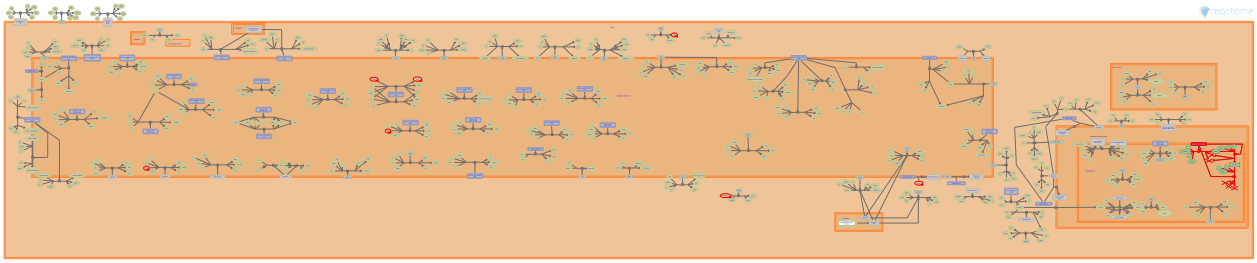
Defective CYP11B2 causes CMO-1 deficiency



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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Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

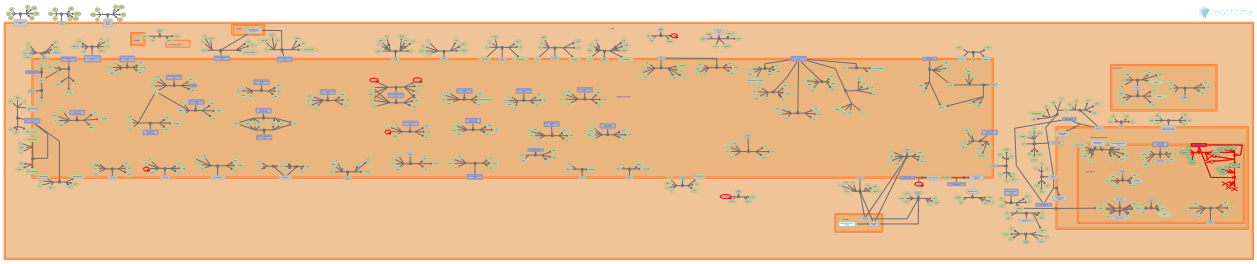
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology, 14*, e1005968. ↗

Defective CYP11B2 causes CMO-1 deficiency ↗

Stable identifier: R-HSA-5579009

Diseases: steroid inherited metabolic disorder



Cytochrome P450 11B2, mitochondrial (CYP11B2 aka aldosterone hydroxylase) is an enzyme necessary for aldosterone biosynthesis via corticosterone (CORST) and 18-hydroxycorticosterone (18HCORST). Defects in CYP11B2 results in disorders of aldosterone synthesis. Corticosterone methyloxidase 1 and 2 deficiencies (CMO-1; MIM:203400 and CMO-2 deficiency; MIM:61060) are autosomal recessive disorders of aldosterone biosynthesis (Mitsuuchi et al. 1993, Bureik et al. 2002). In CMO-1 deficiency, aldosterone is undetectable in plasma, while its immediate precursor, 18HCORST, is low or normal. In CMO-2 deficiency, aldosterone can be low or normal, but at the expense of increased secretion of 18HCORST. Patients with CMO-2 deficiency have elevated plasma 18-hydroxycorticosterone/aldosterone ratios.

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Bureik, M., Bernhardt, R., Lisurek, M. (2002). The human steroid hydroxylases CYP1B1 and CYP11B2. *Biol. Chem., 383*, 1537-51. ↗

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Editions

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Defective CYP11B2 does not oxidise 11DCORST ↗

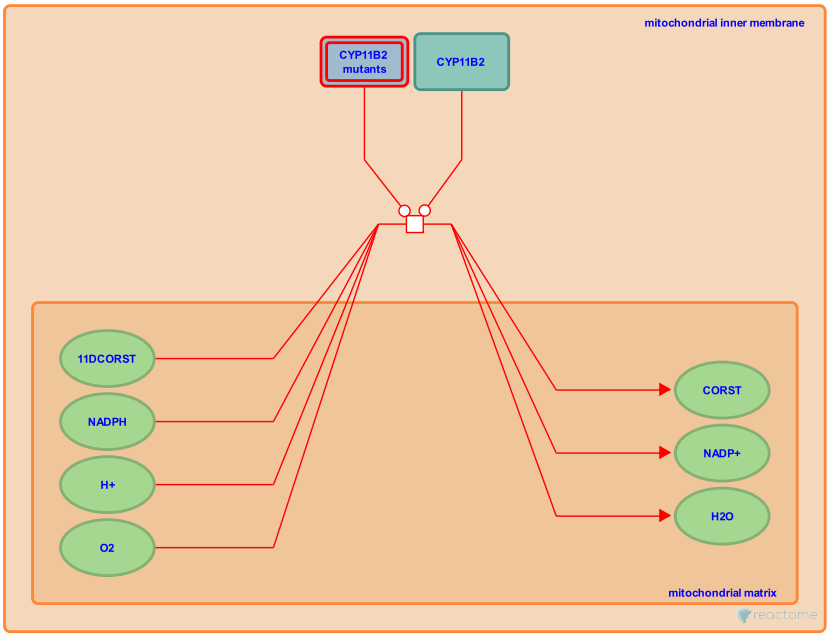
Location: Defective CYP11B2 causes CMO-1 deficiency

Stable identifier: R-HSA-5600598

Type: transition

Compartments: mitochondrial inner membrane, mitochondrial matrix

Diseases: steroid inherited metabolic disorder



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Literature references

Rösler, A., Pascoe, L., White, PC., Curnow, KM., Slutsker, L. (1992). Mutations in the human CYP11B2 (aldosterone synthase) gene causing corticosterone methyloxidase II deficiency. *Proc. Natl. Acad. Sci. U.S.A., 89*, 4996-5000. ↗

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Crock, PA., Montalto, J., Curnow, KM., Dunlop, FM., Funder, JW. (2003). A compound heterozygote case of type II aldosterone synthase deficiency. *J. Clin. Endocrinol. Metab., 88*, 2518-26. ↗

Sólyom, J., Bünger, K., Peter, M., Sippell, WG. (1998). Mutation THR-185 ILE is associated with corticosterone methyl oxidase deficiency type II. *Eur. J. Pediatr., 157*, 378-81. ↗

Editions

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2014-11-03	Reviewed	Nakaki, T.

Defective CYP11B2 does not oxidise 18HCORST ↗

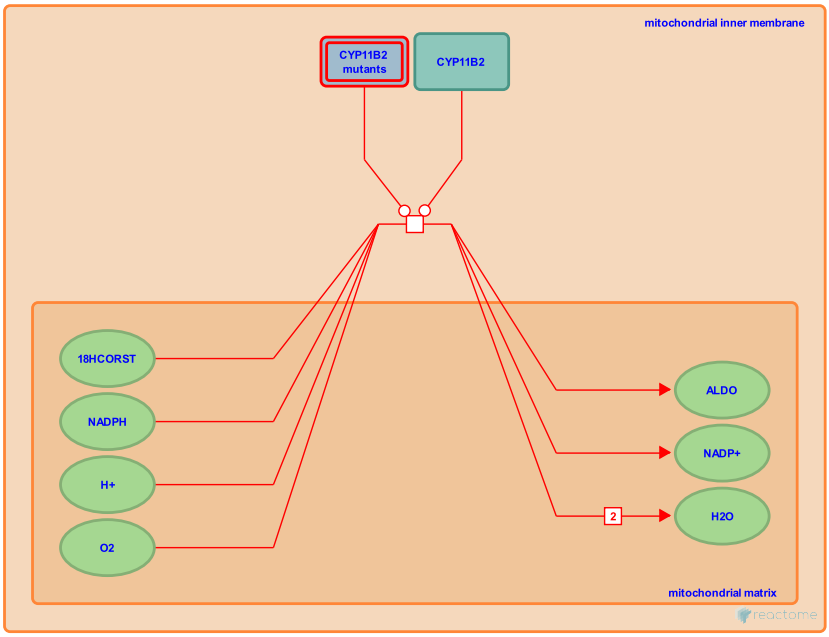
Location: Defective CYP11B2 causes CMO-1 deficiency

Stable identifier: R-HSA-6785244

Type: transition

Compartments: mitochondrial inner membrane, mitochondrial matrix

Diseases: steroid inherited metabolic disorder



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Crock, PA., Montalto, J., Curnow, KM., Dunlop, FM., Funder, JW. (2003). A compound heterozygote case of type II aldosterone synthase deficiency. *J. Clin. Endocrinol. Metab., 88*, 2518-26. ↗

Sólyom, J., Bünger, K., Peter, M., Sippell, WG. (1998). Mutation THR-185 ILE is associated with corticosterone methyl oxidase deficiency type II. *Eur. J. Pediatr., 157*, 378-81. ↗

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Defective CYP11B2 does not oxidise CORST ↗

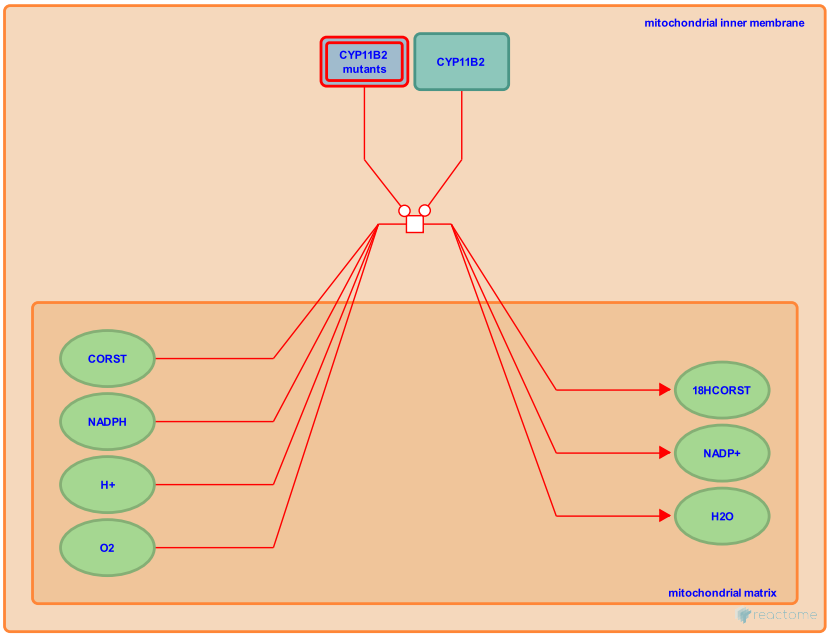
Location: Defective CYP11B2 causes CMO-1 deficiency

Stable identifier: R-HSA-6785245

Type: transition

Compartments: mitochondrial inner membrane, mitochondrial matrix

Diseases: steroid inherited metabolic disorder



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