F2 Gene

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1. Normal Function

The *F2* gene provides instructions for making a protein called prothrombin (also called coagulation factor II). Coagulation factors are a group of related proteins that are essential for normal blood clotting (hemostasis). After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

Prothrombin is made chiefly by cells in the liver. The protein circulates in the bloodstream in an inactive form until an injury occurs that damages blood vessels. In response to injury, prothrombin is converted to its active form, thrombin. Thrombin then converts a protein called fibrinogen into fibrin, the primary protein that makes up blood clots.

Thrombin is also thought to be involved in cell growth and division (proliferation), tissue repair, and the formation of new blood vessels (angiogenesis).

2. Health Conditions Related to Genetic Changes

2.1 Prothrombin Deficiency

More than 50 mutations in the *F2* gene have been found to cause prothrombin deficiency. Most of these mutations change one protein building block (amino acid) in prothrombin. Some mutations drastically reduce the activity of prothrombin and can lead to severe bleeding episodes. Other mutations allow for a moderate amount of activity of prothrombin, typically causing mild bleeding episodes. None of the mutations identified eliminate prothrombin function. Researchers believe that people cannot live with a complete absence of prothrombin.

2.2 Prothrombin Thrombophilia

The mutation that causes most cases of prothrombin thrombophilia changes one DNA building block (nucleotide) in the *F2* gene. Specifically, it replaces the nucleotide guanine with the nucleotide adenine at position 20210 (written G20210A or 20210G>A). This mutation, which occurs in a region of the gene called the 3' untranslated region, causes the gene to be overactive and leads to the production of too much prothrombin. An abundance of prothrombin leads to more thrombin, which promotes the formation of blood clots.

3. Other Names for This Gene

- Blood Coagulation Factor II
- coagulation factor II
- coagulation factor II (thrombin)
- prothrombin B-chain
- PT
- Q7Z7P3_HUMAN
- serine protease

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