Defective CYP27B1 causes VDDR1A

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This is just an excerpt of a full-length report for this pathway. To access the complete report, please download it at the Reactome Textbook.

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https://reactome.org
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: 83

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective CYP27B1 causes VDDR1A

Stable identifier: R-HSA-5579014

Diseases: rickets

Vitamin D3 (cholecalciferol), synthesised in human skin by ultraviolet radiation action on 7-dehydrocholesterol, does not possess any biological activity. Vitamin D hormonal activity requires hydroxylation at the 25 and 1-alpha positions by cytochrome P450 enzymes CYP2R1 and CYP27B1 respectively. Vitamin D 25-hydroxylase (CYP2R1) catalyses the hydroxylation of vitamin D3 to calcidiol (CDL). Subsequent 1-alpha-hydroxylation of CDL by CYP27B1 produces calcitriol (CTL). CTL binds and activates the nuclear vitamin D receptor, with subsequent regulation of physiologic events such as calcium homeostasis, cellular differentiation and proliferation.

Defects in CYP27B1 can cause rickets, vitamin D-dependent 1A (VDDR1A; MIM:264700), a disorder caused by deficiency of the active form of vitamin D (CTL) resulting in defective bone mineralization and clinical features of rickets. To date, 47 mutations have been identified, the majority of them (28) being missense mutations (Kim 2011, Cui et al. 2012).

Literature references


Editions

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Defective CYP27B1 does not hydroxylate CDL

Location: Defective CYP27B1 causes VDDR1A

Stable identifier: R-HSA-5602186

Type: transition

Compartments: cytosol, mitochondrial outer membrane

Diseases: rickets


Literature references


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Table of Contents

Introduction 1

Defective CYP27B1 causes VDDR1A 2

Defective CYP27B1 does not hydroxylate CDL 3

Table of Contents 5