Defective SLC29A3 causes histiocytosis-lymphadenopathy plus syndrome (HLAS)

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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 SLC29A3 causes histiocytosis-lymphadenopathy plus syndrome (HLAS)

Stable identifier: R-HSA-5619063

Diseases: histiocytosis

The human gene SLC29A3 encodes the equilibrative nucleoside transporter 3 (ENT3). It is abundant in many tissues, especially the placenta and is localized intracellularly on the lysosomal membrane. SLC29A3 mediates the reversible transport of nucleosides as well as anticancer and antiviral agents such as cladribine, cordycepin, tubercidin and AZT. Defects in SLC29A3 can cause histiocytosis-lymphadenopathy plus syndrome (HLAS; MIM:602782), an autosomal recessive disorder characterised by combined features from 2 or more of four histiocytic disorders (Morgan et al. 2010, Colmenero et al. 2012, Young et al. 2013).

Literature references


Editions

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Defective SLC29A3 does not transport nucleosides from lysosomal lumen to cytosol

Location: Defective SLC29A3 causes histiocytosis-lymphadenopathy plus syndrome (HLAS)

Stable identifier: R-HSA-5628807