



Hereditary Antithrombin Deficiency

Facts

People with hereditary antithrombin deficiency have lower than normal levels of antithrombin and are at increased risk for venous blood clots, including pulmonary embolism, or PE (clots in the lung) and deep vein thrombosis, or DVT (e.g., clots in the leg).¹ Clots can also appear in veins in other areas of the body, including arms, intestinal tract and around the brain.¹

General Facts

- Antithrombin is a naturally occurring protein that helps regulate the blood clotting mechanism in the body¹. People with hereditary antithrombin deficiency have lower than normal levels of antithrombin, putting them at increased risk for venous thromboembolic events (VTE), including pulmonary embolism and deep vein thrombosis, which can be life threatening, particularly in the high-risk situations of surgery or childbirth.¹ Antithrombin is often referred to as “heparin cofactor” as the action of heparin depends on adequate levels of antithrombin.¹
- Antithrombin deficiency may be caused by either a hereditary deficiency (genetic disorder) or an acquired deficiency (for example, impaired synthesis or increased consumption of antithrombin).
- While hereditary antithrombin deficiency is uncommon, it carries a high risk of thrombosis. The prevalence of hereditary antithrombin deficiency in the general population is approximately one in 2,000 to one in 5,000.^{2,3}
- By the age of 50, approximately 50 percent of people with hereditary antithrombin deficiency will have experienced a VTE.³
- Blood clots caused by DVT affect more than 380,000 people per year in the United States. These clots can dislodge and travel to the lungs and cause PE. Lung clots affect more than 530,000 people in the United States each year, and nearly 33 percent are fatal.⁶ The prevalence of VTEs caused by hereditary antithrombin deficiency has been reported in the literature to be from 2 to as high as 8 percent.⁷
- Patients with hereditary antithrombin deficiency may normally be maintained on a blood thinning agent. When these patients undergo the high risk situations of surgery or childbirth, the blood thinning medication may be discontinued to minimize the risk of excessive bleeding. Antithrombin helps restore normal anticoagulation during the high risk periods.
- Symptoms of DVT may include swelling, often in one leg, leg pain or tenderness, discoloration of the skin and skin on the leg that is warm to the touch.⁶ Patients with hereditary antithrombin deficiency may have clots in uncommon sites such as the upper limb deep veins, mesenteric veins, vena cava, renal veins and retinal veins.¹
- Symptoms of PE may include sudden shortness of breath, sharp, stabbing chest pain, drastically increased heart rate and an unexplained cough that may be bloody.⁶

Epidemiology

- There are two types of hereditary antithrombin deficiency: Type I is a quantitative deficiency characterized by low levels of antithrombin. Type II is a qualitative deficiency characterized by the presence of antithrombin which does not function properly. Type I hereditary antithrombin deficiency patients are thought to be more prone to VTE than Type II patients.^{8,3}
- Hereditary antithrombin deficiency is a genetic disorder inherited in an autosomal dominant pattern. If one parent has hereditary antithrombin deficiency, there is a 50 percent chance their child will inherit it. Men and women are equally affected.¹
- Acquired antithrombin deficiency can result from impaired synthesis (liver disease) or increased consumption (heparin resistance, DIC due to sepsis) of antithrombin.¹

Diagnosis

- There are two types of blood tests to measure antithrombin. The first measures the antithrombin antigen, which is the amount of the protein in the blood. The second measures the antithrombin activity level, which gauges whether the antithrombin present in the blood is working, and is the preferred test.¹

References:

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