HYPERHOMOCYSTEINEMIA

THROMBOPHILIA

Information for Patients

UNIVERSITY OF IOWA HEALTH CARE
Thrombophilia (Hypercoagulability) is a condition in which a person forms blood clots more than normal. Blood clots may occur in the arms or legs (e.g., deep vein thrombosis - DVT), the lungs (e.g., pulmonary embolism - PE) or other organs. Blood clots can cause swelling, pain, or shortness of breath. Other symptoms of blood clots include dizziness, headache, changes in vision, lack of concentration, confusion, slurred speech, numbness, tingling, and weakness in the arms or legs. Blood clots may become life threatening if not recognized and treated early.

Some forms of thrombophilia are inherited from one or both parents. This is called inherited thrombophilia. Examples include:

- Factor V Leiden
- Prothrombin gene mutation
- Antithrombin deficiency
- Protein C deficiency
- Protein S deficiency

Other forms of thrombophilia develop after birth. This is called acquired thrombophilia. Examples include:

- Antiphospholipid syndrome
- Hyperhomocysteinemia

Things that may trigger or cause blood clots to form include: surgery, trauma (injury) or fractures, bed rest or sitting or lying still for several hours at a time, cancer and chemotherapy, intravenous catheters, estrogen use, pregnancy, or air travel. Sometimes, blood clots happen without an identifiable trigger or cause.
HYPERHOMOCYSTEINEMIA

Homocysteine is a naturally occurring amino acid that, if present at a high level in the blood, can produce an increased risk of blood clots. This condition is known as hyperhomocysteinemia. People with hyperhomocysteinemia may get blood clots in either the veins (e.g. deep vein thrombosis and pulmonary embolism) or arteries (e.g. stroke and heart attack).

It is believed that high blood levels of homocysteine can damage the lining of blood vessels. This damage is what can lead to blood clots. In addition to making people prone to blood clots, hyperhomocysteinemia will also increase the risk of birth defects, dementia (e.g., Alzheimer’s disease), and bone fractures.

Common causes of hyperhomocysteinemia include kidney disease, lack of B vitamins (such as folate, vitamin B12, and vitamin B6) in the diet, hypothyroidism, alcoholism, and certain medications.

Diagnosis

The diagnosis of hyperhomocysteinemia is made by measuring the level of total homocysteine in the blood. Some people inherit defects that cause hyperhomocysteinemia. Most people with inherited defects are only mildly affected. In rare cases, however, inherited defects in genes produce a severe form of hyperhomocysteinemia called homocystinuria. People affected with homocystinuria have a very high risk of blood clots, and also may suffer from mental retardation, bone abnormalities, and visual problems.
Treatment
The primary goal of treatment is to lower blood levels of homocysteine to normal. Treatment may consist of giving supplements of folic acid, vitamin B-12, and/or vitamin B-6. It also may include anticoagulant medications (blood thinners), such as aspirin, clopidogrel, heparin, low-molecular weight heparin, or warfarin, to prevent blood clots.

Patients with the severe form of hyperhomocysteinemia (homocystinuria) are often treated with high doses of vitamin B-6 or betaine, and the amount of methionine consumed in the diet may be restricted.

Prevention
People who have thrombophilia can reduce their risk of getting a blood clot by:

♦ **Exercising regularly** - Exercise improves the body’s ability to dissolve clots that have formed and helps prevent blood from clotting inside the veins.

♦ **Avoiding becoming overweight or obese** - Overeating increases levels of the hormone insulin. Insulin can interfere with the normal clotting controls. Obesity can also cause poor circulation in the veins, leading to a higher risk of clotting.

♦ **Avoiding long periods of immobility during illness or when traveling** - When people sit or lie down for several hours at a time, the circulation of the blood slows down. This may increase the risk of clotting.

♦ **Avoiding cigarette smoke** - Smoking decreases the amount of oxygen in the blood and may damage vessel walls, which may lead to clotting. (Being in smoke-filled rooms/areas can also lead to clotting.)

♦ **Refaining from cigarette smoking** - Smoking decreases the amount of oxygen in the blood and may damage vessel walls, potentially leading to clot formation.
寻求医疗建议前进行重大手术
- 血管可能在手术中受损，导致血栓形成的风险高。重要的是你的医生知道你有血栓前体病史，以便在规划任何手术前进行考虑。

寻求医疗建议前使用避孕药或激素替代疗法
- 激素，包括避孕药可能会增加凝血风险。

寻求医疗建议时怀孕或计划怀孕
- 孕妇有血栓前体病史者在怀孕期间有增加血栓形成的风险，也可能会有更高风险的流产和怀孕并发症。
ANTICOAGULANT MEDICATIONS
Anticoagulant medications can cause you to bleed more easily. You may notice that, if you cut yourself, the blood takes longer to clot. You might bruise more easily. If you have any unusual, heavy or prolonged bleeding, severe headaches, visual changes, or stiff neck, call your doctor.

Treatment may include blood thinners (anticoagulant medications) such as heparin, low-molecular weight heparin, warfarin, aspirin, or clopidogrel.

Warfarin has a stronger effect on some people than others. If you take warfarin, your doctor will want to check you frequently with a blood test called the INR (International Normalized Ratio) or Prothrombin Time (PT). This test will tell your doctor how well the warfarin is working. Many other medications can make warfarin more or less strong. Ask your doctor before you take a new medicine, even a nonprescription medicine or vitamin.

If you are pregnant, you should not take warfarin. Warfarin can cause birth defects. If you are pregnant or might become pregnant, talk with your doctor.

COPING WITH STRESS
The diagnosis of thrombophilia and the treatment it may require can cause stress for many patients and their families. It is normal to feel a variety of emotions, including shock, fear, and anger. To cope, many people have found it helpful to become involved in support groups that are made up of other people and their families facing similar issues. Please ask your nurse, social worker, or doctor if you wish to receive more information about support groups. Professional counselors are also available as an option if you feel you need to additional help dealing with your illness and its treatment.
**Activated protein C resistance test:** A blood test that can be used to detect Factor V Leiden.

**Alzheimer’s disease:** A degenerative disease characterized by progressive brain deterioration and dementia.

**Antibody:** A protein substance normally formed by the body to help defend it against infections and foreign substances.

**Anticardiolipin antibody:** A type of antiphospholipid antibody.

**Anticoagulant:** A substance that prevents excessive blood clotting.

**Anticoagulation medication:** A drug that is given to prevent excessive blood clotting. Sometimes called a blood thinner.

**Antiphospholipid antibody:** An autoantibody that reacts with a normal component of blood cells called phospholipid.

**Antiphospholipid syndrome:** A form of acquired thrombophilia caused by autoantibodies that react with a normal component of blood cells called phospholipid.

**Antithrombin:** A blood protein that prevents excessive blood clotting.

**Antithrombin concentrate:** A medication that contains large amounts of antithrombin.

**Antithrombin deficiency:** A form of inherited thrombophilia caused by a mutation in the gene for the blood protein called antithrombin.

**Autoantibody:** An antibody that reacts with normal components of the body.

** Autosomal dominant:** A pattern of inheritance in which, if one parent has a disorder, then each child has a 50:50 chance of inheriting the disorder.

**Aspirin:** A medication that prevents blood clotting by blocking the effects of platelets.

**Autoimmune disease:** A disorder caused by the production of antibodies that react with normal components of the body.
Betaine: A medication used to treat severe hyperhomocysteinemia.

Blood clotting proteins: Proteins in the blood that are involved in the process of blood clotting.

Cell: The basic building blocks of life. Humans are made of many cells of different types.

Chemotherapy: A medication used to treat cancer.

Clopidogrel: A medication that prevents blood clotting by blocking the effects of platelets.

Coagulation: The process by which blood clots.

Cystathionine beta-synthase (CBS): A protein that breaks down homocysteine. Deficiency of CBS can cause hyperhomocysteinemia.

Deep vein thrombosis: A blood clot that forms in the deep veins of the extremities.

DNA: The substance from which the genetic code is made.

Factor V: One of the blood clotting proteins.

Factor V Leiden: A form of inherited thrombophilia caused by a mutation in the gene for the blood clotting protein, factor V.

Folate: One of the B vitamins.

Folic acid: A form of the vitamin folate.

Genetic counselor: A health care provider that is trained to provide information and counseling about the diagnosis and management of genetic (inherited) disorders.

Genetic disorder: An inherited condition passed down in genes through generations in a family.

Genetic test: A test that detects variant in DNA.

Heart attack: A medical condition that occurs when a blood vessel in the heart is blocked, often by a blood clot.

Hematologist: A doctor who specializes in diseases of the blood and bone marrow.

Heparin: An anticoagulant medication.
**Heterozygous:** A genetic condition in which a person has inherited one normal copy and one abnormal copy of a gene.

**Homozygous:** A genetic condition in which a person has inherited two copies of a gene variant.

**Homocysteine:** An amino acid that is normally present in blood.

**Homocystinuria:** A severe form of hyperhomocysteinemia.

**Hypercoagulability:** A condition in which a person has an increased tendency to form blood clots. Also called thrombophilia.

**Hyperhomocysteinemia:** A form of acquired thrombophilia resulting from a high level of homocysteine in the blood.

**Hypothyroidism:** A medical condition caused by deficient thyroid activity.

**Inherited thrombophilia:** A form of thrombophilia that runs in families, and can be inherited from one or both parents.

**International Normalized Ratio (INR):** A blood test used to monitor the effects of anticoagulant medications such as warfarin.

**Intravenous catheter:** A tube that is used to infuse medications into the blood through a vein.

**Livedo reticularis:** A blotchy skin condition that is sometimes seen in people with the antiphospholipid syndrome.

**Low-molecular weight heparin:** An anticoagulant medication.

**Lupus:** A medical condition known as systemic lupus erythematosus.

**Lupus anticoagulant test:** A diagnostic test for antiphospholipid syndrome.

**Methionine:** An amino acid found in proteins in the diet.

**Methylenetetrahydrofolate reductase (MTHFR):** A protein that breaks down homocysteine. Deficiency of MTHFR can cause hyperhomocysteinemia.

**Mutation:** A change in a gene from what is considered normal.

**Phospholipid:** A component of blood cells that is involved in the blood clotting process.
Platelet: A type of blood cell that assists in blood clotting.

**Primary antiphospholipid syndrome:** Antiphospholipid syndrome in someone who does not have another autoimmune disease.

Protein: Essential molecules in the body made up of many amino acids strung together.

Protein C: An anticoagulant that is normally found in blood.

Protein C deficiency: A form of inherited thrombophilia caused by a low level of protein C in the blood.

Protein S: An anticoagulant that is normally found in blood.

Protein S deficiency: A form of inherited thrombophilia caused by a low level of protein S in the blood.

Prothrombin: A blood clotting protein. Also called factor II.

**Prothrombin time (PT)** - A blood test that measures, in seconds, how quickly blood clots. Used to monitor the effects of anticoagulant medications such as warfarin.

Prothrombin gene mutation: A form of inherited thrombophilia caused by a mutation in the gene for the blood clotting protein called prothrombin.

Pulmonary embolism: A blood clot forms in the deep veins of the legs or other locations and then travels to the lung.

**Secondary antiphospholipid syndrome:** Antiphospholipid syndrome in someone who also has another autoimmune disease.

Stroke: A medical condition that occurs when a blood vessel in the brain is blocked, often by a blood clot.

Systemic lupus erythematosus: A medical condition known as Lupus.

Thrombocytopenia: A low number of platelets in the blood.

Thrombophilia: A condition in which a person has an increased tendency to form blood clots. Also called a hypercoagulable state.

Warfarin: An anticoagulant medication.