



[Home](#) → [Medical Encyclopedia](#) → Congenital antithrombin III deficiency

URL of this page: [//medlineplus.gov/ency/article/000558.htm](https://medlineplus.gov/ency/article/000558.htm)

Congenital antithrombin III deficiency

Congenital antithrombin III deficiency is a genetic disorder that causes the blood to clot more than normal.

Causes

Antithrombin III is a protein in the blood that blocks abnormal blood clots from forming. It helps the body keep a healthy balance between bleeding and clotting. Congenital antithrombin III deficiency is an inherited disease. It occurs when a person receives one abnormal copy of the antithrombin III gene from a parent with the disease.

The abnormal gene leads to a low level of the antithrombin III protein. This low level of antithrombin III can cause abnormal blood clots (thrombi) that can block blood flow and damage organs.

People with this condition will often have blood clots at a young age. They are also likely to have family members who have had a blood clotting problem.

Symptoms

People will usually have symptoms of a blood clot. Blood clots in the arms or legs usually cause swelling, redness, and pain. When a blood clot breaks off from where it formed and travels to another part of the body, it is called a thromboembolism. Symptoms depend on where the blood clot travels to. A common place is the lung, where the clot can cause a cough, shortness of breath, pain while taking deep breaths, chest pain, and even death. Blood clots that travel to the brain can cause a stroke.

Exams and Tests

A physical exam may show:

- A swollen leg or arm
- Decreased breath sounds in the lungs
- A rapid heart rate

The health care provider can also order a blood test to check if you have a low level of antithrombin III.

Treatment

A blood clot is treated with blood-thinning medicines (also called anticoagulants). How long you need to take these drugs depends on how serious the blood clot was and other factors. Discuss this with your provider.

Support Groups

More information and support for people with congenital antithrombin III deficiency and their families can be found at:

- National Organization for Rare Disorders -- rarediseases.org/rare-diseases/antithrombin-deficiency/ [<https://rarediseases.org/rare-diseases/antithrombin-deficiency/>]
- MedlinePlus -- medlineplus.gov/genetics/condition/hereditary-antithrombin-deficiency/ [<https://medlineplus.gov/genetics/condition/hereditary-antithrombin-deficiency/>]
- National Blood Clot Alliance -- www.stoptheclot.org/programs-services/ [<https://www.stoptheclot.org/programs-services/>]

Outlook (Prognosis)

Most people have a good outcome if they stay on anticoagulant medicines.

Possible Complications

Blood clots can cause death. Blood clots in the lungs are very dangerous.

When to Contact a Medical Professional

See your provider if you have symptoms of this condition.

Prevention

Once a person is diagnosed with antithrombin III deficiency, all close family members should be screened for this disorder. Blood thinning drugs can prevent blood clots from forming and prevent complications from clotting.

Alternative Names

Deficiency - antithrombin III - congenital; Antithrombin III deficiency - congenital

References

Anderson JAM, Weitz JI. Hypercoagulable states. In: Hoffman R, Benz EJ, Silberstein LE, Heslop HE, Weitz JI, Salama ME, et al, eds. *Hematology: Basic Principles and Practice*. 8th ed. Philadelphia, PA: Elsevier; 2023:chap 138.

Mhyre JM. Hematologic and coagulation disorders. In: Chestnut DH, Wong CA, Tsen LC, et al, eds. *Chestnut's Obstetric Anesthesia: Principles and Practice*. 6th ed. Philadelphia, PA: Elsevier; 2020:chap 44.

Schafer AI. Thrombotic disorders: hypercoagulable states. In: Goldman L, Schafer AI, eds. *Goldman-Cecil Medicine*. 26th ed. Philadelphia, PA: Elsevier; 2020:chap 73.

Review Date 2/2/2023

Updated by: Mark Levin, MD, Hematologist and Oncologist, Monsey, NY. Review provided by VeriMed Healthcare Network. Also reviewed by David C. Dugdale, MD, Medical Director, Brenda Conaway, Editorial Director, and the A.D.A.M. Editorial team.

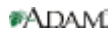
Learn how to cite this page



Health Content
Provider
06/01/2028

A.D.A.M., Inc. is accredited by [URAC](http://www.urac.org), for Health Content Provider (www.urac.org). URAC's [accreditation program](#) is an independent audit to verify that A.D.A.M. follows rigorous standards of quality and accountability. A.D.A.M. is among the first to achieve this important distinction for online health information and services. Learn more about A.D.A.M.'s [editorial policy](#), [editorial process](#), and [privacy policy](#).

The information provided herein should not be used during any medical emergency or for the diagnosis or treatment of any medical condition. A licensed medical professional should be consulted for diagnosis and treatment of any and all medical conditions. Links to other sites are provided for information only – they do not constitute endorsements of those other sites. No warranty of any kind, either expressed or implied, is made as to the accuracy, reliability, timeliness, or correctness of any translations made by a third-party service of the information provided herein into any other language. © 1997-2025 A.D.A.M., a business unit of Ebix, Inc. Any duplication or distribution of the information contained herein is strictly prohibited.



National Library of Medicine 8600 Rockville Pike, Bethesda, MD 20894 U.S. Department of Health and Human Services
National Institutes of Health