Prothrombin Thrombophilia

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Prothrombin thrombophilia is an inherited disorder of blood clotting. Thrombophilia is an increased tendency to form abnormal blood clots in blood vessels.

Keywords: genetic conditions

1. Introduction

People who have prothrombin thrombophilia are at somewhat higher than average risk for a type of clot called a deep venous thrombosis, which typically occurs in the deep veins of the legs. Affected people also have an increased risk of developing a pulmonary embolism, which is a clot that travels through the bloodstream and lodges in the lungs. Most people with prothrombin thrombophilia never develop abnormal blood clots, however. Some research suggests that prothrombin thrombophilia is associated with a somewhat increased risk of pregnancy loss (miscarriage) and may also increase the risk of other complications during pregnancy. These complications may include pregnancy-induced high blood pressure (preeclampsia), slow fetal growth, and early separation of the placenta from the uterine wall (placental abruption). It is important to note, however, that most women with prothrombin thrombophilia have normal pregnancies.

2. Frequency

Prothrombin thrombophilia is the second most common inherited form of thrombophilia after factor V Leiden thrombophilia. Approximately 1 in 50 people in the white population in the United States and Europe has prothrombin thrombophilia. This condition is less common in other ethnic groups, occurring in less than one percent of African American, Native American, or Asian populations.

3. Causes

Prothrombin thrombophilia is caused by a particular mutation in the *F2* gene. The *F2* gene plays a critical role in the formation of blood clots in response to injury. The protein produced from the *F2* gene, prothrombin (also called coagulation factor II), is the precursor to a protein called thrombin that initiates a series of chemical reactions in order to form a blood clot. The particular mutation that causes prothrombin thrombophilia results in an overactive *F2* gene that causes too much prothrombin to be produced. An abundance of prothrombin leads to more thrombin, which promotes the formation of blood clots.

Other factors also increase the risk of blood clots in people with prothrombin thrombophilia. These factors include increasing age, obesity, trauma, surgery, smoking, the use of oral contraceptives (birth control pills) or hormone replacement therapy, and pregnancy. The combination of prothrombin thrombophilia and mutations in other genes involved in blood clotting can also influence risk.

The Gene Associated with Prothrombin Thrombophilia

- F2

4. Inheritance

The risk of developing an abnormal clot in a blood vessel depends on whether a person inherits one or two copies of the *F2* gene mutation that causes prothrombin thrombophilia. In the general population, the risk of developing an abnormal blood clot is about 1 in 1,000 people per year. Inheriting one copy of the *F2* gene mutation increases that risk to 2 to 3 in 1,000. People who inherit two copies of the mutation, one from each parent, may have a risk as high as 20 in 1,000.

5. Other Names for This Condition

- hyperprothrombinemia
- Prothrombin G20210A Thrombophilia

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