Familial Hyperinsulinism (ABCC8-Related)

ABCC8-related familial hyperinsulinism is a rare genetic disorder. It is characterized by very high levels of insulin production. This leads to episodes of low blood sugar, which can cause lane energy, seizures, and brain damage if left untreated. People with ABCC8-related familial hyperinsulinism can have two variants in the ABCC8 gene.

play 4814b20d44, you do not have the variants we tested.

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

0 variants 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 the 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 ABCC8 gene.

• To identify individuals not at risk for ABCC8-related familial hyperinsulinism.

• To identify family members at risk for autosomal dominant hyperinsulinism.

Limitations

• Does not test for variants in other genes (such as KCNJ11 and GLUT2) that are also associated with familial hyperinsulinism.

Important Ethnities

• 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 ABCC8-related familial hyperinsulinism in people of Ashkenazi Jewish descent.

You still have a chance of being a carrier for ABCC8-related familial hyperinsulinism.

You may still have up to a 1 in 1,793 chance of carrying a variant that could cause disease in your kids.

See Scientific Details

About Familial Hyperinsulinism

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

When it develops

Symptoms typically develop during infancy or early childhood.

Typical signs and symptoms

• High levels of insulin

• Low blood sugar

• Seizures or fainting

• Fatigue

• Stomach pain

Read more at "Genetic Home Reference" GenetRef.org

Ethnicity most affected

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

How is it treated

There is currently no known cure. Treatment depends on the severity of the condition. Some people can manage their health with diet and medication. Others may need surgery to remove part of the pancreas.

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 Genetic Counselor

See your results with a healthcare professional.

Get support

Learn more about this condition and connect with support groups.

Take action

Receive up to ED when you share family and friends on ShareAlike. Our shared data is
ABCC8-related familial hyperinsulinism is caused by variants in the ABCC8 gene.

You have no variants detected by this test.

**Variant Details**

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>Gene</th>
<th>Variant Information</th>
<th>Exonic</th>
<th>Reference(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>310189 c.64G&gt;C</td>
<td>ABCC8</td>
<td>p.Asp21Leu</td>
<td>Yes</td>
<td>none</td>
</tr>
</tbody>
</table>

**Text Interpretation**

**Post-Test Carrier Risk**

This report provides an estimate of the risk for carrier risk for people of Armenian descent.

- For people with similar ancestry who are not carrier risk for the condition.
- The test result is not actionable.

**Warnings and Limitations**

- This test does not cause or cure the condition.
- This test does not diagnose or manage the condition.
- Results indicate a carrier status and do not provide information on whether the individual has the condition.
- Results may vary by ethnicity and other factors.

**Test Details**

**Indications for Use**

- This test is performed to screen for variants in the ABCC8 gene that cause familial hyperinsulinism.
- People with a personal or family history of the condition are eligible for testing.

**Test Relevance**

- The test can help determine if someone is a carrier of the condition.
- Results can inform reproductive decisions.

**References**


**Analysis/Phenotype**

Access to clinical care is determined by results from this test.

**Change Log**

Your report may include updated clinical background information. The Change Log describes updates and revisions to the report.