Clarity Genetics

Genetics Uncoded:

Facts about Pseudocholinesterase Deficiency

What Your Test Results Mean

Carriers typically show no symptoms of pseudocholinesterase deficiency; however, carriers are at an increased risk of having a child with pseudocholinesterase deficiency. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Pseudocholinesterase Deficiency Explained

Pseudocholinesterase deficiency is an inherited condition that affects the body's ability to metabolize choline ester drugs. Choline ester drugs are muscle relaxants that temporarily relax and paralyze the skeletal muscles and muscles used for breathing; these are usually used to facilitate the placement of breathing tubes. Individuals with the disorder are unable to metabolize choline ester drugs efficiently and after a normal dose of a choline ester drug will be paralyzed for up to several hours. Individuals with psuedocholinesterase deficiency must be closely monitored and supported by mechanical ventilation any time a choline ester drug is administered. No other symptoms are associated with this condition.

How the Genetics Work

Pseudocholinesterase deficiency is an autosomal recessive disorder caused by pathogenic variants in the *BCHE* gene. In general, individuals have two copies of the BCHE gene. Carriers of pseudocholinesterase deficiency have a single variant in one copy of the *BCHE* gene while individuals with pseudocholinesterase deficiency have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

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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