TFR2 gene
transferrin receptor 2

Normal Function

The TFR2 gene provides instructions for making a protein called transferrin receptor 2. Studies suggest that this receptor helps iron enter liver cells (hepatocytes). In the blood, iron binds to a protein called transferrin for transport and delivery to the liver and other tissues. On the cell surface, transferrin binds to transferrin receptor 2, and iron is allowed to enter the cell. Additionally, this receptor helps sense and 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

At least nine mutations that cause a form of hereditary hemochromatosis designated as type 3 have been identified in the TFR2 gene. Some mutations in the TFR2 gene 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 likely impair the ability to regulate importation of iron into certain cells.

Mutations in the TFR2 gene are also thought to contribute to low levels of hepcidin in the body, which allows too much iron to be 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.
Chromosomal Location

Cytogenetic Location: 7q22.1, which is the long (q) arm of chromosome 7 at position 22.1

Molecular Location: base pairs 100,620,416 to 100,642,780 on chromosome 7 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HFE3
- TFR2_HUMAN
- Transferrin Receptor Protein 2

Additional Information & Resources

Clinical Information from GeneReviews
- TFR2-Related Hereditary Hemochromatosis
  https://www.ncbi.nlm.nih.gov/books/NBK1349

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

Catalog of Genes and Diseases from OMIM
- TRANSFERRIN RECEPTOR 2
  http://omim.org/entry/604720

Research Resources
- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TFR2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TFR2%5Bgene%5D
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14633776

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10802645

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15749659

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12393650

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16634833

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11826284

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15711214

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12737947

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15147384

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15070436
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15175440

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16315138

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12547237

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12531241

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