Defective ABCA3 causes pulmonary surfactant metabolism dysfunction 3 (SMDP3)

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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 ABCA3 causes pulmonary surfactant metabolism dysfunction 3 (SMDP3)

Stable identifier: R-HSA-5688399

Diseases: newborn respiratory distress syndrome, interstitial lung disease

ATP-binding cassette sub-family A member 3 (ABCA3) plays an important role in the formation of pulmonary surfactant, probably by transporting phospholipids such as phosphatidylcholine (PC) and phosphatidylglycerol (PG) from the ER membrane to lamellar bodies (LBs). PC and PG are the major phospholipid constituents of pulmonary surfactant. LBs are the surfactant storage organelles of type II epithelial cells from where phospholipids can be secreted together with surfactant proteins (SFTPs) into the alveolar airspace. Defects in ABCA3 can cause pulmonary surfactant metabolism dysfunction type 3 (S-MDP3; MIM:610921), resulting in respiratory distress in newborns and interstitial lung disease (ILD) in children (Whitsett et al. 2015).

Literature references


Editions

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<tr>
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Defective ABCA3 does not transport PC, PG from ER membrane to lamellar body

Location: Defective ABCA3 causes pulmonary surfactant metabolism dysfunction 3 (SMDP3)

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