Antithrombin Deficiency



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Characteristics

Autosomal Dominant
Domino
Antithrombin III Deficiency is inherited in an autosomal dominant fashion. It can also be acquired secondary to DIC, liver disease, or nephrotic syndrome.

Increased Thrombin and Factor X
Up-arrow Trombone and Factory (10) tin.
Antithrombin III is a plasma protease inhibitor that has a role in inactivating thrombin. The lack of antithrombin in antithrombin III deficiency causes a decrease in thrombin inhibition. This will result in increased thrombin and factor X.

Heparin Resistance
Hippie-heron Resistance-band
The action of heparin requires antithrombin to work properly. In antithrombin III deficiency, heparin can not work well if it's administered alone. This is known as heparin resistance.

Hypercoagulability
Hiker-clogs
The decreased inhibition of thrombin in antithrombin III deficiency will cause a hypercoagulable state. Patients can present with thromboses at a young age such as deep vein thrombosis and pulmonary embolism. Venous blood vessels are affected more than arterial blood vessels.

Diagnosis

Family History
Family Portrait
On average, 50% of children of parents heterozygous for antithrombin III deficiency (one affected) can inherit this disease. Therefore, information about family history is essential in these patients.

Normal PT, PTT, and Bleeding Time
Normal-sign Blood Clock and Normal PT and PTT
Antithrombin III deficiency does not interfere with the production of platelets nor with intrinsic or extrinsic coagulation factors. This will result in a normal bleeding time, aPTT, and PT, respectively.

Antithrombin-Heparin Cofactor Assay
Ant-tie-trombone and Hippie Heron
Antithrombin-heparin cofactor assay is a measurement of the ability of antithrombin to bind heparin and neutralize thrombin or factor Xa. Depending upon the enzyme used, the test can be either a thrombin inhibition assay or a factor Xa inhibition assay (most preferred test). Diagnosis is based on the plasma level of antithrombin.

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Management

Factor Xa Inhibitors

(10) Tin with A-apple in Inhibiting-chains

Factor Xa inhibitors are used as an alternative to heparin in treating thromboses in antithrombin deficiency. Instead of giving a higher dose of heparin, factor Xa inhibitors act independently of antithrombin levels.

Direct Thrombin Inhibitors

Direct Trombone in Inhibiting-chains

Heparin may not be effective in treating antithrombin III deficiency due to its dependency on antithrombin activity. To overcome this issue, a direct thrombin inhibitor is used for anticoagulation.

Low Molecular Weight Heparin

Down Arrow Molecule Weight Hippie Heron

Low Molecular Weight Heparin is recommended in pregnant patients with antithrombin deficiency. A larger dose is needed due to heparin resistance in this disorder.

Antithrombin Replacement

Ant-tie-trombone

Replacement of antithrombin III is indicated in patients with severe thrombosis, recurrent thromboses, or difficulty achieving sufficient anticoagulation.

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