**Genetics Uncoded:**

**FACTS ABOUT PSEUDOCHOLINESTERASE DEFICIENCY**

*AKA Butyrylcholinesterase deficiency, Cholinesterase II deficiency, Deficiency of butyrylcholine esterase, Pseudocholinesterase E1 deficiency, Succinylcholine sensitivity, Suxamethonium sensitivity*

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**What Your Test Results Mean**

Testing results indicate that you are a carrier of pseudocholinesterase deficiency. Carriers typically show no symptoms of pseudocholinesterase deficiency; however, carriers are at an increased risk of having a child with pseudocholinesterase deficiency. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

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**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 pseudocholinesterase 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.

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**How the Genetics Work**

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

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**Recommended Next Steps**

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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**Questions?**

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.