



## Pseudocholinesterase deficiency

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.

## **Inheritance Pattern**

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

## **Diagnosis & Management**

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Deficiency of butyrylcholine esterase  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1283400/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22pseudocholinesterase+deficiency%22>

### Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Cholinesterase (blood test)  
<https://medlineplus.gov/ency/article/003358.htm>

## **Additional Information & Resources**

### Health Information from MedlinePlus

- Encyclopedia: Anesthesia -- What to Ask Your Doctor  
<https://medlineplus.gov/ency/patientinstructions/000183.htm>
- Encyclopedia: Cholinesterase (blood test)  
<https://medlineplus.gov/ency/article/003358.htm>
- Health Topic: Metabolic Disorders  
<https://medlineplus.gov/metabolicdisorders.html>

### Genetic and Rare Diseases Information Center

- Pseudocholinesterase deficiency  
<https://rarediseases.info.nih.gov/diseases/7482/pseudocholinesterase-deficiency>

### Additional NIH Resources

- National Institute of General Medical Sciences: Understanding Anesthesia  
<https://www.nigms.nih.gov/education/Inside-Life-Science/Pages/understanding-anesthesia.aspx>
- News in Health: Waking Up to Anesthesia  
<https://newsinhealth.nih.gov/2011/04/waking-up-anesthesia>

### Educational Resources

- MalaCards: butyrylcholinesterase deficiency  
[https://www.malacards.org/card/butyrylcholinesterase\\_deficiency](https://www.malacards.org/card/butyrylcholinesterase_deficiency)
- Orphanet: Butyrylcholinesterase deficiency  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=132](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=132)
- Royal (UK) College of Anaesthetists: Information about Anaesthesia  
<https://www.rcoa.ac.uk/patientinfo>

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/pseudocholinesterase-deficiency/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28pseudocholinesterase+d+eficiency%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>

## Catalog of Genes and Diseases from OMIM

- BUTYRYLCHOLINESTERASE  
<http://omim.org/entry/177400>

### Sources for This Summary

- Garcia DF, Oliveira TG, Molfetta GA, Garcia LV, Ferreira CA, Marques AA, Silva WA Jr. Biochemical and genetic analysis of butyrylcholinesterase (BChE) in a family, due to prolonged neuromuscular blockade after the use of succinylcholine. *Genet Mol Biol*. 2011 Jan;34(1):40-4. doi: 10.1590/S1415-47572011000100008. Epub 2011 Mar 1.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21637541>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3085371/>
- Gätke MR, Bundgaard JR, Viby-Mogensen J. Two novel mutations in the BCHE gene in patients with prolonged duration of action of mivacurium or succinylcholine during anaesthesia. *Pharmacogenet Genomics*. 2007 Nov;17(11):995-9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18075469>
- Leadingham CL. A case of pseudocholinesterase deficiency in the PACU. *J Perianesth Nurs*. 2007 Aug;22(4):265-71; quiz 272-4. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17666297>
- Levano S, Ginz H, Siegemund M, Filipovic M, Voronkov E, Urwyler A, Girard T. Genotyping the butyrylcholinesterase in patients with prolonged neuromuscular block after succinylcholine. *Anesthesiology*. 2005 Mar;102(3):531-5.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15731589>
- Maiorana A, Roach RB Jr. Heterozygous pseudocholinesterase deficiency: a case report and review of the literature. *J Oral Maxillofac Surg*. 2003 Jul;61(7):845-7. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12856264>
- Soliday FK, Conley YP, Henker R. Pseudocholinesterase deficiency: a comprehensive review of genetic, acquired, and drug influences. *AANA J*. 2010 Aug;78(4):313-20. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20879632>
- Yen T, Nightingale BN, Burns JC, Sullivan DR, Stewart PM. Butyrylcholinesterase (BCHE) genotyping for post-succinylcholine apnea in an Australian population. *Clin Chem*. 2003 Aug;49(8):1297-308.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12881446>
- Zelinski T, Coghlan G, Mauthe J, Triggs-Raine B. Molecular basis of succinylcholine sensitivity in a prairie Hutterite kindred and genetic characterization of the region containing the BCHE gene. *Mol Genet Metab*. 2007 Feb;90(2):210-6. Epub 2006 Dec 12.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17166756>
- Zencirci B. Pseudocholinesterase enzyme deficiency: a case series and review of the literature. *Cases J*. 2009 Dec 4;2:9148. doi: 10.1186/1757-1626-2-9148.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20062665>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2803945/>

---

Reprinted from Genetics Home Reference:

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

Reviewed: April 2012

Published: November 12, 2019

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services