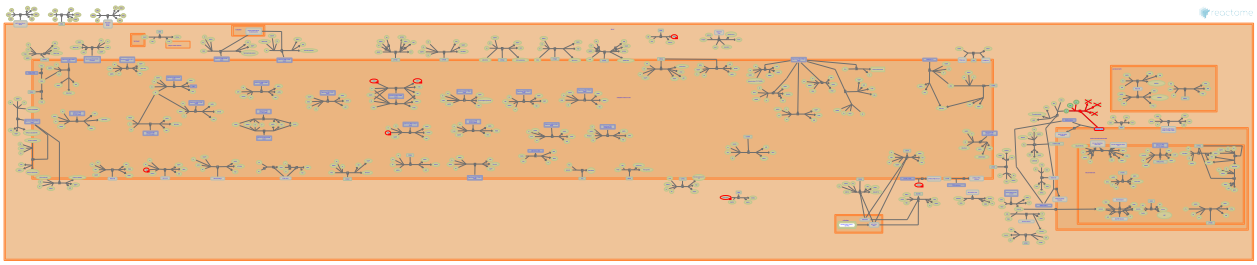
Defective MAOA causes BRUNS



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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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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

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

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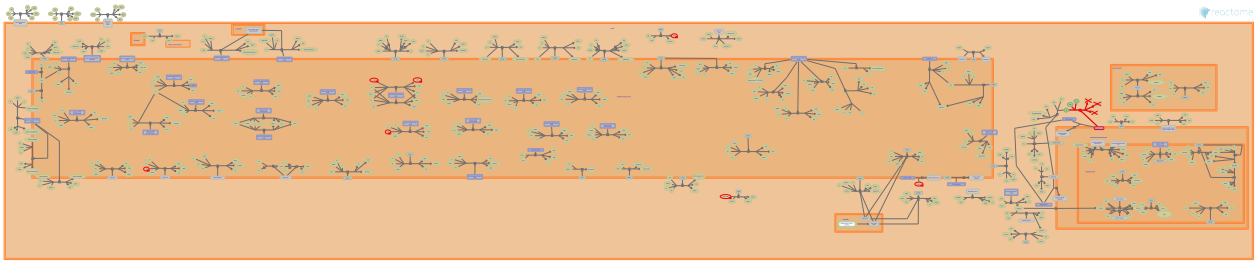
Defective MAOA causes BRUNS ↗

Stable identifier: R-HSA-5579012

Diseases: disease of mental health



Amine oxidase (flavin-containing) A (MAOA) catalyses the oxidative deamination of biogenic and dietary amines, the regulation of which is critical for mental state homeostasis. MAOA, located on the mitochondrial outer membrane and requiring FAD as cofactor (Weyler 1989), preferentially oxidises biogenic amines such as 5-hydroxytryptamine (5HT), dopamine, noradrenaline and adrenaline. Defects in MAOA can cause Brunner syndrome (BRUNS; MIM:300615), a form of X-linked non-dysmorphic mild mental retardation. Male patients are affected by mild mental retardation and exhibit abnormal behaviour, including impulsive aggression (Brunner et al. 1993, Shih et al. 1999, Shih 2004).

Literature references

Shih, JC. (2004). Cloning, after cloning, knock-out mice, and physiological functions of MAO A and B. *Neurotoxicology , 25*, 21-30. ↗

Shih, JC., Ridd, MJ., Chen, K. (1999). Role of MAO A and B in neurotransmitter metabolism and behavior. *Pol J Pharmacol, 51*, 25-9. ↗

Brunner, HG., Nelen, M., Ropers, HH., van Oost, BA., Breakefield, XO. (1993). Abnormal behavior associated with a point mutation in the structural gene for monoamine oxidase A. *Science, 262*, 578-80. ↗

Editions

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

Defective MAOA does not oxidatively deaminate 5HT ↗

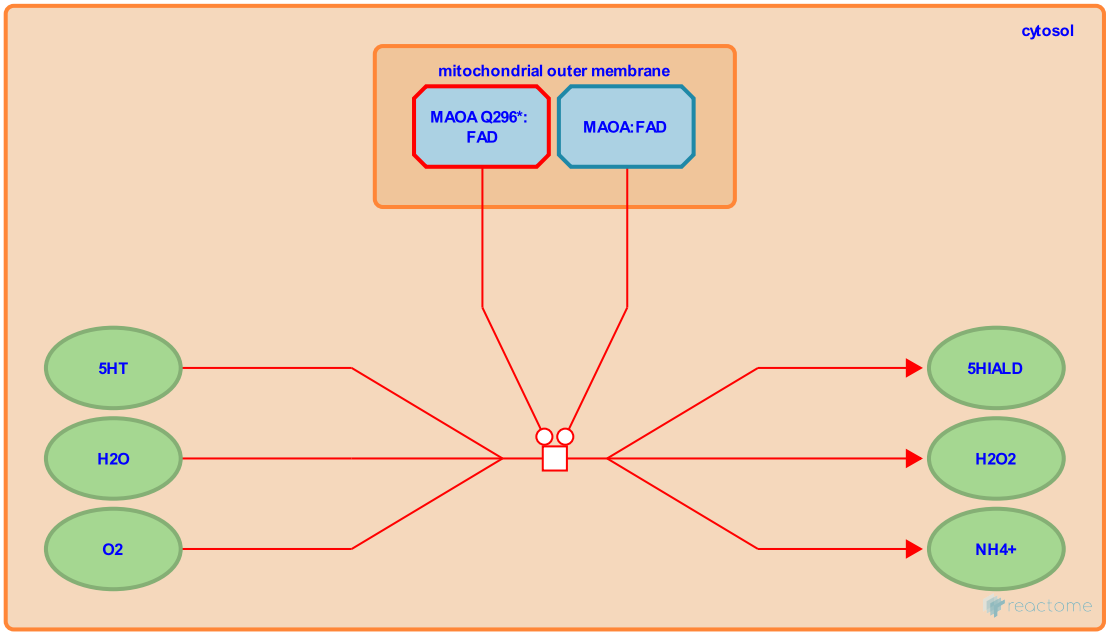
Location: Defective MAOA causes BRUNS

Stable identifier: R-HSA-5603108

Type: transition

Compartments: cytosol, mitochondrial outer membrane

Diseases: disease of mental health



Amine oxidase (flavin-containing) A (MAOA) catalyses the oxidative deamination of biogenic and dietary amines, the regulation of which is critical for mental state homeostasis. MAOA, located on the mitochondrial outer membrane and requiring FAD as cofactor, preferentially oxidises biogenic amines such as 5-hydroxytryptamine (5HT), dopamine, noradrenaline and adrenaline. Defects in MAOA can cause Brunner syndrome (BRUNS; MIM:300615), a form of X-linked non-dysmorphic mild mental retardation. Male patients are affected by mild mental retardation and exhibit abnormal behaviour, including impulsive aggression. A mutation that causes BRUNS is Q296* (Brunner et al. 1993).

Literature references

Brunner, HG., Nelen, M., Ropers, HH., van Oost, BA., Breakefield, XO. (1993). Abnormal behavior associated with a point mutation in the structural gene for monoamine oxidase A. *Science, 262*, 578-80. ↗

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