Protein C Deficiency

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Protein C deficiency is a disorder that increases the risk of developing abnormal blood clots; the condition can be mild or severe.

genetic conditions

1. Introduction

Individuals with mild protein C deficiency are at risk of a type of blood clot known as a deep vein thrombosis (DVT). These clots occur in the deep veins of the arms or legs, away from the surface of the skin. A DVT can travel through the bloodstream and lodge in the lungs, causing a life-threatening blockage of blood flow known as a pulmonary embolism (PE). While most people with mild protein C deficiency never develop abnormal blood clots, certain factors can add to the risk of their development. These factors include increased age, surgery, inactivity, or pregnancy. Having another inherited disorder of blood clotting in addition to protein C deficiency can also influence the risk of abnormal blood clotting.

In severe cases of protein C deficiency, infants develop a life-threatening blood clotting disorder called purpura fulminans soon after birth. Purpura fulminans is characterized by the formation of blood clots in the small blood vessels throughout the body. These blood clots block normal blood flow and can lead to localized death of body tissue (necrosis). Widespread blood clotting uses up all available blood clotting proteins. As a result, abnormal bleeding occurs in various parts of the body, which can cause large, purple patches on the skin. Individuals who survive the newborn period may experience recurrent episodes of purpura fulminans.

2. Frequency

Mild protein C deficiency affects approximately 1 in 500 individuals. Severe protein C deficiency is rare and occurs in an estimated 1 in 4 million newborns.

3. Causes

Protein C deficiency is caused by mutations in the *PROC* gene. This gene provides instructions for making protein C, which is found in the bloodstream and is important for controlling blood clotting. Protein C blocks the activity of (inactivates) certain proteins that promote blood clotting.

Most of the mutations that cause protein C deficiency change single protein building blocks (amino acids) in protein C, which disrupts its ability to control blood clotting. Individuals with this condition do not have enough functional protein C to inactivate clotting proteins, which results in the increased risk of developing abnormal blood clots. Protein C deficiency can be divided into type I and type II based on how mutations in the *PROC* gene affect protein C. Type I is caused by *PROC* gene mutations that result in reduced levels of protein C, while type II is caused by *PROC* gene mutations that result in the production of an altered protein C with reduced activity. Both types of mutations can be associated with mild or severe protein C deficiency; the severity is determined by the number of *PROC* gene mutations an individual has.

The Gene Associated with Protein C Deficiency

- PROC

4. Inheritance

Protein C deficiency is inherited in an autosomal dominant pattern, which means one altered copy of the *PROC* gene in each cell is sufficient to cause mild protein C deficiency. Individuals who inherit two altered copies of this gene in each cell have severe protein C deficiency.

5. Other Names for This Condition

- hereditary thrombophilia due to protein C deficiency
- PROC deficiency

References

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