Defective SLC17A5 causes Salla disease (SD) and ISSD

Broer, S., Jassal, B.

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

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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 SLC17A5 causes Salla disease (SD) and ISSD

Stable identifier: R-HSA-5619035

Diseases: inherited metabolic disorder

SLC17A5 encodes a lysosomal sialic acid transporter, sialin (AST, membrane glycoprotein HP59) which exports sialic acid (N-acetylenuraminic acid, Neu5Ac) derived from the degradation of glycoconjugates from lysosomes. This export is dependent on the proton electrochemical gradient across the lysosomal membrane. SLC17A5 is present in the pathological tumor vasculature of the lung, breast, colon, and ovary, but not in the normal vasculature, suggesting that the protein may be critical to pathological angiogenesis. Sialin is not expressed in a variety of normal tissues, but is significantly expressed in human fetal lung. Defects in SLC17A5 cause Salla disease (SD) and infantile sialic acid storage disorder (ISSD aka N-acetylenuraminic acid storage disease, NSD). These diseases belong to the sialic acid storage diseases (SASDs) and are autosomal recessive neurodegenerative disorders characterised by hypotonia, cerebellar ataxia and mental retardation with patients excreting large amounts of free Neu5Ac in urine. ISSD is a severe infantile form of SASD with a more severe clinical course than SD (Verheijen et al. 1999, Aula et al. 2000).

Literature references


Editions

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Defective SLC17A5 does not cotransport Neu5Ac, H+ from lysosomal lumen to cytosol

**Location:** Defective SLC17A5 causes Salla disease (SD) and ISSD

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