Variant SLC6A20 contributes towards hyperglycinuria (HG) and iminoglycinuria (IG)

Broer, S., Jassal, B.
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)
Variant SLC6A20 contributes towards hyperglycinuria (HG) and iminoglycinuria (IG)

Stable identifier: R-HSA-5619101

Diseases: amino acid metabolic disorder

SLC6A20 encodes the sodium- and chloride-dependent transporter SIT1 and mediates the sodium-dependent uptake of imino acids such as L-proline, N-methyl-L-proline and pipecolate as well as N-methylated amino acids and glycine (Broer & Gether 2012, Schweikhard & Ziegler 2012). The human protein is expressed in the intestine and kidney. A common SNP in the SLC6A20 gene, a 596C-T transition that results in a thr199-to-met (T199M) substitution can contribute towards iminoglycinuria (IG; MIM:242600) or hyperglycinuria (HG; MIM:138500) (Broer et al. 2008). Overall, mutations in SLC36A2 together with polymorphisms in the modifiers SLC6A20, SLC6A18, and SLC6A19 constitute the genetic basis for these phenotypes.

Literature references


Editions

<table>
<thead>
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<th>Date</th>
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<tbody>
<tr>
<td>2014-08-22</td>
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<td>Jassal, B.</td>
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Variant SLC6A20 does not cotransport L-Pro, Na+ from extracellular to cytosol

**Location:** Variant SLC6A20 contributes towards hyperglycinuria (HG) and iminoglycinuria (IG)

**Stable identifier:** R-HSA-5660694