

Rotor syndrome

Description

Rotor syndrome is a relatively mild condition characterized by elevated levels of a substance called bilirubin in the blood (hyperbilirubinemia). Bilirubin is produced when red blood cells are broken down. It has an orange-yellow tint, and buildup of this substance can cause yellowing of the skin or whites of the eyes (jaundice). In people with Rotor syndrome, jaundice is usually evident shortly after birth or in childhood and may come and go; yellowing of the whites of the eyes (also called conjunctival icterus) is often the only symptom.

There are two forms of bilirubin in the body: a toxic form called unconjugated bilirubin and a nontoxic form called conjugated bilirubin. People with Rotor syndrome have a buildup of both unconjugated and conjugated bilirubin in their blood, but the majority is conjugated.

Frequency

Rotor syndrome is a rare condition, although its prevalence is unknown.

Causes

The *SLCO1B1* and *SLCO1B3* genes are involved in Rotor syndrome. Mutations in both genes are required for the condition to occur. The *SLCO1B1* and *SLCO1B3* genes provide instructions for making similar proteins, called organic anion transporting polypeptide 1B1 (OATP1B1) and organic anion transporting polypeptide 1B3 (OATP1B3), respectively. Both proteins are found in liver cells; they transport bilirubin and other compounds from the blood into the liver so that they can be cleared from the body. In the liver, bilirubin is dissolved in a digestive fluid called bile and then excreted from the body.

The *SLCO1B1* and *SLCO1B3* gene mutations that cause Rotor syndrome lead to abnormally short, nonfunctional OATP1B1 and OATP1B3 proteins or an absence of these proteins. Without the function of either transport protein, bilirubin is less efficiently taken up by the liver and removed from the body. The buildup of this substance leads to jaundice in people with Rotor syndrome.

[Learn more about the genes associated with Rotor syndrome](https://medlineplus.gov/genetics/)

- *SLCO1B1*
- *SLCO1B3*

Inheritance

This condition is inherited in an autosomal recessive pattern. In autosomal recessive inheritance, both copies of a gene in each cell have mutations. In Rotor syndrome, an affected individual must have mutations in both the *SLCO1B1* and the *SLCO1B3* gene, so both copies of the two genes are altered. The parents of an individual with this condition each carry one altered copy of both genes, but they do not show signs and symptoms of the condition.

Other Names for This Condition

- Hyperbilirubinemia, Rotor type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Rotor syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220991/>)

Genetic and Rare Diseases Information Center

- Rotor syndrome (<https://rarediseases.info.nih.gov/diseases/218/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- HYPERBILIRUBINEMIA, ROTOR TYPE; HBLRR (<https://omim.org/entry/237450>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28rotor+syndrome%5BTIAB%5D%29+OR+%28rotor+type+hyperbilirubinemia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

References

- Strassburg CP. Hyperbilirubinemia syndromes (Gilbert-Meulengracht, Crigler-Najjar, Dubin-Johnson, and Rotor syndrome). *Best Pract Res Clin Gastroenterol*. 2010 Oct; 24(5):555-71. doi: 10.1016/j.bpg.2010.07.007. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20955959>)
- van de Steeg E, Stranecky V, Hartmannova H, Noskova L, Hrebicek M, Wagenaar E, van Esch A, de Waart DR, Oude Elferink RP, Kenworthy KE, Sticova E, al-Edreesi M, Knisely AS, Knoch S, Jirsa M, Schinkel AH. Complete OATP1B1 and OATP1B3 deficiency causes human Rotor syndrome by interrupting conjugated bilirubin reuptake into the liver. *J Clin Invest*. 2012 Feb; 122(2):519-28. doi:10.1172/JCI59526. Epub 2012 Jan 9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/2232210>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3266790/>)
- Wolkoff AW, Wolpert E, Pascasio FN, Arias IM. Rotor's syndrome. A distinct inheritable pathophysiologic entity. *Am J Med*. 1976 Feb; 60(2):173-9. doi:10.1016/0002-9343(76)90426-5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/766621>)

Last updated March 1, 2013