

PROTEIN S DEFICIENCY AND BLOOD CLOTS

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 with 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.

What is Protein S deficiency and why does it make a clot?

Protein S is a protein that prevents clotting (an anticoagulant), and requires vitamin K for its production and works with Protein C. The function of protein S is to stop the action of two activated clotting proteins named factor V and factor VIII. If there is not enough (deficiency) of the Protein S, these clotting proteins remain activated increased the tendency for blood to clot.

How do I get Protein S deficiency?

Protein S deficiency can be inherited (present at birth) past from parents to child. It can also be acquired (after birth) as in vitamin K deficiency, with certain medications or with infection. If the Protein S deficiency is inherited, it is the presence of a specific gene that is passed on to you from your parents. 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 gene and one normal or two abnormal Protein S genes. Having one abnormal Protein S gene (heterozygous) will result in a slightly higher risk of developing a thrombosis than people who do not have the gene, but having two genes (homozygous) makes the risk much greater and potentially fatal. 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, or other coagulation abnormalities such as Factor V Leiden.

How do I know if I have Protein S deficiency?

Protein S deficiency is a silent abnormality. The only way to know if you have Protein S deficiency is by a blood test. The test will determine the level of Protein S in your blood. The test can be performed with results in 2-3 days. There are no particular symptoms of Protein S deficiency unless you have a

clot. The most common sites for a clot is in the legs (deep vein thrombosis – DVT) and in the chest (pulmonary embolism). Blood clots can also be in the brain. Manifestation of Protein S deficiency can be extremely variable. Many people with Protein S deficiency never develop thrombosis, yet, some have recurrent clotting before the age of 20. The diagnosis is suspected in patients with a history of thrombosis and in those with a family history of thrombosis.

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 Protein S deficiency?

You should be tested for other possible inherited or acquired clotting disorders. Treatment of Protein S deficiency depends on each individual and clinical circumstance. If you have only the Protein S deficiency you do not need treatment unless your blood starts to clot. If you have or have had a blood clot and have an abnormal Protein S level, 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 may 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 second-hand smoke
4. Exercise regularly and stay well hydrated
5. Use support stocking 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
8. If you become pregnant, consult your GYN and Hematologist
9. Tell the doctor or surgeon prior to having surgery about your Protein S deficiency

If you have any questions about your diagnosis, please contact the Mountain States Hemophilia and Thrombosis Center at 303-724-1154 or the Children's Hospital Thrombosis Clinic at 303-861-6972.

