Defective CYP27B1 does not hydroxylate CDL

Jassal, B., Nakaki, T.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

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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

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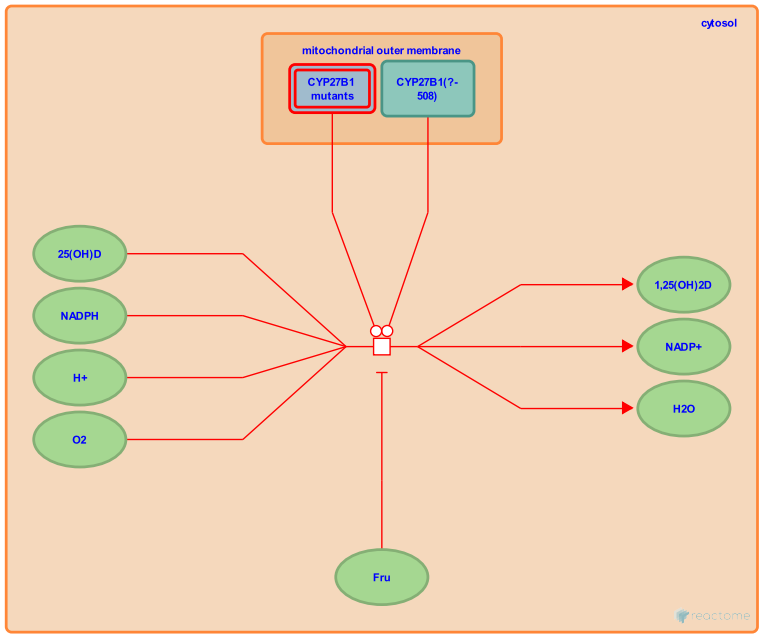
Stable identifier: R-HSA-5602186

Type: transition

Compartments: mitochondrial outer membrane, cytosol

Diseases: rickets



The second step in vitamin D3 activation requires hydroxylation of 25-hydroxyvitamin D3 (calcidiol) to 1alpha-25-dihydroxyvitamin D3 (calcitriol). This conversion is mediated by 25-hydroxyvitamin D-1alpha hydroxylase (CYP27B1) (Zehnder et al. 2002, Fritsche et al. 2003). 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 (Kim 2011). Mutations causing complete loss of function of CYP27B1 include R107H, G125E, R335P, P382S, R389G, R389H and D320Tfs*32 (Kitanaka et al. 1998, Wang et al. 2002, Wang et al. 1998).

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Editions

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