**What is Prothrombin?**

- Prothrombin is a protein that causes blood to coagulate and form blood clots. A genetic mutation (called G20210A) in the production of this protein is a risk factor for thrombosis (blood clots) including deep venous thrombosis (DVT). This mutation in the gene encoding the clotting factor prothrombin is found in about 1 in 50 persons in the US. It raises the risk of thrombosis significantly for both males and females in all age groups.

- The Prothrombin G20210A mutation increases circulating prothrombin levels. This appears to create a hypercoagulable state.

**Risks associated with Prothrombin**

- This gene provides instructions for making a protein called prothrombin (also called coagulation factor II). Coagulation factors are essential proteins for normal blood clotting. After an injury, clots protect the body by sealing off damaged blood vessels, preventing additional blood loss.

- This mutation causes the gene to be overactive and leads to the excess production of prothrombin, which may lead to high rates of blood clot formation.

- People who have prothrombin mutation G20210A have a 2-to-3 fold increase in the risk of DVT (Deep Vein Thrombosis). Persons who have this mutation plus the factor V Leiden mutation have a 10-to-20 fold increase in thrombotic risk.

- Other factors also increase the risk of blood clots in people with prothrombin thrombophilia (a disorder that causes overcoagulation of the blood). These factors include increasing age, obesity, trauma, surgery, smoking, the use of oral contraceptives (birth control pills) or hormone replacement therapy, and pregnancy.

**Who should be tested?**

- Those who have had a blood clot in one of the deep veins of the body (also called deep vein thrombosis or DVT)

- Those who have had a blood clot that has traveled to the lung (called a pulmonary embolism or PE)

- Those who have had a blood clot in an unusual site (such as the mesenteric or cerebral sinus vein)

- Those who have suffered a heart attack or stroke at a young age

- Those who have a history of recurrent pregnancy loss or stillbirth.

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### Results - F2 G20210A Mutation

There are three possible outcomes:

**Negative:** zero copies of this genetic mutation

- This genotype indicates normal enzyme activity and is not associated with any increased risks of thrombosis (blood clots).

- Normal risk of abnormal blood clots is 1 in 1,000 per year (0.10%)  

**Heterozygous:** one copy of this genetic mutation

- This result is associated with an increased risk of deep vein thrombosis (excessive blood clotting in veins).

- Inheriting one copy of this gene mutation may increase that risk to 2 to 3 in 1,000. (0.2 – 0.3%) compared to normal (0.1%).

- The G20210A mutation is associated with an increased risk of myocardial infarction (heart attack) (4-fold in women, and a 1.5-fold increase in risk for men).

- This result is also associated with an increase in first trimester miscarriage and pregnancy complication rates compared to the general population. Please note: Many women with this mutation go through one or multiple normal pregnancies with no difficulties.

**Homozygous Positive:** Two copies of this genetic mutation

- This result is associated with increased risk of deep vein thrombosis (excessive blood clotting in veins).

- The homozygous positive subject has increased risk for blood clot formation by 10 fold over a ten year period.

- This result is also associated with an increased risk of myocardial infarction (heart attack) (4-fold in women, and a 1.5-fold increase in risk for men).

- Evidence suggests that increased prothrombin levels, which occur when a person has this genetic mutation, might affect critical aspects of placental development (e.g., cell adhesion, smooth muscle proliferation, and vasculogenesis). Please note: Many women with this mutation go through one or multiple normal pregnancies with no difficulties.

- This result is associated with an increase in first trimester miscarriage and pregnancy complication rates.