F13A1 Gene

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Coagulation factor XIII A chain

genes

1. Normal Function

The *F13A1* gene provides instructions for making one part, the A subunit, of a protein called factor XIII. This protein is part of a group of related proteins called coagulation factors that are essential for normal blood clotting. They work together as part of the coagulation cascade, which is a series of chemical reactions that forms blood clots in response to injury. After an injury, clots seal off blood vessels to stop bleeding and trigger blood vessel repair. Factor XIII acts at the end of the cascade to strengthen and stabilize newly formed clots, preventing further blood loss.

Factor XIII in the bloodstream is made of two A subunits (produced from the *F13A1* gene) and two B subunits (produced from the *F13B* gene). When a new blood clot forms, the A and B subunits separate from one another, and the A subunit is cut (cleaved) to produce the active form of factor XIII (factor XIIIa). The active protein links together molecules of fibrin, the material that forms the clot, which strengthens the clot and keeps other molecules from breaking it down.

Studies suggest that factor XIII has additional functions, although these are less well understood than its role in blood clotting. Specifically, factor XIII is likely involved in other aspects of wound healing, immune system function, maintaining pregnancy, bone formation, and the growth of new blood vessels (angiogenesis).

2. Health Conditions Related to Genetic Changes

2.1 Factor XIII Deficiency

At least 140 mutations in the *F13A1* gene have been found to cause inherited factor XIII deficiency, a rare bleeding disorder. Without treatment, affected individuals have a greatly increased risk of abnormal bleeding episodes, including life-threatening bleeding inside the skull (intracranial hemorrhage). *F13A1* gene mutations severely reduce the amount or activity of the A subunit of factor XIII. In most people with these mutations, the level of functional factor XIII in the bloodstream is less than 5 percent of normal. This loss of factor XIII activity weakens new blood clots and prevents them from stopping blood loss effectively.

2.2 Other Disorders

Several common variations (polymorphisms) in the *F13A1* gene have been studied as possible risk factors for diseases involving abnormal blood clotting. The most common *F13A1* gene polymorphism changes a single protein building block (amino acid) in a critical part of the A subunit, replacing the amino acid valine with the amino acid leucine at protein position 34 (written as Val34Leu or V34L). This genetic change speeds up the activation of factor XIII. The Val34Leu polymorphism has been studied in relation to heart disease, stroke, recurrent pregnancy loss, and several other conditions. However, the results of these studies have been conflicting, and it remains unclear whether the polymorphism represents a major risk factor for any of these conditions.

3. Other Names for This Gene

- bA525O21.1 (coagulation factor XIII, A1 polypeptide)
- coagulation factor XIII A chain precursor
- coagulation factor XIII, A polypeptide
- coagulation factor XIII, A1 polypeptide
- coagulation factor XIIIa
- F13A
- factor XIIIa
- fibrin stabilizing factor, A subunit
- fibrinoligase
- FSF, A subunit
- protein-glutamine gamma-glutamyltransferase A chain
- TGase
- transglutaminase A chain
- transglutaminase. plasma

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