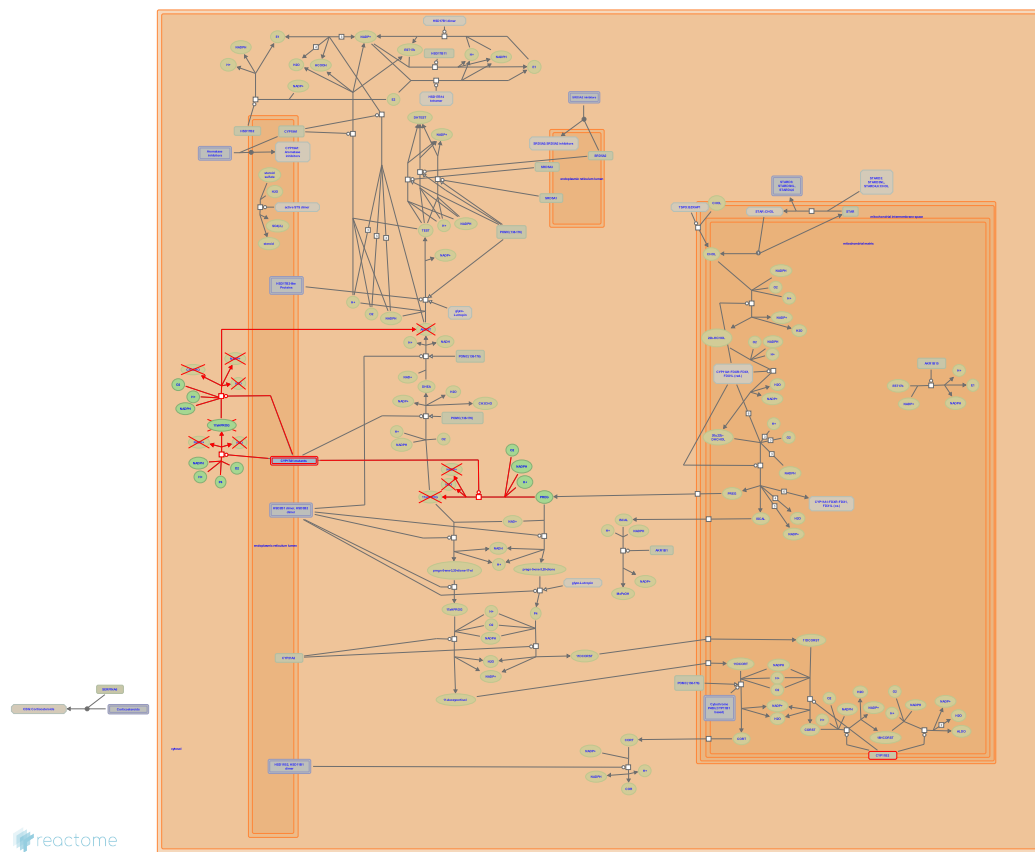


Defective CYP17A1 causes AH5



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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](https://reactome.org/about/reactome-textbook/).

11/05/2024

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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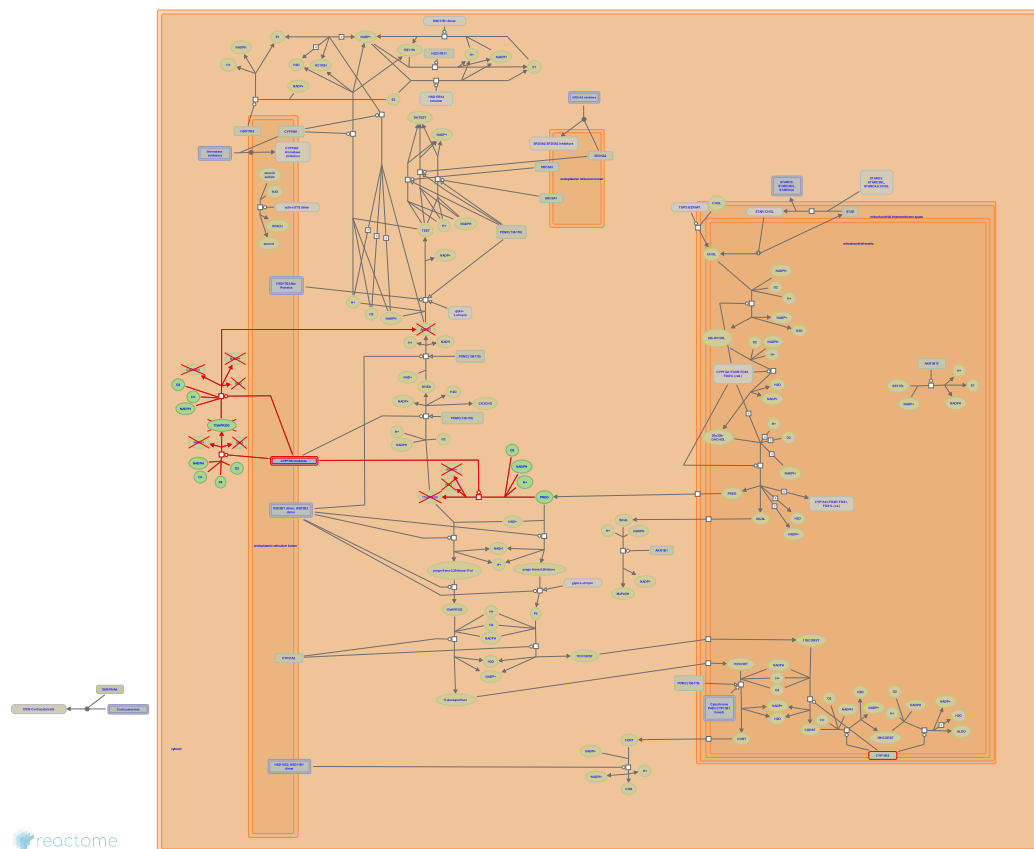
Reactome database release: 88

This document contains 1 pathway and 3 reactions ([see Table of Contents](#))

Defective CYP17A1 causes AH5 ↗

Stable identifier: R-HSA-5579028

Diseases: adrenal gland disease



Steroid 17- α -hydroxylase/17,20 lyase (CYP17A1) mediates both 17- α -hydroxylase and 17,20-lyase activity, allowing the adrenal glands and gonads to synthesise both 17- α -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. Common symptoms include mild hypocortisolism, ambiguous genitalia in genetic males or failure of the ovaries to function at puberty in genetic females, metabolic alkalosis due to hypokalemia and low-renin hypertension. CYP17A1 can have defects in either or both of 17- α -hydroxylase and 17,20-lyase activities thus patients can show combined partial 17- α -hydroxylase/17,20-lyase deficiency or isolated 17,20-lyase deficiency traits (Yanase et al. 1992, Kater & Biglieri 1994, Fluck & Miller 2006, Miller 2012).

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Editions

2014-06-06	Authored, Edited	Jassal, B.
2014-11-03	Reviewed	Nakaki, T.

Defective CYP17A1 does not 17-hydroxylate PREG ↗

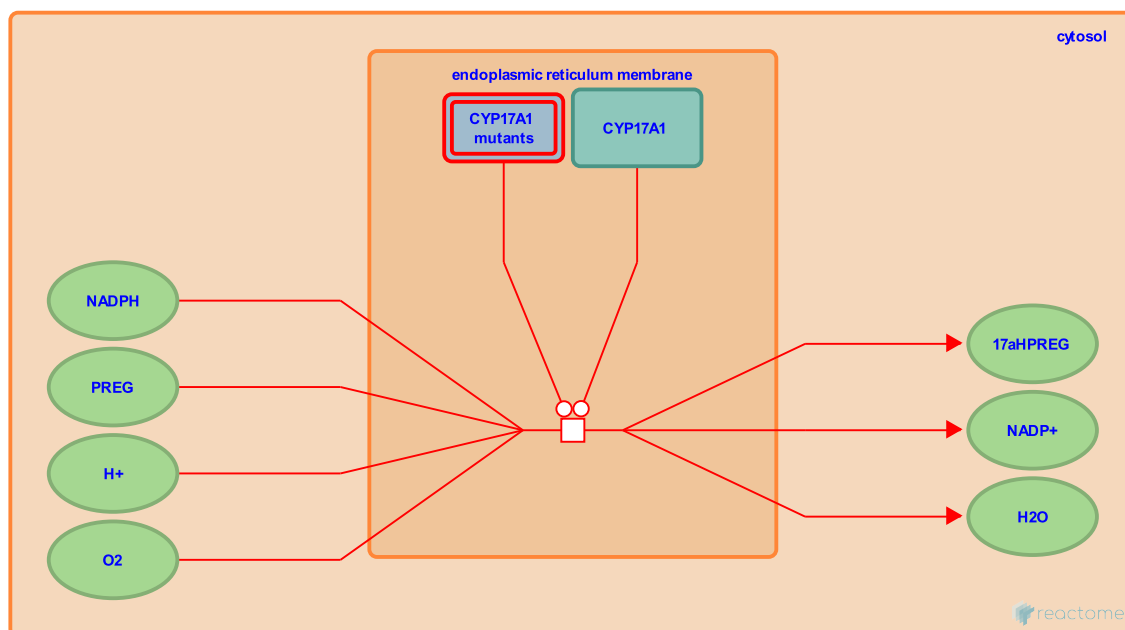
Location: Defective CYP17A1 causes AH5

Stable identifier: R-HSA-5601843

Type: transition

Compartments: endoplasmic reticulum membrane, cytosol

Diseases: adrenal gland disease



Steroid 17- α -hydroxylase/17,20 lyase (CYP17A1) mediates both 17- α -hydroxylase and 17,20-lyase activity, allowing the adrenal glands and gonads to synthesise both 17- α -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- α -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).

Literature references

- Martin, RM., de Oliveira, ML., Costa, EM., Villar, H., Lin, CJ., Longui, CA. et al. (2003). P450c17 deficiency in Brazilian patients: biochemical diagnosis through progesterone levels confirmed by CYP17 genotyping. *J. Clin. Endocrinol. Metab.*, 88, 5739-46. ↗
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Defective CYP17A1 does not 17-hydroxylate P4 [↗](#)

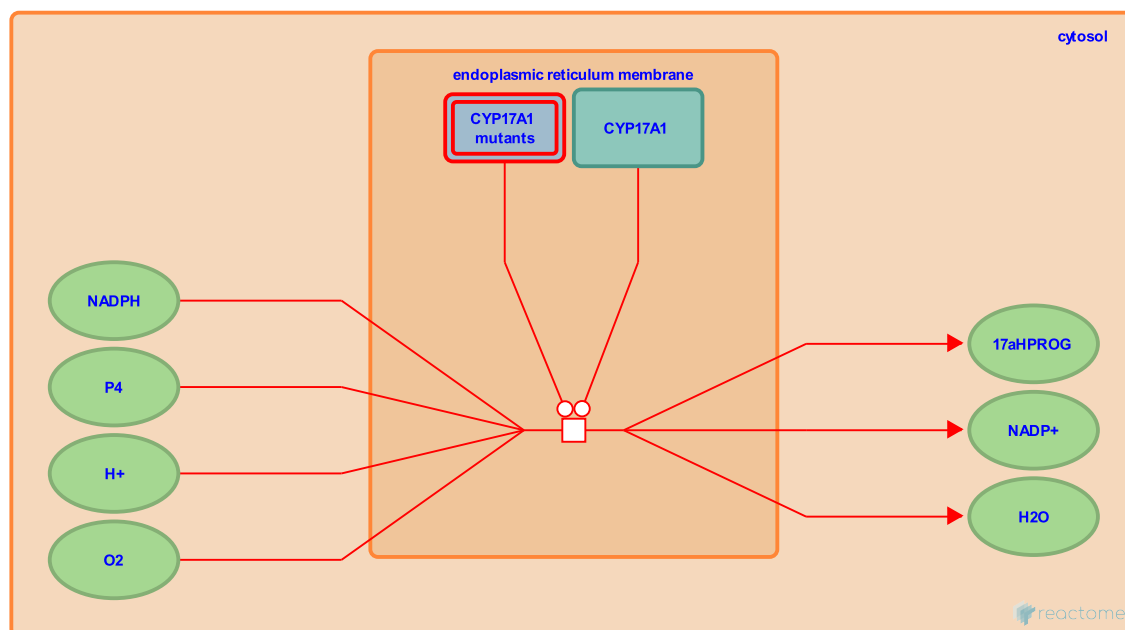
Location: [Defective CYP17A1 causes AH5](#)

Stable identifier: R-HSA-9035954

Type: transition

Compartments: endoplasmic reticulum membrane, cytosol

Diseases: adrenal gland disease



Steroid 17- α -hydroxylase/17,20 lyase (CYP17A1) mediates both 17- α -hydroxylase and 17,20-lyase activity, allowing the adrenal glands and gonads to synthesise both 17- α -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- α -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).

Literature references

- Martin, RM., de Oliveira, ML., Costa, EM., Villar, H., Lin, CJ., Longui, CA. et al. (2003). P450c17 deficiency in Brazilian patients: biochemical diagnosis through progesterone levels confirmed by CYP17 genotyping. *J. Clin. Endocrinol. Metab.*, 88, 5739-46. [↗](#)
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2014-11-03	Reviewed	Nakaki, T.

Defective CYP17A1 does not cleave 17aHPROG ↗

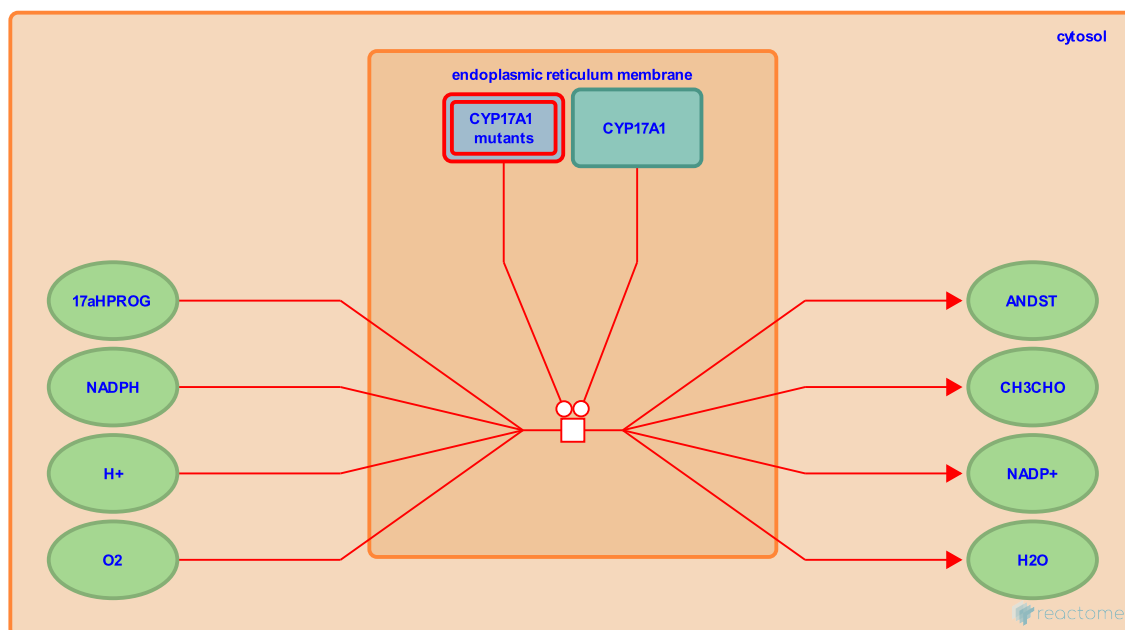
Location: Defective CYP17A1 causes AH5

Stable identifier: R-HSA-9035956

Type: transition

Compartments: endoplasmic reticulum membrane, cytosol

Diseases: adrenal gland disease



Steroid 17- α -hydroxylase/17,20 lyase (CYP17A1) mediates both 17- α -hydroxylase and 17,20-lyase activity, allowing the adrenal glands and gonads to synthesise both 17- α -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- α -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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