

Pseudocholinesterase Deficiency

What Your Results Mean

Test results indicate that you are a carrier of pseudocholinesterase deficiency. Some carriers can show symptoms, while others show no symptoms at all. Carriers also have an increased chance to have a child with pseudocholinesterase deficiency; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.



Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for pseudocholinesterase deficiency, each of your children has a 1 in 4 (25%) chance or greater to have the condition.

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What is Pseudocholinesterase Deficiency?

Pseudocholinesterase deficiency is an inherited condition that causes an increased sensitivity to certain muscle relaxant drugs used during general anesthesia, known as choline esters. These fast-acting drugs, such as succinylcholine and mivacurium, are given to relax the muscles during surgical procedures or in emergencies when a breathing tube must be inserted quickly. Normally, these drugs are broken down (metabolized) by the body within a few minutes of being administered, allowing the muscles to move again. However, people with pseudocholinesterase deficiency may not be able to move or breathe on their own for a few hours after the drugs are administered. People with pseudocholinesterase deficiency may also have increased sensitivity to certain other drugs, including the local anesthetic procaine, and to specific agricultural pesticides.



Prognosis

Prognosis is excellent with early diagnosis and treatment.

Treatment

There is no cure for pseudocholinesterase deficiency. Affected individuals must be supported with a machine to help them breathe (mechanical ventilation) until the choline ester drugs are cleared from the body. Doctors may also consider using other types of muscle relaxants.



Resources

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/pseudocholinesterase-deficiency>

National Society of Genetic Counselors

<https://www.nsgc.org/>