INHERITED THROMBOPHILIA SCREENING AND FAMILY TESTING

PATIENT INFORMATION SHEET

WHAT IS THROMBOSIS?

Thrombosis is a medical term for a blood clot.

A deep vein thrombosis (DVT) is a blood clot that occurs most commonly in the deep vein of the leg but can occur in any vein in the body.

If left untreated, DVT can move to the lung where it causes pulmonary embolism (PE). Pulmonary embolism causes shortness of breath, chest pain, and tiredenss. Once DVT has moved to the lung, the clot is stuck there and cannot move. Because blood clots in the lungs cannot move, PEs do not cause strokes or heart attacks. About half the time people with DTV also have PE.

WHAT IS THROMBOPHILIA SCREENING?

Blood tests that can look for the presence of a thrombophilia gene or a lower level of the specific protein or protein activity. These genes can be passed on through the family.

WHO SHOULD BE TESTED?

- Most people do not need testing as the results will not affect or change your treatment.
- Sometimes testing is done at the wrong time and results can be confusing. For example some blood tests cannot be done if you are taking an anticoagulant (also called a blood thinner).
- Thrombophilia testing is expensive.
- Life insurance may be more expensive or harder to get if you test positive for a thrombophilia.

WHAT IS THROMBOPHILIA?

Thrombophilia is a tendency to form excessive blood clots. Thrombophilia can be caused by inherited genetic changes (also called mutations) that may make your blood clot more easily than other people. If you have tested positive, then you have inherited the genetic change from your mother and/or father.

Some examples of thrombophilia:

Factor V Leiden:
Interferes with the body's ability to protect from thrombosis. It is a common genetic mutation that occurs in about 1 in 20 people. Factor V (5) helps your blood clot, so having too much encourages clots.

Prothrombin Gene Mutation:
Common genetic mutation that occurs in about 2 or 3 in 100 people. It increases the amount of the clotting protein prothrombin, which increases the chance of thrombosis.

Protein C Deficiency:
Rare disorder that occurs in about 3 in 1000 people. Protein C is a protein in the blood that stops your blood from making clots, so having a lower amount than normal encourages clotting.

Protein S Deficiency:
Rare disorder that occurs in about 3 in 1000 people. Protein S is a protein in the blood that stops your blood from making clots, so having a lower amount than normal encourages clotting.

Antithrombin Deficiency:
Rare disorder that occurs in about 1 in 2500 people. Antithrombin is a protein in the blood that stops your blood from making clot. Low antithrombin levels reduce the body's ability to stop excessive clot formation and put patients at risk for thrombosis.

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WHAT IF I HAVE A THROMBOPHILIA?

- Having a thrombophilia may slightly increase your chance of forming a first-time blood clot. This is still a small chance.
- If you have a blood clot, usually other things that cause blood clots are more important than thrombophilia.
- If you are pregnant or on estrogen, talk to your doctor to see if this is important.

WHAT ABOUT MY FAMILY?

There is no need to test everyone in your family.

Having a close relative with a blood clot increases your chance of forming a blood clot, regardless of thrombophilia testing. It is important for family members to know the symptoms of blood clots, and things that can increase their risk, so they can talk to their healthcare professional if these happen to them.

TAKE AWAY MESSAGE

- Thrombophilia testing does not change how most people are treated for blood clots
- Testing for thrombophilia should not be done in most people

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