SLC52A2 gene

solute carrier family 52 member 2

Normal Function

The *SLC52A2* gene provides instructions for making a riboflavin transporter protein called RFVT2 (formerly known as RFT3). This protein moves (transports) a vitamin called riboflavin (also called vitamin B_2) across the cell membrane. The RFVT2 protein is found at especially high levels in cells of the brain and spinal cord and is important for absorbing riboflavin from the bloodstream into these tissues.

In the cells of the body, including those in the brain and spinal cord, 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

At least 11 mutations in the *SLC52A2* 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. Some of the gene mutations involved in riboflavin transporter deficiency neuronopathy prevent production of the RFVT2 protein. Others lead to production of an abnormal protein with impaired ability to transport riboflavin. 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.

Other Names for This Gene

- BVVLS2
- D15Ertd747e
- FLJ11856

- G protein-coupled receptor 172A
- GPCR41
- GPR172A
- hRFT3
- PAR1
- PERV-A receptor 1
- porcine endogenous retrovirus A receptor 1
- putative G-protein coupled receptor GPCR41
- RFT3
- RFVT2
- riboflavin transporter 3
- solute carrier family 52 (riboflavin transporter), member 2
- solute carrier family 52, riboflavin transporter, member 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC52A2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79581[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SLC52A2%5BTIAB%5D%29+OR+%28solute+carrier+family+52++,+member+2%5BTIAB%5D%29%29+OR+%28%28G+protein-coupled+receptor+172A%5BTIAB%5D%29+OR+%28GPCR41%5BTIAB%5D%29+OR+%28GPR172A%5BTIAB%5D%29+OR+%28RFT3%5BTIAB%5D%29+OR+%28RFVT2%5BTIAB%5D%29+OR+%28hRFT3%5BTIAB%5D%29+OR+%28putative+G-protein+coupled+receptor+GPCR41%5BTIAB%5D%29+OR+%28riboflavin+transporter+3%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 2; SLC52A2 (https://omim.org/entry/607882)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/79581)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC52A2[gene])

References

- Cali E, Dominik N, Manole A, Houlden H. Riboflavin Transporter Deficiency.2015 Jun 11 [updated 2021 Apr 8]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R)[Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Availablefrom http://www.ncbi.nlm.nih.gov/books/NBK299312/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26072523)
- Ciccolella M, Corti S, Catteruccia M, Petrini S, Tozzi G, Rizza T, Carrozzo R, Nizzardo M, Bordoni A, Ronchi D, D'Amico A, Rizzo C, Comi GP, Bertini E. Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laeredisease: two novel mutations. J Med Genet. 2013 Feb;50(2):104-7. doi:10.1136/jmedgenet-2012-101204. Epub 2012 Dec 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23243084)
- Haack TB, Makowski C, Yao Y, Graf E, Hempel M, Wieland T, Tauer U, Ahting U, Mayr JA, Freisinger P, Yoshimatsu H, Inui K, Strom TM, Meitinger T, Yonezawa A, Prokisch H. Impaired riboflavin transport due to missense mutations in SLC52A2causes Brown-Vialetto-Van Laere syndrome. J Inherit Metab Dis. 2012Nov; 35(6):943-8. doi: 10.1007/s10545-012-9513-y. Epub 2012 Aug 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22864630) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3470687/)
- Johnson JO, Gibbs JR, Megarbane A, Urtizberea JA, Hernandez DG, Foley AR, Arepalli S, Pandraud A, Simon-Sanchez J, Clayton P, Reilly MM, Muntoni F, Abramzon Y, Houlden H, Singleton AB. Exome sequencing reveals riboflavintransporter mutations as a cause of motor neuron disease. Brain. 2012 Sep; 135(Pt9):2875-82. doi: 10.1093/brain/aws161. Epub 2012 Jun 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22740598) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3437022/)
- Yao Y, Yonezawa A, Yoshimatsu H, Masuda S, Katsura T, Inui K. Identificationand comparative functional characterization of a new human riboflavin transporterhRFT3 expressed in the brain. J Nutr. 2010 Jul;140(7):1220-6. doi:10.3945/jn.110.122911. Epub 2010 May 12. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20463145)
- Yonezawa A, Inui K. Novel riboflavin transporter family RFVT/SLC52:identification, nomenclature, functional characterization and genetic diseases ofRFVT/SLC52. Mol Aspects Med. 2013 Apr-Jun;34(2-3):693-701. doi:10.1016/j.mam.2012.07.014. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23506902)

Genomic Location

The *SLC52A2* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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