Clarity Genetics

Genetics Uncoded:

Facts about

ABCC8-Related Hyperinsulinism

What Your Test Results Mean

Some individuals with one *ABCC8* variant show no symptoms of *ABCC8*-related hyperinsulinism while others have symptoms ranging from congenital hyperinsulinism to type 2 diabetes mellitus in adulthood. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

ABCC8-Related Hyperinsulinism Explained

ABCC8-related hyperinsulinism is an inherited disorder characterized by hypoglycemia, or low blood sugar. Individuals with ABCC8-related hyperinsulinism release insulin into the bloodstream even in the absence of glucose. The low level of sugar in the blood increases the risk for seizures and brain damage and can ultimately lead to death.

Treatment for individuals with ABCC8-related hyperinsulinism includes glucose infusions as well as other medications and frequent eating to prevent complications associated with hypoglycemia. In some cases it may be necessary to surgically remove most of the pancreas. Even with treatment, some individuals may have some degree of brain damage or learning disability.

How the Genetics Work

Pathogenic variants in the *ABCC8* gene are associated with *ABCC8*-related hyperinsulinism. In general, individuals have two copies of the *ABCC8* gene. Individuals with *ABCC8*-related hyperinsulinism typically have variants in both copies of their *ABCC8* genes, one inherited from each parent. In addition to the autosomal recessive form, *ABCC8*-related hyperinsulinism caused by a single *ABCC8* variant, E1506K, has been associated with symptoms ranging from congenital hyperinsulinism to type 2 diabetes mellitus in adulthood.

Questions?

Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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