

## **Prothrombin G 20210A Mutation**

Prothrombin is a protein found in the human body that helps blood clot. Too little of Prothrombin will result in bleeding and too much Prothrombin causes the blood to clot. Prothrombin circulates in the blood and when activated is then converted into Thrombin. This action is what forms Fibrinogen which is another clotting factor that converts it to Fibrin strands. These strands help make up part of a clot.

Prothrombin G 20210A, also called Factor II, is most commonly seen in people of European origin and is seen equally in men and women. The Factor II (Prothrombin) mutation is in fact the second most common genetic defect for inherited thrombosis, with Factor V Leiden being the most common. This genetic mutation on the Prothrombin gene causes individuals with the mutation to have a risk of developing a Deep Venous Thrombosis (DVT). The risk is twice as great with this mutation than someone who does not have the mutation in regards to the formation of a blood clot or DVT. This risk is relatively low and most people with this mutation are not affected.

Prothrombin G 20210A does not increase the risk of arterial thrombosis, so there is no increase in risk of heart attacks or strokes associated with this genetic mutation. If you have inherited a single gene from one of your parents you are heterozygous for the mutation. If you inherited 2 genes, one from each parent, then you are homozygous for the mutation. It is very rare for an individual to have 2 genes of Prothrombin G 20210A mutation. However, some individuals who have Factor V Leiden mutation may also have the Prothrombin G 20210A Mutation.

Women with Prothrombin G 20210A are at a 16 times greater risk of developing a DVT (blood clot) if they use estrogen-based birth control pills. It is also advised that women with this mutation avoid hormone replacement therapy. There are some circumstances that may alter the recommendations and consultation with a Hematologist is advised. Pregnancy may be complicated with this mutation and should be discussed with their Obstetrician.

### Sources

<http://www.fvleiden.org/publications/g20210a.html>

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=ptt>