Defective ABCD1 does not transfer LCFAs from cytosol to peroxisomal matrix

Jassal, B., Shukla, S.
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 reaction (see Table of Contents)
Defective ABCD1 does not transfer LCFAs from cytosol to peroxisomal matrix

**Stable identifier:** R-HSA-5684043

**Type:** transition

**Compartments:** peroxisomal membrane, cytosol

**Diseases:** adrenoleukodystrophy

The 70-kDa peroxisomal membrane protein (PMP70) and the adrenoleukodystrophy protein (ALDP aka ABCD1) are half ATP binding cassette (ABC) transporters in the peroxisome membrane. They are involved in metabolic transport of long and very long chain fatty acids into peroxisomes. Mutations in the ALD gene result in the X-linked neurodegenerative disorder adrenoleukodystrophy (ALD; MIM:300100). ABCD1 deficiency impairs the peroxisomal beta-oxidation of very long-chain fatty acids (VLCFA) and facilitates their further chain elongation by ELOVL1 resulting in accumulation of VLCFA in plasma and tissues. While all patients with ALD have mutations in the ABCD1 gene, there is no general genotype-phenotype correlation. Mutations causing ALD include R617C, S606L, M1V, G277R, R554H and W77_L82del (Krasemann et al. 1996, Fanen et al. 1994, Engelen et al. 2011, Coll et al. 2005, Park et al. 2014).

**Literature references**


## Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Role</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2015-03-18</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
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