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 low energy, seizures, and brain damage if left untreated. People with ABCC8-related familial hyperinsulinism often have two variants in the ABCC8 gene.

Your result for this test cannot be determined.

We may not always be able to report a result for this test. This can happen if there is a test error or if a person has two copies of a variant tested.

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 ABCC8 gene.
- To identify carrier status for ABCC8-related familial hyperinsulinism.

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 variants in other genes (such as KCNJ11) that are also associated with familial hyperinsulinism.

Important Ethnicities

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

About Familial Hyperinsulinism

Also known as: Congenital Hyperinsulinism, Persistent Hyperinsulinemiac Hypoglycemia of Infancy (PHHI)

When it develops

Symptoms typically develop during infancy or in early childhood.

Typical signs and symptoms

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

Read more at: Genetics Home Reference® GeneReviews®

How it's treated

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

Consider talking to a healthcare professional if you are concerned about this report.

If you think you might have symptoms or if this condition runs in your family, consult with a healthcare professional.

Print report

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

Learn more about this condition and connect with support groups.

Learn more