

Hereditary Antithrombin Deficiency

Antithrombin III (AT3) deficiency is an inherited disorder which can cause abnormal development of blood clot. These clots are mainly seen in the form of a Deep Vein Thrombosis or DVT. Individuals with a DVT are at risk for developing a potential life-threatening clot to the lungs called a Pulmonary Embolism or PE. These leg clots can break off and travel in the bloodstream and become lodged in the lungs. Anti-thrombin is a protein in the blood that is naturally present in all individuals that acts like a natural blood thinner. Low Antithrombin levels result in more clot formations.

The occurrence of AT3 deficiency is estimated to occur in about 1 in 2,000 to 3,000 individuals. There are 2 types of the Antithrombin III deficiency.

Type 1: An individual does not produce enough Antithrombin. This is caused by inherited gene mutation. There are circumstances that may also give rise to this type and include liver cirrhosis, severe kidney disease, cancer or trauma.

Type 2: An individual may produce a normal amount of protein but it has an abnormal structure. Because of this abnormality, the protein does not work correctly and the activity level of the protein is low.

Antithrombin III is an autosomal dominant disorder in which one copy of the mutated gene is enough to cause the disorder. Inheritance of 2 copies of the mutated gene is not compatible with life. About one half of all individuals with AT3 deficiency will experience a blood clot in their life time. There are certain conditions that may raise this risk and they include: obesity, smoking, sedentary lifestyle and the presence of additional clotting disorders, like Factor V Leiden.

Women with Antithrombin III deficiency are at high risk for developing clots during and after delivery. Treatment with an anticoagulant may be recommended depending on the individual. Women are at an increased risk of clots with Estrogen-based oral contraception pill and should be avoided . More information may be found at the National Institute of Health web site and the Genetics Home Reference page.

References

National Institute of Health: Genetics Home reference. Retrieved from <http://ghr.nlm.nih.gov/condition/hereditary-antithrombin-deficiency>