TMPRSS6 Gene

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Transmembrane serine protease 6: The TMPRSS6 gene provides instructions for making a protein called matriptase-2.

genes

1. Normal Function

The *TMPRSS6* gene provides instructions for making a protein called matriptase-2. This protein is part of a signaling pathway that controls the levels of another protein called hepcidin, which is a key regulator of iron balance in the body. When blood iron levels are low, this signaling pathway reduces hepcidin production, allowing more iron from the diet to be absorbed through the intestines and transported out of storage sites (particularly in the liver and spleen) into the bloodstream. Iron is an essential component of hemoglobin, which is the molecule in red blood cells that carries oxygen.

2. Health Conditions Related to Genetic Changes

2.1. Iron-refractory iron deficiency anemia

At least 40 mutations in the *TMPRSS6* gene have been found to cause an inherited form of anemia called iron-refractory iron deficiency anemia. This condition is characterized by a shortage (deficiency) of iron in the bloodstream that is resistant (refractory) to treatment with iron.

TMPRSS6 gene mutations greatly reduce the amount of functional matriptase-2, preventing it from controlling hepcidin levels. The resulting elevation in hepcidin activity blocks the absorption of iron through the intestines and the release of iron from storage. When not enough iron is available in the bloodstream, less hemoglobin is produced and red blood cells cannot carry oxygen to the body's cells and tissues effectively. The shortage of oxygen causes the signs and symptoms of anemia, which can include tiredness (fatigue), weakness, and pale skin.

3. Other Names for This Gene

- matriptase 2
- matriptase-2
- membrane-bound mosaic serine proteinase matriptase-2

- transmembrane protease serine 6
- transmembrane protease, serine 6
- type II transmembrane serine protease 6

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