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 most often have two variants in the ABCC8 gene.

You are at risk for developing symptoms of ABCC8-related familial hyperinsulinism. Your risk may also be relevant if you're considering having children.

How To Use This Test

This test does not diagnose ABCC8-related familial hyperinsulinism. If the result is unexpected, please discuss this report with a healthcare professional.

Play dr46bc644bb, you have two of the variants we tested. You are at risk for developing symptoms of familial hyperinsulinism. Your result may also be relevant if you're considering having children.

Intended Use

- 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 KPNB1 that are also associated with familial hyperinsulinism.

Important Ethnicities

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

You are at risk for developing symptoms of familial hyperinsulinism. Your result may also be relevant if you're considering having children.

About Familial Hyperinsulinism

Also known as Congenital Hyperinsulinism, Persistent Hyperinsulinism, Hyperglycemia of Infancy (PHI), Type 2 Diabetes.

When it develops

Symptoms usually develop during infancy or in early childhood.

Typical signs and symptoms

- High levels of insulin
- Low blood sugar
- Low energy
- Inability to sleep
- Seizures
- Brain damage

Matters most affected

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

How it's treated

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

Read more at: [Genetic Home Reference](https://ghr.nimh.nih.gov/condition/familial-hyperinsulinism)

It is important to talk to a healthcare professional if you are concerned about your results.

You are at risk for developing symptoms of familial hyperinsulinism. It is important to consult with a healthcare professional about your result.

If you're looking for genetic counseling or to help you and your partner understand if additional testing might be appropriate.

Learn more about this condition and connect with support groups.

See Frequently Asked Questions for more information.
ABCBC8-related familial hypereumismus is caused by variants in the ABCC8 gene.

You have two variants detected by this test.

Test Details

Indications for Use

The Cardio-REL Preventive Cardiomyopathy Risk Evaluation (REL PREVENT) is indicated for the detection of two rare genetic defects in the ABCBC8 gene that may be associated with familial hypereumismus. These defects are called “variants of unknown significance” and are determined by providing only sufficient data to establish whether the variant is likely to be pathogenic.

Test Procedure

CardioGen Chip & Gene Analysis.

The “carrier detection test” is in essence a test of the percentage of carriers of the condition that can be identified. This is done by determining the carrier status of the child and using the information obtained to provide reliable and sufficiently accurate data on the prevalence of the condition.

Analysis Progress

Pregnancy was determined by the results from the test results from sequencing. The PGE2 levels were measured in all cases. The test may be performed at a lower level of accuracy, with the results of the performance analysis of the test being used to assess the possible need for additional testing.

References

Frequently Asked Questions

ARCCX-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 loss of energy, weakness, and even loss of consciousness. People with ARCCX-related familial hyperinsulinism are at risk for early-onset diabetes. Some have been diagnosed with diabetes before birth. The symptoms of this condition may vary from one family to another. It is important to talk with a healthcare professional if you are concerned about your results.

Is this answer helpful? Yes No

What does this test do? This test looks for three genetic variants in the ARCCX gene that are linked to familial hyperinsulinism. People with two variants are at risk of developing symptoms of familial hyperinsulinism, and they are most likely to pass a variant on to each of their children. People with one variant may have a small chance of developing from hyperinsulinism called homozygous hyperinsulinism. They may also pass a variant on to their children. This test does not include all possible genetic variants associated with familial hyperinsulinism. Is this answer helpful? Yes No

What does this test not do? This test does not diagnose familial hyperinsulinism. Only a healthcare professional can do that. This test does not include all possible variants in the ARCCX gene that are linked to familial hyperinsulinism. This test does not include variants in other genes such as KCNJ11 that are linked to familial hyperinsulinism. Is this answer helpful? Yes No

My report says two variants were detected. What does this mean? This means you have two of the genetic variants we tested. You are most likely to pass a variant on to each of your parents. People with this mutation may be at risk for developing symptoms of familial hyperinsulinism. It is important to talk with a healthcare professional if you are concerned about your result. This result does mean you will most likely pass a variant on to each of your children. Is this answer helpful? Yes No

What does this mean for developing symptoms of familial hyperinsulinism? Symptoms of familial hyperinsulinism include excessive muscle production, low blood sugar, low energy levels, seizures, and possibly brain damage. If this condition is left untreated, people with this mutation may be at risk for developing symptoms of familial hyperinsulinism. This result does mean you will most likely pass a variant on to each of your children. Is this answer helpful? Yes No

The report says this test is most relevant for people of Ashkenazi Jewish decent. What if I’m not of Ashkenazi Jewish decent? The genetic variants are treated most commonly in people of Ashkenazi Jewish decent, but they may also be found in people of other ethnicities. If you have two variants, you are at risk for developing symptoms of familial hyperinsulinism. In addition, you will still most likely pass a variant on to each of your children. Is this answer helpful? Yes No

My report says I have two variants linked to familial hyperinsulinism. What are some things I could do? Based on your genetic result, you are at risk for developing symptoms of familial hyperinsulinism. It is important to talk to a healthcare professional if you are concerned about your result. If your family has a genetic counselor, they can help you and your partner understand if additional testing might be appropriate. You may also want to discuss your results with other family members. Because you share DNA with your relatives, your genetic result could also be relevant for them. Is this answer helpful? Yes No

How could my result affect my children? Because you have two variants, you are most likely to pass a variant on to each of your children. If your partner is a carrier for familial hyperinsulinism, each of your children is likely to have 50% chance of having the condition. For mode with this result, if your partner is not a carrier each child may still have a small chance of having the condition. (One study in people of Ashkenazi Jewish decent estimated that the risk is 1 in 230.) If you are not sure if your partner is not a carrier, your children are most likely at risk of having the condition. You can learn more about the best care plan for ARCCX-related familial hyperinsulinism in the following resources:

1. Genetic Home Reference
2. GeneticFacts

A genetic counselor can help you and your partner understand if additional testing might be appropriate. Learn more about genetic counseling. Is this answer helpful? Yes No

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