SLC52A3 gene

solute carrier family 52 member 3

Normal Function

The *SLC52A3* gene (previously called the *C20orf54* gene) provides instructions for making a riboflavin transporter protein called RFVT3 (formerly known as RFT2). This protein moves (transports) a vitamin called riboflavin (also called vitamin B_2) across the cell membrane. Riboflavin cannot be made by the body, so it must be obtained from the food a person eats. The RFVT3 protein is found at especially high levels in cells of the small intestine and is important for absorbing riboflavin during digestion so that the vitamin can be used in the body.

In the cells of the body, riboflavin is the core component of molecules called flavin adenine dinucleotide (FAD) and flavin mononucleotide (FMN). These molecules function as coenzymes, which means they help enzymes carry out chemical reactions. FAD and FMN are involved in many different chemical reactions and are required for a variety of cellular processes. One important role of these coenzymes is in the production of energy for cells. FAD and FMN are also involved in the breakdown (metabolism) of carbohydrates, fats, and proteins.

Health Conditions Related to Genetic Changes

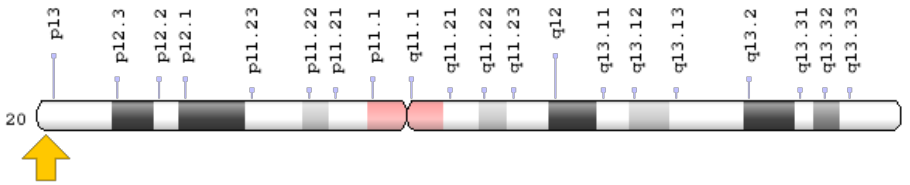
Riboflavin transporter deficiency neuronopathy

More than two dozen mutations in the *SLC52A3* gene have been found to cause riboflavin transporter deficiency neuronopathy. This neurological condition encompasses two disorders that were previously considered to be separate: Brown-Vialetto-Van Laere syndrome and Fazio-Londe disease. The gene mutations involved in this condition lead to production of abnormal RFVT3 proteins. Some mutations lead to the production of an altered protein that cannot get to the cell membrane, so it is unable to transport riboflavin into the cell. Other mutations lead to a version of the protein that can get to the cell membrane, but its function as a transporter is impaired. These changes impair the absorption of riboflavin in the small intestine. The resulting shortage of riboflavin leads to a reduction of FAD and FMN. However, it is unclear how these changes lead to the nerve problems that cause hearing loss, muscle weakness in the face and limbs, and breathing problems in people with the disorder.

Chromosomal Location

Cytogenetic Location: 20p13, which is the short (p) arm of chromosome 20 at position 13

Molecular Location: base pairs 760,080 to 775,985 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bA371L19.1
- BVVLS
- C20orf54
- hRFT2
- MGC10698
- RFT2
- RFT2_HUMAN
- RFVT3
- riboflavin transporter 2
- solute carrier family 52 (riboflavin transporter), member 3
- solute carrier family 52, riboflavin transporter, member 3

Additional Information & Resources

Clinical Information from GeneReviews

- Riboflavin Transporter Deficiency Neuronopathy
  https://www.ncbi.nlm.nih.gov/books/NBK299312

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28riboflavin+transporter+2%5BTIAB%5D%29+OR+%28RFT2%5BTIAB%5D%29+OR+%28BVVLS%5BTIAB%5D%29+OR+%28hRFT2%5BTIAB%5D%29+OR+%28C20orf54%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 3
  http://omim.org/entry/613350

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC52A3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC52A3%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:16187
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:113278
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/113278
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