Familial Hyperinsulinism (ABCC8-Related)

ABCC8-related familial hyperinsulinism is a rare genetic disorder that is characterized by very high levels of insulin production. The defect is a mutation of the ABCC8 gene, which can cause near-lethal hunger, and even serious damage if untreated. People with ABCC8-related familial hyperinsulinism often have two variants in the ABCC8 gene.

`play-cd4dfe3c01`, you have one of the variants we tested.

You could pass this variant on to your children.

1 variant detected
in the ABCC8 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 Table
- See Scientific Details
- See Frequently Asked Questions

Intended Uses

- Tests for multiple variants in the ABCC8 gene.
- To identify carrier status for ABCC8-related familial hyperinsulinism.

Limitations

- Does not test for all possible variants for the condition.
- Does not detect changes in other genes such as A-RUN which are also associated with familial hyperinsulinism.

Important Ethnicities

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

You are a carrier.

You could pass this variant on to your children.

We detected one variant for ABCC8-related familial hyperinsulinism.

Your results may be relevant for you if you're thinking about starting a family.

If you and your partner are both carriers, each child has a 25% chance of having this condition. For male carriers, even if your partner is not a carrier, each child may still have a 50% chance of having the condition. For female carriers, you and your partner are not expected to be at risk of passing the condition unless your partner or your partner's family has a history of consideration testing if they plan to have children.

About Familial Hyperinsulinism

Also known as Congenital Hypoglycaemia, Persistent Hypoglycaemia Hypoglycaemia of Infancy (PHHI)

When it develops

Symptoms typically develop during infancy or in early childhood.

- High levels of insulin
- Low blood sugar
- Low energy
- Irritability
- Seizures
- Brain damage

Read more at: Genetic Alliance Reference® Geneticfinders®

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish, central European, and South Asian descent.

How it’s treated

There is currently no cure known. Treatment depends on the severity of the condition. Some people might need healthy blood glucose levels through medication or diet. Others may require surgery to remove part of the pancreas.

Consider talking to a healthcare professional if you are thinking about having children.

- If you're trying to have a family, genetic counseling can help you and your partner understand if additional testing might be appropriate.
- If you have other concerns about your results, consult a healthcare professional.

Learn more about other conditions and variants with support groups.

- See all Frequently Asked Questions for more information.
- Learn more about this condition and variants with support groups.

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ABC2-related familial hyperinsulinism is caused by variants in the ABC22 gene.

The ABC22 gene contains instructions for making a protein called sulfonate-amine reuptake protein 1. This protein is found in the pancreas and helps control the flow of a form of insulin that is released into the blood. Certain mutations in ABC22 can disrupt this function, resulting in a constant release of insulin and low blood sugar levels.

Read more at Genetic Home Reference®

You have one variant detected by this test.

<table>
<thead>
<tr>
<th>Chromosome 11</th>
<th>Gene - ABC22</th>
<th>Variants Detected</th>
<th>Variant Type</th>
<th>Position</th>
<th>Genotype</th>
<th>Reference</th>
<th>Evidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>11</td>
<td>ABC22</td>
<td>p.Thr321Met</td>
<td>Missense</td>
<td>321</td>
<td>c.959T&gt;C</td>
<td>1:1539</td>
<td>1:1539</td>
</tr>
</tbody>
</table>

**Indications for Use**

The 23andMe MGx® Carrier Status Report for Familial Hyperinsulinism (ABC22-Mediated) is indicated for the detection of these variants in the ABC22 gene. This test is intended to be used to determine carrier status for ABC22-related familial hyperinsulinism in adults, but cannot determine a person's risk for two copies of a tested variant. The report also displays if a result is associated with personal risk for the condition. It is recommended for use in individuals with a family history of this disease.

**Special Considerations**

- Symptoms of familial hyperinsulinism may vary between individuals with the condition even if they have the same genetic variant.
- There are many different professional guidelines in the U.S. for carrier testing for conditions. However, 23andMe's testing for familial hyperinsulinism is not intended for use in patients of Ashkenazi Jewish descent or in people with a history of having children with the condition.

**Test Performance Summary**

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified in a carrier detection test. This test has a lower detection rate than a disease-specific test because it is designed to detect all known variants that cause this condition.

- Adulthood Jewish: 87% (95)
- Entirely, particularly from central eastern: 4% (9)

**Analytical Performance**

Accuracy was determined by comparing results from the test with results from sequencing. Greater than 99% of known results were correct. While this study may provide positive or false negative results, more details on the analytical performance of this test can be found in 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 affordability is not covered by the Affordable Care Act may not cover the cost of genetic testing. Individuals in this situation should discuss genetic testing with a healthcare provider.
- There may be a requirement for you to see a healthcare professional for any medical issues.
- If you are concerned about your results, consult with a healthcare professional.

**Data Protection**

This is not genotypic data, nor does it involve personal health information, which 23andMe does not collect or use.

**References**


**Change Log**

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

**Date** | **Change**
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Mar 20, 2019 | Familial hyperinsulinism (ABC22-Mediated) report is updated.
Frequently Asked Questions

**ABCC6-related Familial Hypersensitivity**

This test looks for three genetic variants in the ABCC6 gene that are linked to familial hypersensitivity.

- People with two variants are at risk of developing symptoms of familial hypersensitivity, and they will mostly pass a variant on to each of their children.
- People with one variant may have a small degree of sensitivity if familial hypersensitivity has a genetic basis. They may also pass on a variant to their children.

This test does not include all possible genetic variants associated with familial hypersensitivity.

- Is this test useful? Yes

This test does not diagnose familial hypersensitivity. Only a healthcare professional can do that.

This test does not include all possible variants in the ABCC6 gene that are linked to familial hypersensitivity.

- Is this test useful? Yes

My variant was not selected. What does this mean?

This means you have one of the genetic variants tested, but you are not a carrier for familial hypersensitivity. You do not pass the variant on to each of your children. If your partner is a carrier for ABCC6-related familial hypersensitivity, each of your children may have a 50% chance of having the condition.

For males with this result, if your partner is not a carrier, each child will still have a small chance of having a form of familial hypersensitivity caused by familial hypercalciuria. One study in people of ethnicity [Asian] [Asian] showed that the risk is 1 in 500. If you have this result, if your partner is not a carrier, your children are not likely to have the condition.

Most people with your genetic result do not have familial hypersensitivity. However, a small fraction of people with the variant may develop familial hypercalciuria, typically by early adulthood. One study in people of [Asian] [Asian] showed that about 1 in 100 people who have a single variant from their mother may eventually develop familial hypercalciuria. People who inherit a single variant from their mother are not expected to develop familial hypercalciuria.

You cannot learn more about how a hereditary pattern for ABCC6-related familial hypersensitivity from the following resources:

- Genetic Home Reference
- GeneReviews

If you have questions about your results, a genetics counselor may be able to help. Learn more about genetic counseling.

- Is this test useful? Yes

My report says I have a small percentage of people with my genetic result may develop familial hypersensitivity. What does this suggest?

Most people with only one variant do not have familial hypersensitivity.

However, a small fraction of people with only one variant may develop a form of familial hypercalciuria, which affects part of the kidney. One study in people of [Asian] [Asian] showed that about 1 in 100 people who inherit a single variant from their father may eventually develop familial hypercalciuria. People who develop symptoms typically do so by early adulthood. People who inherit a single variant from their mother are not expected to develop familial hypercalciuria.

You can learn more about how a hereditary pattern for ABCC6-related familial hypersensitivity from the following resources:

- Genetic Home Reference
- GeneReviews

If you have questions about your results, a genetics counselor may be able to help. Learn more about genetic counseling.

- Is this test useful? Yes

My report says I have one variant linked to familial hypersensitivity. What can I know from this result?

Based on your genetic result, you could pass a variant on to each of your children. If you're starting a family, a genetics counselor can help you and your partner understand additional testing might be appropriate.

In addition, a small fraction of people with this result may develop a form of familial hypercalciuria caused by familial hypercalciuria. Consider talking to a healthcare professional about your result.

You may also want to share your results with your family. Because you share [DNA] [DNA] with your relatives, your genetic result could be relevant to your family members.

- Is this test useful? Yes

How will this result affect my children?

Between you and your partner, you could pass this variant on to each of your children. If your partner is a carrier for ABCC6-related familial hypersensitivity, each of your children may have a 50% chance of having the condition.

For males with this result, if your partner is not a carrier, each child will still have a small chance of having the condition. One study in people of [Asian] [Asian] showed that the risk is 1 in 500. If you have this result, if your partner is not a carrier, your children are not likely to have the condition.

You can learn more about how a hereditary pattern for ABCC6-related familial hypersensitivity from the following resources:

- Genetic Home Reference
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If you have questions about your results, a genetics counselor can help you and your partner understand additional testing might be appropriate. Learn more about genetic counseling.

- Is this test useful? Yes

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

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