Protein C Deficiency

Protein C deficiency is an inherited disorder which is characterized by low levels of Protein C in the blood. The role of Protein C is to balance the slow-flowing venous circulation, in which there is prolonged exposure of procoagulant proteins and platelet phospholipids to the vessel wall. This may explain why protein C deficiency appears to be associated with venous but not arterial thrombosis.

There are two types of Protein C deficiency:

Type I Deficiency refers the quantity of Protein C that is produced and circulating. Heterozygous individuals usually exhibit approximately one half that of normal patient's blood that do not have Protein C deficiency. Some families have a severe thrombotic tendency and other remain asymptomatic. Individuals with Protein C deficiency may also have a secondary mutation such as Factor V Leiden, which has been associated with a more severe form of the deficiency.

Type II deficiency is less common then type I and is associated with decreased functional activity of the protein C with a normal quantity in circulation.

Protein C deficiency by plasma level alone is found in 1 in 200 to 1 in 500 persons in the general population. However, many affected individuals remain asymptomatic throughout life. Protein C deficiency is present in approximately 2-5% of patients presenting with VTE. Severe homozygous or compound heterozygous protein C deficiency occurs in approximately 1 in 500,000 to 1 in 750,000 live births.

The main risk in a Heterozygous individual is the risk of a DVT or deep vein thrombosis. Arterial clots are not associated with heterozygous Protein C deficiency. Most common sites for a DVT are in the deep veins of the lower leg. However, there is an elevated risk for both Mesenteric and cerebral sinus clots to form. A serious event that can be initiated by a DVT would be the development of a Pulmonary Embolism or PE. Approximately 40% of patients with Protein C deficencey present with a PE and 60% suffer repeated clots if anticoagulation is discontinued.

Protein C deficiency may be weakly associated with late and recurrent pregnancy loss. However, it is not uncommon and should be discussed with your Gynecologist.

In certain circumstance, Protein C Deficiency can be acquired and is not inherited. These conditions include:

- Acute thrombosis (blood clot)

- Warfarin therapy
- Liver disease
- Vitamin K deficiency
- Sepsis
- Disseminated intravascular coagulation (DIC)
- Certain chemotherapeutic agents (eg, L-asparaginase)

In rare occurrences an individual with Protein C deficiency may develop Warfarin-induced skin necrosis. This is a medical emergency that required prompt treatment.

Infants that present with a condition called neonatal Purpura Fuminans is also a medical emergency that requires prompt medical treatment.

Deep Vein Thrombosis in patients with protein C deficiency is managed in much the same way as it is for patients with DVT due to other causes. Because the risk of recurrent DVT in protein C deficient patients may be as high as 60%, long-term anticoagulation is often recommended, particularly following a spontaneous thromboembolic event.

References

National Institute of Health: Protein C deficiency: http://www.nlm.nih.gov/medlineplus/