TFR2 gene

transferrin receptor 2

Normal Function

The *TFR2* gene provides instructions for making a protein called transferrin receptor 2. The main function of this protein is to help iron enter liver cells (hepatocytes). On the surface of hepatocytes, the receptor binds to a protein called transferrin, which transports iron through the blood to tissues throughout the body. When transferrin binds to transferrin receptor 2, iron is allowed to enter the cell.

Additionally, transferrin receptor 2 can bind to other proteins to help regulate iron storage levels in the body by controlling the levels of another protein called hepcidin. Hepcidin is a protein that determines how much iron is absorbed from the diet and released from storage sites in the body in response to iron levels.

Health Conditions Related to Genetic Changes

Hereditary hemochromatosis

About 50 mutations in the *TFR2* gene cause type 3 hemochromatosis, a form of hereditary hemochromatosis that begins in early adulthood, usually before age 30. Hereditary hemochromatosis is a disorder that causes the body to absorb too much iron from the diet. The excess iron accumulates in, and eventually damages, the body's tissues and organs.

Some *TFR2* gene mutations prevent the production of transferrin receptor 2. Other mutations result in proteins that have an incorrect sequence of protein building blocks (amino acids) or proteins that are too short to function normally. These mutations prevent the protein from binding to transferrin, blocking iron from entering hepatocytes.

Mutations in the *TFR2* gene are also thought to contribute to low levels of hepcidin in the body, which results in too much iron being absorbed from the diet. When this occurs, the excess iron is stored in the body's tissues, especially the liver. Iron overload leads to the organ damage and other signs and symptoms of type 3 hemochromatosis.

Other Names for This Gene

- HFE3

- TFR2_HUMAN
- Transferrin Receptor Protein 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TFR2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7036[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TFR2%5BTIAB%5D%29+OR+%28transferrin+receptor+2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TRANSFERRIN RECEPTOR 2; TFR2 (https://omim.org/entry/604720)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7036)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TFR2[gene])

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Genomic Location

The *TFR2* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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