Defective SLC6A19 causes Hartnup disorder (HND)

Broer, S., Jassal, B.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of Creative Commons Attribution 4.0 International (CC BY 4.0) License. For more information see our license.

11/05/2020
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.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references


Reactome database release: 72

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective SLC6A19 causes Hartnup disorder (HND)

Stable identifier: R-HSA-5659735

Diseases: Hartnup disease

SLC6A19 encodes the sodium-dependent neutral amino acid transporter B(0)AT1 and mediates the uptake of neutral amino acids across the plasma membrane accompanied by uptake of a sodium ion. The protein is abundantly expressed in the small intestine and kidney (Broer & Gether 2012, Schweikhard & Ziegler 2012). Defects in SLC6A19 can cause Hartnup disorder (HND; MIM:234500), an autosomal recessive abnormality of renal and gastrointestinal neutral amino acid transport characterised by increased urinary and intestinal excretion of neutral amino acids. Symptoms include transient manifestations of rashes, cerebellar ataxia and psychotic behaviour (Broer 2009, Cheon et al. 2010). Some mutations in SLC6A19 are thought to contribute to the phenotypes iminoglycinuria (IG; MIM:242600) and hyperglycinuria (HG; MIM:138500) (Broer et al. 2008).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2014-08-22</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2015-08-04</td>
<td>Reviewed</td>
<td>Broer, S.</td>
</tr>
</tbody>
</table>
Defective SLC6A19 does not cotransport neutral amino acids, Na+ from extracellular region to cytosol

Location: Defective SLC6A19 causes Hartnup disorder (HND)

Stable identifier: R-HSA-5659734