Defective CYP17A1 does not 17-hydroxylate PREG

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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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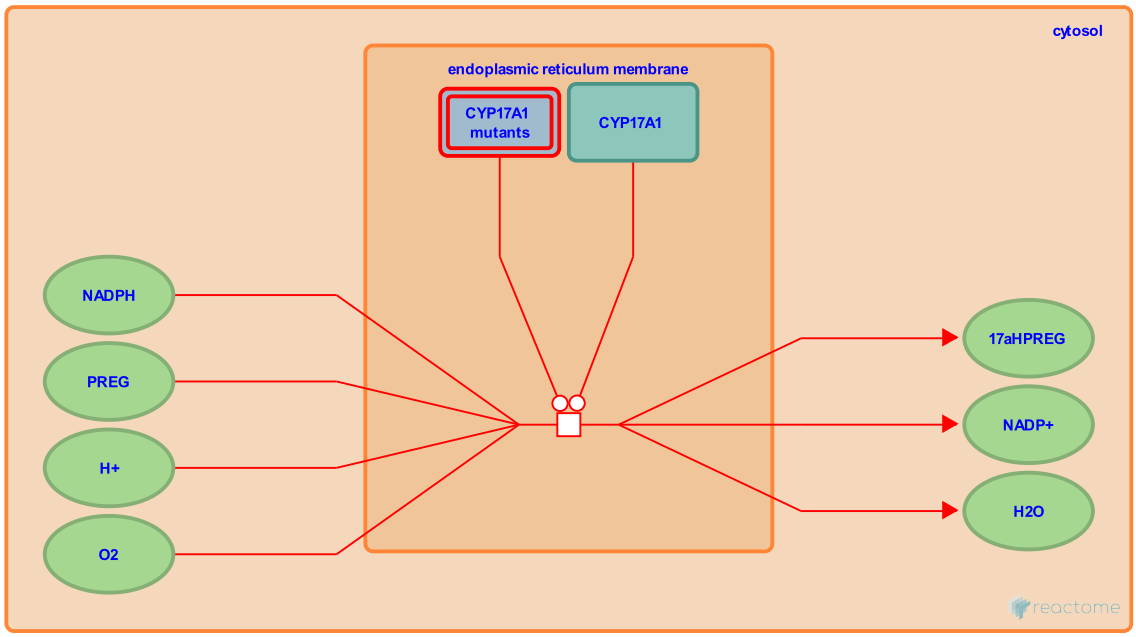
Stable identifier: R-HSA-5601843

Type: transition

Compartments: cytosol, endoplasmic reticulum membrane

Diseases: adrenal gland disease



Steroid 17-alpha-hydroxylase/17,20 lyase (CYP17A1) mediates both 17-alpha-hydroxylase and 17,20-lyase activity, allowing the adrenal glands and gonads to synthesise both 17-alpha-hydroxylated glucocorticoids and sex steroids respectively (Kagimoto et al. 1998). Defects in CYP17A1 can cause Adrenal hyperplasia 5 (AH5), a form of congenital adrenal hyperplasia (CAH), a common recessive disease due to defective synthesis of cortisol and sex steroids. Mutations causing combined 17-alpha-hydroxylase/17,20-lyase deficiency include S106P, R96W, W17*, R362C, W406R and R96Q (Lin et al. 1991, Laflamme et al. 1996, Suzuki et al. 1998, Martin et al. 2003, Brooke et al. 2006). Mutations causing isolated 17,20-lyase deficiency are R358Q and R347H (Geller et al. 1997, Van den Akker et al. 2002).

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Editions

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