

PROTHROMBIN MUTATION G20210 AND BLOOD CLOTting

What is normal clotting?

A blood clot (also call a thrombus or thrombosis) is one of our natural defenses against bleeding. Normally, blood is maintained in a fluid state in the body because of a delicate balance between proteins that make clots (also called coagulants or factors) and proteins that prevent clots (also called anticoagulants). Blood clotting occurs when there is damage to the blood vessel. 1st, the platelets, which are tiny blood cells like sticky jigsaw puzzle pieces, plug the injured vessel. At the same time they release chemicals that start the clotting process. The result is like a domino effect that eventually leads to the formation of a blood clot. Whenever a blood clot is formed, anticoagulants are also activated to prevent excess clot formation and start the clot breakdown process. It is a delicate balancing act between bleeding and clotting.

What happens when a clot is formed?

Clots can occur in veins (venous) or arteries (arterial). Arteries are tougher, elasticized blood vessels that carry blood away from the heart and deliver oxygen to the body. Arterial blood clots may cause stroke, heart attack, or damage to limbs or other organs. Veins are thinner, collapsible blood vessels that carry blood back from the tissues to the heart. Venous blood clots are often associated with surgery, pregnancy, birth control pills, IV lines or other external factors. Other common risks of thrombosis include cigarette smoking, high blood pressure, high cholesterol, and diabetes.

How do I get the prothrombin mutation?

The prothrombin mutation is not a disease; it is the presence of a specific gene that is passed on to you from your parents (inherited). As with all inherited diseases we inherit one gene from our mother and one from our father and so it is possible to inherit two normal genes or one abnormal prothrombin gene and one normal or two abnormal prothrombin genes. Having one abnormal prothrombin gene (called heterozygous) will result in a slightly higher risk of developing a thrombosis than people who do not have the gene, but having two abnormal prothrombin genes (called homozygous) makes the risk much greater. There is further risk of thrombosis if you have a second risk factor such as oral birth control pills, estrogen therapy, pregnancy, immobilization, surgery, infection, increased age, protein C deficiency or other coagulation abnormalities.

How do I know if I have the prothrombin mutation?

The prothrombin mutation is a silent abnormality. The only way to know if you have prothrombin mutation is by a blood test. The test will determine the presence or absence of the mutation and distinguishes between the heterozygous and homozygous type. The test can be performed with results in 5-7 days.

What are the symptoms of the prothrombin mutation?

There are no specific symptoms of the prothrombin mutation unless you have a clot. The most common sites for clots are in the legs (deep vein thrombosis - DVT) and in the chest (pulmonary embolism - PE). Manifestation of the prothrombin mutation can be extremely variable. Many

people with the prothrombin mutation never develop a clot; yet, some have recurrent clotting before the age of 30. The diagnosis is suspected in patients with a history of thrombosis and in those with a family history.

How do I know if I should be tested?

People should be tested if they:

- Have a family history of clotting or thrombophilia
- Have a history themselves of clotting
- Have a history of transient ischemic attacks or premature stroke
- Have a history of repeated miscarriages or stillbirths

How do I get treated if I have the prothrombin mutation?

You should be tested for other possible inherited or acquired clotting disorders. Treatment of the prothrombin mutation depends on the clinical circumstances such as another inherited or acquired clotting abnormality. If you have only the prothrombin mutation you do not need treatment unless your blood starts to clot. If you have or have had a blood clot and have the abnormal prothrombin gene, you will be put on anticoagulant therapy for a period of time ranging from 3 months to life. The length of time you are on anticoagulants depends on several issues such as; how many blood clots you have had, how serious your clot was and how many additional risk factors you have. Protective anticoagulation therapy may be needed in times where your risk for developing a clot is greater such as pregnancy, surgery, or long plane/car rides. Discuss this with your Hematologist.

How do I keep from getting a clot?

1. Avoid long periods of bed rest. Avoid prolonged sitting or standing in one position. Don't cross ankles or legs while sitting or lying. Keep your feet higher than your hips while sitting. While resting, occasionally move your legs, ankles and toes to promote circulation
2. On long car or plane rides, get up and move around for ~15 minutes at least every 2 hours
3. Do not smoke or be around others who are smoking
4. Exercise regularly and stay well hydrated
5. Use support stockings (such as JOBST compression stockings or TED compression stockings) if you have severe varicose veins or if you have had a clot in your legs
6. Avoid knee socks or hosiery that might limit blood flow. Avoid tight diapers around the legs in infants
7. Do not take Birth Control pills without discussing it with your Hematologist
8. If you become pregnant, consult your GYN and Hematologist about the prothrombin abnormal gene
9. Tell the doctor or surgeon prior to having surgery about having the prothrombin abnormal gene

If you have any questions about your diagnosis, please contact KC Clevenger, Coagulation Nurse Practitioner at 303.861.6972 or the Hematology fellow on call if after hours or on the weekend at 303.861.6740.