Defective SLC3A1 causes cystinuria (CSNU)

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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)
Defective SLC3A1 causes cystinuria (CSNU)

Stable identifier: R-HSA-5619113

Diseases: cystinuria

Neutral and basic amino acid transport protein rBAT (SLC3A1) and b(0,+)-type amino acid transporter 1 (SLC7A9) are linked by a disulfide bridge to form system b(0,+)-like activity in the high affinity transport of neutral and dibasic amino acids and cystine. The SLC7A9:SLC3A1 heterodimer mediates the electrogenic exchange of extracellular amino acids such as L-arginine (L-Arg) and L-lysine (L-lys) and cystine (CySS-, the oxidised form of L-cysteine) for intracellular neutral amino acids such as L-leucine (L-Leu). These solute carriers are mainly expressed in the kidney and small intestine where they remove dibasic amino acids and cystine from the renal tubular and intestinal lumen respectively (Schweikhard & Ziegler 2012). Defects in SLC3A1 (or SLC7A9) can cause cystinuria (CSNU; MIM:220100), an autosomal recessive disorder characterised by impaired epithelial cell transport of cystine and dibasic amino acids in the proximal renal tubule and GI tract. The build-up and low solubility of cystine causes the formation of calculi in the urinary tract, resulting in obstructive uropathy, pyelonephritis and in severe cases, renal failure (Palacin et al. 2001, Mattoo & Goldfarb 2008, Fotiadis et al. 2013, Saravakos et al. 2014). Cystinuria is subcategorized as type A (mutations on SLC3A1) and type B (mutations on SLC7A9).

Literature references


Editions

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<tr>
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Defective SLC3A1 (in SLC7A9:SLC3A1) does not exchange L-Arg, CySS-, L-Lys for L-Leu

Location: Defective SLC3A1 causes cystinuria (CSNU)

Stable identifier: R-HSA-5655702