Disorders of transmembrane transporters

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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: 81

This document contains 3 pathways (see Table of Contents)

https://reactome.org
Disorders of transmembrane transporters

Stable identifier: R-HSA-5619115

Proteins with transporting functions can be roughly classified into 3 categories: ATP hydrolysis-coupled pumps, ion channels, and transporters. Pumps utilize the energy released by ATP hydrolysis to power the movement of substrates across the membrane against their electrochemical gradient. Channels in their open state can transfer substrates (ions or water) down their electrochemical gradient at an extremely high efficiency (up to 108 s⁻¹). Transporters facilitate the movement of a specific substrate either against or with their concentration gradient at a lower speed (about 102 -104 s⁻¹); as generally believed, conformational change of the transporter protein is involved in the transfer process. Diseases caused by defects in these transporter proteins are detailed in this section. Disorders associated with ABC transporters and SLC transporters are annotated here (Dean 2005).

Literature references


Editions

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The ATP-binding cassette (ABC) transporters form a large family of transmembrane proteins that utilise the energy from the hydrolysis of ATP to facilitate the movement of a wide variety of substrates against a concentration gradient across membrane bilayers. Substrates include amino acids, lipids, inorganic ions, peptides, saccharides, peptides for antigen presentation, metals, drugs, and proteins. Of the 48 known ABC transporters in humans, 15 are associated with a defined human disease (Tarling et al. 2013, Woodward et al. 2011, Kemp et al. 2011, Ueda 2011, Chen & Tiwari 2011).

**Literature references**


SLC transporter disorders

Location: Disorders of transmembrane transporters

Stable identifier: R-HSA-5619102

The solute-carrier gene (SLC) superfamily encodes membrane-bound transporters comprising 55 gene families with at least 362 putatively functional protein-coding genes. The gene products include passive transporters, symporters and antiporters and are located in all cellular and organelle membranes. Curated here is a list of SLCs, where mutations within them can result in disease (Hediger et al. 2013).

Literature references


Editions

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