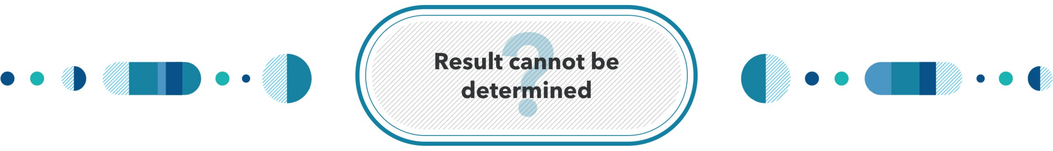
Familial Hyperinsulinism (ABCC8-Related)

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.

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.



If you are concerned about this report, please consult with a healthcare professional about additional testing.

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 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

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

Familial Hyperinsulinism (ABCC8-Related)

Scientific Details

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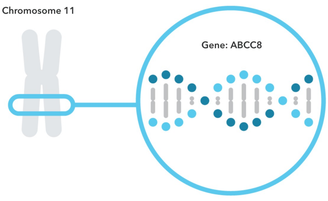
ABCC8-related familial hyperinsulinism is caused by variants in the ABCC8 gene.

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



Your result cannot be determined.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*	Additional Information
F1388del **Gene:** ABCC8 **Marker:** rs151344624	**Not determined**	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 **C** Typical 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
V187D **Gene:** ABCC8 **Marker:** rs137852672	**A** Typical copy from one of your parents **A** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [6, 8] | 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.