

Prothrombin G20210A Mutation



PLAY PICMONIC

Pathophysiology

Second Most Common Genetic Clotting Disorder in Caucasians

(2) Tutu on Caucasians with Clogs

Prothrombin G2O21OA mutation is the second most common genetic clotting disorder in Caucasians. The first and most common disorder is factor V Leiden Thrombophilia.

Autosomal Dominant

Domino

Prothrombin G2O21OA Mutation is inherited in an autosomal dominant fashion. Therefore, information about family history is essential for these patients. On average, 50% of children of heterozygous parents (one affected) can inherit this disease.

Genetic Point Mutation

Pointy Mutant

This disorder is characterized by a genetic point mutation in the prothrombin gene in the 3' Untranslated Region.

3' Untranslated Region

(3) Tree No-sign-train-lotion

Mutation of prothrombin G2O21OA affects the terminal 3' nucleotide of the 3' untranslated region of the Prothrombin mRNA, resulting in increased prothrombin level in the plasma. Prothrombin G2O21OA mutation occurs from missense mutation with a characteristic of guanine replacing adenine.

Increased PT

Up-arrow Pro PT-hourglass

Prothrombin time can increase in this disorder by 1,5-2 times normal.

Clinical Features

Hypercoagulability

Hiker-clog State

The high level of prothrombin in the Prothrombin G2O21OA Mutation will lead to an increased risk of clotting (hypercoagulability). These include a blood clot in the deep veins, legs (DVT), and clots that travel to the lung (pulmonary embolism).



Increased Risk of Thromboembolism

Up-arrow Risk Trombone-Elmo

The presence of recurrent DVT in a patient, especially in young patients, can lead to suspicion of inherited blood clotting disorder (thrombophilia). Healthcare providers should include prothrombin G2O21OA mutation as one of the differential diagnoses.

Considerations

Avoid Oral Contraceptives

Avoid-sign with OCP-case

Oral contraceptives have a risk of thrombosis and cardiovascular events and should be avoided in prothrombin G2O21OA mutation patients.

Caution During Pregnancy

Caution-tape Pregnancy

Pregnancy can cause increased procoagulants and decreased anticoagulants. These conditions and the presence of blood stasis due to compression from pregnancy will increase the risk of thrombosis in prothrombin G2O21OA mutation patients.

Caution After Surgery

Caution-tape Surgeon

Patients after surgery should be monitored due to the risk of blood stasis with thrombosis due to prolonged immobility.

Caution During Long Flights

Caution-tape Long Airplane

Long flights should be taken cautiously in patients with prothrombin G2O21OA mutation due to increased thrombosis resulting from prolonged immobility in the passenger seat.

Treatment

Anticoagulation

Ant-tie-clogs

Anticoagulation is used in patients with Prothrombin G20210A Mutation with the presence of thromboses such as DVT or pulmonary embolism. Warfarin may be used as an anticoagulant to manage this disorder. Patients without thrombosis are not given the anticoagulant routinely, but counseling should be held with high-risk conditions such as pregnancy, surgery, and acute illness.