Rotor syndrome

Rotor syndrome (OMIM #237450 (https://omim.org/entry/237450)) is one of the causes of **juvenile conjugated hyperbilirubinemias'**. It is a rare autosomal recessive disease. It appears with the simultaneous defect of two organic anion transporting polypeptides (OATPs), proteins OATP1B1 and OATP1B3 (products of the genes SLCO1B1 and SLCO1B3).

The symptoms of Rotor syndrome are very similar to Dubin-Johnson syndrome. Shortly after birth or in infancy, mild conjugated hyperbilirubinemia develops and '*bilirubinuria* appears. Both syndromes can be distinguished by special examinations (with Rotor syndrome, the liver absorbs diagnostic substances of an anionic nature more slowly; Dubin-Johnson syndrome, on the other hand, is characterized by pigment deposits in hepatocytes).

The frequency of Rotor syndrome is estimated to be approximately **1 in 1 million'**; however, it is thought to vary widely in different populations.

Since the OATP1B1/3 transporters, in addition to conjugated bilirubin, take up a number of other substances **including many drugs**, it is assumed that some drugs administered in the usual dosage may also show life-threatening toxicity in patients with Rotor syndrome.

Links

Related Articles

- Juvenile hyperbilirubinemia

References

- VAN DE STEEG, Evita, Viktor STRÁNECKÝ a Hana HARTMANNOVÁ, et al. Complete OATP1B1 and OATP1B3 deficiency causes human Rotor syndrome by interrupting conjugated bilirubin reuptake into the liver. *J Clin Invest* [online]. 2012, vol. 122, no. 2, s. 519-28, dostupné také z <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3266790/?tool=pubmed>. ISSN 0021-9738 (print), 1558-8238. DOI: 10.1172/JCI59526.