Defective SLC26A4 causes Pendred syndrome (PDS)

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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 SLC26A4 causes Pendred syndrome (PDS)

Stable identifier: R-HSA-5619046

Diseases: goiter, sensorineural hearing loss

Solute carrier (SLC) genes that code chloride (Cl-) / bicarbonate (HCO3-) exchanger proteins are in the SLC4 and SLC26 families. SLC26A4 (pendrin) is thought to act as a chloride/anion exchanger but in the thyroid and inner ear, it also contributes to the conditioning of the endolymphatic fluid by mediating iodide (I-) transport. Defects in SLC26A4 can cause Pendred syndrome (PDS; MIM:274600), an autosomal recessive disorder characterised by congenital sensorineural hearing loss in association with thyroid goiter (Choi et al. 2011, Pesce & Kopp 2014).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Author(s)</th>
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<tbody>
<tr>
<td>2014-08-22</td>
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<td>Jassal, B.</td>
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SLC26A4 does not transport I- from cytosol to extracellular region

**Location:** Defective SLC26A4 causes Pendred syndrome (PDS)

**Stable identifier:** R-HSA-5627870