# Pseudocholinesterase deficiency

## Description

Pseudocholinesterase deficiency is a condition that results in increased sensitivity to certain muscle relaxant drugs used during general anesthesia, called choline esters. These fast-acting drugs, such as succinylcholine and mivacurium, are given to relax the muscles used for movement (skeletal muscles), including the muscles involved in breathing. The drugs are often employed for brief 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, at which time the muscles can 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. Affected individuals must be supported with a machine to help them breathe (mechanical ventilation) until the drugs are cleared from the body.

People with pseudocholinesterase deficiency may also have increased sensitivity to certain other drugs, including the local anesthetic procaine, and to specific agricultural pesticides. The condition causes no other signs or symptoms and is usually not discovered until an abnormal drug reaction occurs.

## Frequency

Pseudocholinesterase deficiency occurs in 1 in 3,200 to 1 in 5,000 people. It is more common in certain populations, such as the Persian Jewish community and Alaska Natives.

## Causes

Pseudocholinesterase deficiency can be caused by mutations in the *BCHE* gene. This gene provides instructions for making the pseudocholinesterase enzyme, also known as butyrylcholinesterase, which is produced by the liver and circulates in the blood. The pseudocholinesterase enzyme is involved in the breakdown of choline ester drugs. It is likely that the enzyme has other functions in the body, but these functions are not well understood. Studies suggest that the enzyme may be involved in the transmission of nerve signals.

Some *BCHE* gene mutations that cause pseudocholinesterase deficiency result in an abnormal pseudocholinesterase enzyme that does not function properly. Other

mutations prevent the production of the pseudocholinesterase enzyme. A lack of functional pseudocholinesterase enzyme impairs the body's ability to break down choline ester drugs efficiently, leading to abnormally prolonged drug effects.

Pseudocholinesterase deficiency can also have nongenetic causes. In these cases, the condition is called acquired pseudocholinesterase deficiency; it is not inherited and cannot be passed to the next generation. Activity of the pseudocholinesterase enzyme can be impaired by kidney or liver disease, malnutrition, major burns, cancer, or certain drugs.

Learn more about the gene associated with Pseudocholinesterase deficiency

- BCHE

## Inheritance

When due to genetic causes, this condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive disorder have one copy of the altered gene in each cell and are called carriers. They can pass on the gene mutation to their children, but they do not usually experience signs and symptoms of the disorder. In some cases, carriers of *BCHE* gene mutations take longer than usual to clear choline ester drugs from the body, but not as long as those with two copies of the altered gene in each cell.

## Other Names for This Condition

- Butyrylcholinesterase deficiency
- Cholinesterase II deficiency
- Deficiency of butyrylcholine esterase
- Pseudocholinesterase E1 deficiency
- Succinylcholine sensitivity
- Suxamethonium sensitivity

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deficiency of butyrylcholinesterase (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1283400/)

Genetic and Rare Diseases Information Center

- Butyrylcholinesterase deficiency (https://rarediseases.info.nih.gov/diseases/7482/in

dex)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Pseudocholinesterase deficiency%22)

Catalog of Genes and Diseases from OMIM

- BUTYRYLCHOLINESTERASE; BCHE (https://omim.org/entry/177400)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28pseudocholinesterase+deficiency%5BTIAB%5D%29+OR+%28butyrylcholinesterase+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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