Defective SLC12A3 causes Gitelman syndrome (GS)

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)
Defective SLC12A3 causes Gitelman syndrome (GS)

Stable identifier: R-HSA-5619087

Diseases: Gitelman syndrome

The SLC12A3 gene encodes for the Thiazide-sensitive sodium-chloride cotransporter (TSC). TSC mediates sodium and chloride removal from the distal convoluted tubule of the kidney. Defects in SLC12A3 are the cause of Gitelman syndrome (GS aka familial hypokalemic hypomagnesemia; MIM:263800). GS is an autosomal recessive disorder characterised by hypokalemic metabolic alkalosis, hypomagnesemia, and hypocalciuria. Patients can present with periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. GS has overlapping features with Bartter syndrome (caused by defects in SLC12A1). This cotransporter is the major target for thiazide-type diuretics, used in the treatment of hypertension, extracellular fluid overload and renal stone disease (Nakhoul et al. 2012).

Literature references


Editions

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<td>2014-08-22</td>
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<td>Jassal, B.</td>
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Defective SLC12A3 does not cotransport Cl-, Na+ from extracellular region to cytosol

**Location:** Defective SLC12A3 causes Gitelman syndrome (GS)

**Stable identifier:** R-HSA-5623705