

Rush Hemophilia & Thrombophilia Center
Prothrombin Gene Mutation Educational Resource

The following information is provided to you for educational purposes. Please contact a staff member of the Rush Hemophilia & Thrombophilia Center if you have questions or concerns. This information is intended to supplement the education that you received in the clinic during our face-to-face visit. If you are experiencing any symptoms described here, go to the closest emergency room for evaluation and possible treatment.

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What is the prothrombin gene mutation?

The prothrombin gene mutation is a genetically determined trait that increases the risk for blood clots. About 1-2% of the general Caucasian population is heterozygous (one copy) for the prothrombin gene mutation. This gene abnormality is less common in Asians and African Americans. The estimated increase in the risk of developing a blood clot if you are heterozygous (one copy) for the prothrombin gene mutation is 2-5 fold (1 in 3700) and increases to 16 fold (1 in 800) for women who take oral contraceptive pills. During pregnancy, a women's risk of blood clots is already increased and the presence of this gene mutation, further increases that risk. The risk for people who are homozygous (two abnormal copies) is even greater.

What problems may develop because of the prothrombin gene mutation?

1. Venous thrombosis (blood clot in veins) such as
 - ! Deep vein thrombosis (DVT) (veins in legs and arms)
 - ! Superficial thrombophlebitis
 - ! Pulmonary embolism (PE) (blood clot in lung)
 - ! Sinus vein thrombosis (veins around the brain)
 - ! Mesenteric vein thrombosis (intestinal veins)
 - ! Budd-Chiari syndrome (liver veins)
2. **Arterial clots (stroke, heart attack) in selected patients (some smokers)**

It is likely but not proven that the prothrombin gene mutation increases the risk of arterial thrombosis (stroke, heart attack). Therefore it is important for these individuals to eliminate other risk factors including: smoking, high blood pressure, high cholesterol, and obesity and to increase physical activity and exercise regularly.

3. **Transient ischemic attacks (TIA's)**
4. **Possibly with stillbirth or recurrent unexplained miscarriage**
5. **Preeclampsia and/or eclampsia (toxemia while pregnant)**

What is Deep Vein Thrombosis?

Deep vein thrombosis is a medical condition for blood clotting. This is a process for formation of thrombi that either partially or completely block circulation in a deep vein, generally in the lower extremities. These veins—the iliac, femoral, popliteal, and tibial—bear primary responsibility for returning deoxygenated blood to the heart. Unlike the superficial veins just below the skin surface, the deep veins are surrounded by powerful muscles that contract to force blood back to the heart. One-way valves inside the veins prevent backflow of blood between muscle contractions. The quick and efficient return of blood to the heart using the power of the leg muscles is a crucial phase of the circulatory process.

What are the symptoms of Deep Vein Thrombosis?

DVT may cause pain, swelling, and discoloration of the affected area, and skin that is warm to the touch. But up to 50% of deep vein thromboses produce minimal symptoms or are completely "silent."

Symptoms of DVT may include:

- ! Pain
- ! Tenderness
- ! Swelling
- ! Discoloration
- ! Warmth

What is the risk of DVT?

The major risk associated with DVT is development of pulmonary embolism (PE). A fragment of a blood clot breaks loose from the wall of the vein and migrates to the lungs, where it blocks a pulmonary artery or one of its branches.

Symptoms of PE may include

- ! Shortness of breath
- ! Apprehension
- ! Rapid pulse
- ! Sweating
- ! Sharp chest pain
- ! Bloody sputum
- ! Fainting

For more information, please contact a staff member of the Rush Hemophilia & Thrombophilia Center.