Hereditary Antithrombin Deficiency

Subjects: Genetics & Heredity

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Hereditary antithrombin deficiency is a disorder of blood clotting. People with this condition are at higher than average risk for developing abnormal blood clots, particularly a type of clot that occurs in the deep veins of the legs. This type of clot is called a deep vein thrombosis (DVT).

genetic conditions

1. Introduction

Affected individuals also have an increased risk of developing a pulmonary embolism (PE), which is a clot that travels through the bloodstream and lodges in the lungs. In hereditary antithrombin deficiency, abnormal blood clots usually form only in veins, although they may rarely occur in arteries.

About half of people with hereditary antithrombin deficiency will develop at least one abnormal blood clot during their lifetime. These clots usually develop after adolescence.

Other factors can increase the risk of abnormal blood clots in people with hereditary antithrombin deficiency. These factors include increasing age, surgery, or immobility. The combination of hereditary antithrombin deficiency and other inherited disorders of blood clotting can also influence risk. Women with hereditary antithrombin deficiency are at increased risk of developing an abnormal blood clot during pregnancy or soon after delivery. They also may have an increased risk for pregnancy loss (miscarriage) or stillbirth.

2. Frequency

Hereditary antithrombin deficiency is estimated to occur in about 1 in 2,000 to 3,000 individuals. Of people who have experienced an abnormal blood clot, about 1 in 20 to 200 have hereditary antithrombin deficiency.

3. Causes

Hereditary antithrombin deficiency is caused by mutations in the *SERPINC1* gene. This gene provides instructions for producing a protein called antithrombin (previously known as antithrombin III). This protein is found in the bloodstream and is important for controlling blood clotting. Antithrombin blocks the activity of proteins that promote blood clotting, especially a protein called thrombin.

Most of the mutations that cause hereditary antithrombin deficiency change single protein building blocks (amino acids) in antithrombin, which disrupts its ability to control blood clotting. Individuals with this condition do not have enough functional antithrombin to inactivate clotting proteins, which results in the increased risk of developing abnormal blood clots.

3.1. The gene associated with Hereditary antithrombin deficiency

- SERPINC1

4. Inheritance

Hereditary antithrombin deficiency is typically inherited in an autosomal dominant pattern, which means one altered copy of the *SERPINC1* gene in each cell is sufficient to cause the disorder. Inheriting two altered copies of this gene in each cell is usually incompatible with life; however, a few severely affected individuals have been reported with mutations in both copies of the *SERPINC1* gene in each cell.

5. Other Names for This Condition

- Antithrombin III Deficiency
- Congenital Antithrombin III Deficiency

References

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