Print

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.

play+d4b6c64fbb, 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.

2 variants detected
in the ABCC8 gene

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

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
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 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.

You are at risk for developing symptoms of familial hyperinsulinism.

Your result may also be relevant if you're considering having children.

Talk to a healthcare professional.

People with your result are at risk for developing symptoms of familial hyperinsulinism. A healthcare professional can answer questions you may have about your results. See Frequently Asked Questions for more information.

Your results may be relevant for your family.

Because you have two variants, you will most likely pass a variant on to each of your children. **If your partner is a carrier** for ABCC8-related familial hyperinsulinism, each child most likely has a **50% chance** of having this condition. For males with this result, even if your partner does not have a variant, each child may still have a small chance (less than 1%) of having the condition. For females with this result, your children are not expected to be at risk of having the condition unless your partner has a variant. Your relatives may also wish to consider testing if they plan to have children. See Frequently Asked Questions for more information.

About Familial Hyperinsulinism

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

When it develops

Symptoms typically develop during infancy or in early childhood.

Ethnicities most affected

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

Typical signs and symptoms

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

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.

Read more at: Genetics Home Reference GeneReviews

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.

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

See our Frequently Asked Questions for more information.

FAQs

Learn more about this condition and connect with support groups.

Learn more

Familial Hyperinsulinism (ABCC8-Related)

Scientific Details

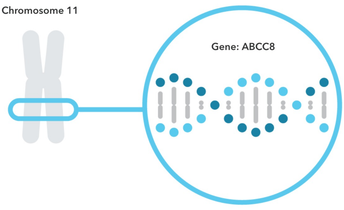
ABCC8-related familial hyperinsulinism is a rare genetic disorder. It is characterized by very high levels of insulin production. This leads to episodes of low blood sugar, which can cause low energy, seizures, and brain damage if left untreated. People with ABCC8-related familial hyperinsulinism most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

ABCC8-related familial hyperinsulinism is caused by variants in the ABCC8 gene.

ABCC8

The ABCC8 gene contains instructions for making a protein called sulfonylurea receptor 1. This protein is found in the pancreas and helps control the amount of insulin that is released into the blood. Certain variants in ABCC8 disrupt this function, resulting in a constant release of insulin and low blood sugar levels.

Read more at Genetics Home Reference



You have two variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*			Additional Information
F1388del **Gene:** ABCC8 **Marker:** rs151344624	(–) Variant copy from one of your parents		**GAA** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 5, 7] | ClinVar
3992-9G>A **Gene:** ABCC8 **Marker:** rs151344623	**C** Typical copy from one of your parents		**T** Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 5, 7, 9] | 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 Details

Indications for Use

The 23andMe PGS Carrier Status Report for Familial Hyperinsulinism (ABCC8-Related) is indicated for the detection of three variants in the ABCC8 gene. This test is intended to be used to determine carrier status for ABCC8-related familial hyperinsulinism in adults, but cannot determine if a person has two copies of a tested variant. This report also describes if a result is associated with personal risk for developing symptoms of ABCC8-related familial hyperinsulinism, but it does not describe a person's overall risk of developing symptoms. This test is most relevant for people of Ashkenazi Jewish descent.

Special Considerations

- Symptoms of familial hyperinsulinism may vary between people with the condition even if they have the same genetic variants.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition. However, ACOG notes that testing for familial hyperinsulinism may be considered for people of Ashkenazi Jewish descent who are considering having children.

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.

Ashkenazi Jewish	97%	[5]
Finnish, particularly from central Finland	41%	[8]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to 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 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

1. Cartier EA et al. (2001). "Defective trafficking and function of KATP channels caused by a sulfonylurea receptor 1 mutation associated with persistent hyperinsulinemic hypoglycemia of infancy." Proc Natl Acad Sci U S A. 98(5):2882-7.
2. Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.
3. Dunne MJ et al. (2004). "Hyperinsulinism in infancy: from basic science to clinical disease." Physiol Rev. 84(1):239-75.
4. Glaser B et al. (2003). "Familial Hyperinsulinism." [Updated 2013 Jan 24]
5. Glaser B et al. (2011). "ABCC8 mutation allele frequency in the Ashkenazi Jewish population and risk of focal hyperinsulinemic hypoglycemia." Genet Med. 13(10):891-4.
6. Huopio H et al. (2002). "Acute insulin response tests for the differential diagnosis of congenital hyperinsulinism." J Clin Endocrinol Metab. 87(10):4502-7.
7. Nestorowicz A et al. (1996). "Mutations in the sulonylurea receptor gene are associated with familial hyperinsulinism in Ashkenazi Jews." Hum Mol Genet. 5(11):1813-22.
8. Otonkoski T et al. (1999). "A point mutation inactivating the sulfonylurea receptor causes the severe form of persistent hyperinsulinemic hypoglycemia of infancy in Finland." Diabetes. 48(2):408-15.
9. Thomas PM et al. (1995). "Mutations in the sulfonylurea receptor gene in familial persistent hyperinsulinemic hypoglycemia of infancy." Science. 268(5209):426-9.

Change Log

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

Date	Change
March 23, 2018	Familial Hyperinsulinism (ABCC8-Related) report created.

Familial Hyperinsulinism (ABCC8-Related)

Frequently Asked Questions

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.

ABCC8-Related Familial Hyperinsulinism

What does this test do?

This test looks for three genetic variants in the ABCC8 gene that are linked to familial hyperinsulinism.

People with two variants are at risk of developing symptoms of familial hyperinsulinism, and they will most likely pass a variant on to each of their children.

People with one variant may have a small chance of developing a form of hyperinsulinism called focal 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 ABCC8 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 most likely inherited one variant from each of your parents.

People with this result are at risk for developing symptoms of familial hyperinsulinism. It's important to talk with a healthcare professional if you are concerned about your result.

This result also means you will most likely pass a variant on to each of your children.

Is this answer helpful? Yes No

What does **at risk for developing symptoms of familial hyperinsulinism** mean?

Symptoms of familial hyperinsulinism include excessive insulin production, low blood sugar, low energy, irritability, seizures, and possible brain damage, if the condition is left untreated.

People with your genetic result are at risk for developing these symptoms. Most people who develop symptoms do so by early adulthood.

A healthcare professional can answer any questions you may have about your results.

Is this answer helpful? Yes No

The report says the test is most relevant for people of **Ashkenazi Jewish** descent. What if I'm not of Ashkenazi Jewish descent?

The genetic variants we tested are most common in people of Ashkenazi Jewish descent, but they are also found in people of other ethnicities. Because you have two variants, you are still 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 you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

You may also want to share 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 will most likely pass a variant on to each of your children. If your partner is a carrier for familial hyperinsulinism, each of your children most likely has a 50% chance of having this condition.

For males 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 descent estimated that the risk is 1 in 270.) For females with this result, if your partner is not a carrier, your children are not likely at risk of having the condition.

You can learn more about the inheritance pattern for ABCC8-related familial hyperinsulinism from the following resources:

- Genetics Home Reference
- GeneReviews

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.