

SLCO1B3 gene

solute carrier organic anion transporter family member 1B3

Normal Function

The *SLCO1B3* gene provides instructions for making a protein called organic anion transporting polypeptide 1B3, or OATP1B3. This protein is found in liver cells; it transports compounds from the blood into the liver so that they can be cleared from the body. For example, the OATP1B3 protein transports bilirubin, which is a yellowish substance that is produced when red blood cells are broken down. In the liver, bilirubin is dissolved in a digestive fluid called bile and then excreted from the body. The OATP1B3 protein also transports certain hormones, toxins, and drugs into the liver for removal from the body. Some of the drugs transported by the OATP1B3 protein include statins, which are used to treat high cholesterol; heart disease medications; certain antibiotics; and some drugs used for the treatment of cancer.

Health Conditions Related to Genetic Changes

Rotor syndrome

Mutations in the *SLCO1B3* gene are involved in Rotor syndrome. This condition is characterized by elevated levels of bilirubin in the blood that can cause yellowing of the skin and whites of the eyes (jaundice). For this condition to occur, individuals must have mutations in the *SLCO1B3* gene and a related gene called *SLCO1B1*. This related gene provides instructions for making a protein called OATP1B1, which has a similar transport function to OATP1B3. In some cases, the condition is caused by a deletion of genetic material that removes parts of both the *SLCO1B3* and *SLCO1B1* genes, so no functional OATP1B3 or OATP1B1 protein is made. Most mutations that cause Rotor syndrome lead to abnormally short, nonfunctioning OATP1B3 and OATP1B1 proteins. Without the function of either transport protein, bilirubin is less efficiently taken up by the liver and cleared from the body. The buildup of this substance leads to jaundice in people with Rotor syndrome.

Other Names for This Gene

- HBLRR
- liver-specific organic anion transporter 2
- LST-2

- OATP-8
- OATP1B3
- OATP8
- organic anion transporter 8
- organic anion-transporting polypeptide 8
- SLC21A8
- SO1B3_HUMAN
- solute carrier family 21 (organic anion transporter), member 8
- solute carrier organic anion transporter family, member 1B3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLCO1B3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=28234\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=28234[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SLCO1B3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+180+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER ORGANIC ANION TRANSPORTER FAMILY, MEMBER 1B3; SLCO1B3 (<https://omim.org/entry/605495>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/28234>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLCO1B3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLCO1B3[gene]))

References

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- Niemi M, Pasanen MK, Neuvonen PJ. Organic anion transporting polypeptide 1B1: a genetically polymorphic transporter of major importance for hepatic drug uptake.

Pharmacol Rev. 2011 Mar;63(1):157-81. doi: 10.1124/pr.110.002857. Epub 2011 Jan 18. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21245207>)

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Genomic Location

The *SLCO1B3* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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