Defective SLCO1B3 causes hyperbilirubinemia, Rotor type (HBLRR)

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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

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Literature references


Reactome database release: 72

This document contains 1 pathway and 1 reaction (see Table of Contents)

https://release.reactome.org
Defective SLCO1B3 causes hyperbilirubinemia, Rotor type (HBLRR)

Stable identifier: R-HSA-5619058

Diseases: bilirubin metabolic disorder

In the body, solute carrier organic anion transporter family member 1B3 (SLCO1B3) is expressed on the basolateral surfaces of hepatocytes and may play a role in the uptake of bilirubin (BIL), a breakdown product of heme that requires conjugation and excretion from the body. Defects in SLCO1B3 can cause hyperbilirubinemia, Rotor type (HBLRR; MIM:237450), an autosomal recessive form of primary conjugated hyperbilirubinemia. Mild jaundice, not associated with hemolysis, develops shortly after birth or in childhood (van de Steeg et al. 2012, Sticova & Jirsa 2013, Keppler 2014).

Literature references


Editions

<table>
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<th>Action</th>
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Defective SLCO1B3 does not transport BIL from extracellular region (blood) to cytosol (hepatocyte)

Location: Defective SLCO1B3 causes hyperbilirubinemia, Rotor type (HBLRR)

Stable identifier: R-HSA-5661198