parents |
| Gene: FAH    |                | Biological explanation |
| Marker: ISO12861 |              | Typical variant DNA sequence(s) |
|              |                | Percent of 23andMe customers with variant |
|              |                | References [2, 4] | ClinVar |

| IV512+5G>A   | G              | Typical copy from one of your parents |
| Gene: FAH    |                | Biological explanation |
| Marker: ISO12865 |              | Typical variant DNA sequence(s) |
|              |                | Percent of 23andMe customers with variant |
|              |                | References [1, 2, 4, 6, 7, 11] | ClinVar |

| IV56-1G>T    | G              | Typical copy from one of your parents |
| Gene: FAH    |                | Biological explanation |
| Marker: ISO12867 |              | Typical variant DNA sequence(s) |
|              |                | Percent of 23andMe customers with variant |
|              |                | References [1, 2, 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

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of French Canadian, Ashkenazi Jewish, Finnish, European, and Turkish 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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk Factor</th>
<th>Example Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>French Canadian</td>
<td>1 in 200</td>
<td>[3]</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 149,000,000</td>
<td>[10]</td>
</tr>
<tr>
<td>Finnish</td>
<td>1 in 870</td>
<td>[10]</td>
</tr>
<tr>
<td>European</td>
<td>1 in 370</td>
<td>[10]</td>
</tr>
<tr>
<td>Turkish</td>
<td>1 in 210</td>
<td>[10]</td>
</tr>
</tbody>
</table>

View technical article on estimating post-test carrier risk.
Test Details

Indications for Use

The 23andMe PGx 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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>French Canadian</td>
<td>90%</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>&gt;99%</td>
</tr>
<tr>
<td>Finnish</td>
<td>88%</td>
</tr>
<tr>
<td>European</td>
<td>60%</td>
</tr>
<tr>
<td>Turkish</td>
<td>30%</td>
</tr>
</tbody>
</table>

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 196 samples with known variant status. 196 out of 196 genotype results were correct. Fewer than 1 in 100,000 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

10. Snidman King L et al. (1993). "Tyrosinemia Type I." 1