Protein S deficiency

Protein S deficiency is an inherited disorder which is characterized by low levels of Protein S in the blood. Protein S helps control blood clotting by blocking the activity of certain proteins that promote the formation of blood clots.

Low levels of Protein S may result in blood clots or DVT. Individuals with mild Protein S deficiency are at increased risk for a DVT that may break off and travel in the blood stream and become lodged in the lungs, causing a Pulmonary Embolism (PE). Pulmonary embolisms are potentially life threatening and in most cases are a result of a DVT. Individuals with low-levels of Protein S may also be at risk for a PE without evidence of a DVT. There are certain conditions that increase this risk and include: increased age, surgery, immobility and pregnancy.

Protein S deficiency is caused by a mutation on the PROS1 gene. Most individuals inherit one gene and this is referred to as being Heterozygous. If an individual inherits 2 genes, one from each parent, then they are Homozygous for the protein S deficiency. Homozygous individuals are very rare and would have severe protein S deficiency. Mild protein S deficiency occurs in approximately 1 in 500 individuals.

Women who are pregnant with protein S deficiency may have experienced a miscarriage. It is recommended that woman with this type of clotting disorder should avoid estrogen-based birth control pills. In some individuals, anticoagulation with a blood thinner may be necessary. Consideration should be made for anticoagulation with pregnancy, surgery, trauma and chronic conditions that are known to predispose individuals to clots.

More information can be obtained from the National Institute of Health and the genetic Home Reference page.

References

National Institute of Health: Genetic Home Reference. http://ghr.nlm.nih.gov/condition/protein-s-deficiency