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Congenital protein C or S deficiency

Congenital protein C or S deficiency is a lack of proteins C or S in the fluid part of the blood. The proteins are natural substances that help prevent blood clots.

Causes

Congenital protein C or S deficiency is an inherited disorder. This means it is passed down through families. Congenital means it is present at birth.

The disorder causes abnormal blood clotting.

On average, 1 in 350 people has one normal gene and one variant gene for protein C deficiency.

Protein S deficiency is rare and its prevalence is unknown.

Symptoms

If you have this condition, you are more likely to develop blood clots, especially at a younger age or in unusual areas, such as the veins in the abdominal cavity. The symptoms are the same as for deep vein thrombosis, and include:

- Pain or tenderness in the affected area
- Redness or swelling in the affected area

Exams and Tests

Your health care provider will perform a physical exam and ask about your medical history and symptoms.

Lab tests will be done to check the levels of proteins C and S.

Treatment

Blood-thinning medicines are used to treat and prevent blood clots.

Outlook (Prognosis)

The outcome is usually good with treatment, but symptoms may return, especially if blood-thinning agents are stopped.

Possible Complications

Complications may include:

- Childhood stroke
- More than one pregnancy loss (recurrent miscarriage)
- Recurrent clots in the veins
- Pulmonary embolism (blood clot in a lung artery)

In rare cases, using warfarin to thin the blood and prevent clots can cause brief increased clotting and severe skin wounds. People with these conditions are at risk if they are not treated with the blood-thinning medicine heparin before taking warfarin.

When to Contact a Medical Professional

Contact your provider if you have symptoms of clotting in a vein (swelling and redness of the leg).

Prevention

If your provider diagnoses you with this disorder, you should be careful to prevent clots from forming. This can occur when the blood moves slowly in the veins, such as from prolonged bed rest during an illness, surgery, pregnancy or hospital stay. It may also occur after long airplane or car trips.

Alternative Names

Protein S deficiency; Protein C deficiency

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