

Antithrombin III Deficiency

Antithrombin III (AT-III) is a protein made in the liver. It inhibits coagulation and limits the forming of blood clots.

A shortage of AT-III affects the normal process of coagulation and can lead to excessive blood clotting. There are two categories of AT-III deficiency. Patients with Type I deficiency have reduced amounts of AT-III protein and functional activity, while patients with Types II and III deficiency have normal protein levels, but some of it does not function properly.

Antithrombin-III deficiency can cause or lead to thrombosis, a clot forming in a blood vessel. If a clot attached to a blood vessel wall breaks loose and travels in the bloodstream, it is called an embolus. An embolus that reaches a blood vessel in the lungs is called a pulmonary embolism. This type of clot can block the blood vessel, cut off the oxygen supply to the lung tissue, and, in some cases, cause death.

Signs and Symptoms

Patients with AT-III deficiency may have thromboembolic problems that begin in early adulthood. Clots forming in the legs, which may cause pain and swelling, and pulmonary embolism, which often causes chest pain and coughing, are most commonly reported. Pregnancy, the use of oral contraceptives, and surgery may contribute to the thromboembolic event.

Causes

Antithrombin III deficiency is usually inherited and affects males and females equally. AT-III deficiency is found in approximately 1 in 2,000 to 5,000 persons. All family members should be tested if there is history of the disease.

Acquired AT-III deficiency can occur as a result of other conditions. It has been reported in patients with liver disease, patients receiving certain kinds of chemotherapy, and patients using oral contraceptives. Oral contraceptives tend to lower AT-III levels, and their use should be evaluated regularly.

Treatment

The goal of treatment is to prevent thrombosis. For patients who have had a thromboembolic event, treatment with anticoagulating drugs may be necessary. Heparin is commonly prescribed immediately during a critical event followed by warfarin for long-term use. Heparin is also the drug of choice during pregnancy because it does not cross the placenta from the mother to the baby.

Replacement therapy using purified human AT-III infusion is now available. However, it is not generally recommended due to the high cost, the risk of infection, and the need for frequent intravenous (IV) administration. Patients with AT-III deficiency who are undergoing surgery or who have extensive thrombosis may be given transfusions of fresh plasma or a heat-treated AT-III concentrate.

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