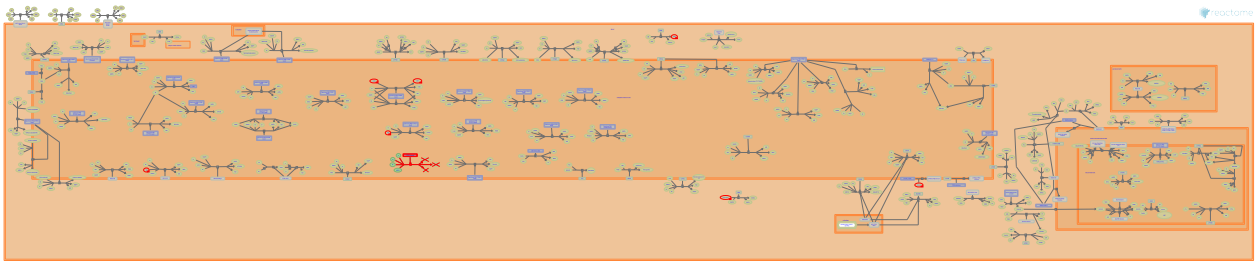
Defective CYP2U1 causes SPG56



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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. ↗

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

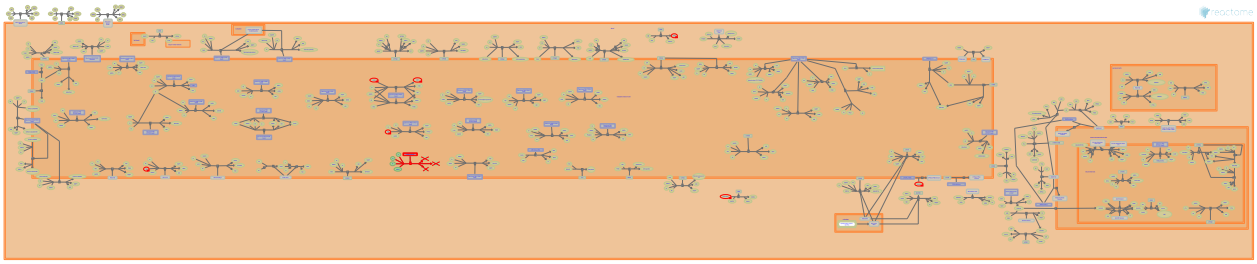
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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 CYP2U1 causes SPG56 ↗

Stable identifier: R-HSA-5579011

Diseases: hereditary spastic paraplegia



Cytochrome P450 2U1 (CYP2U1) catalyses the hydroxylation of arachidonic acid, docosahexaenoic acid and other long chain fatty acids, generating bioactive eicosanoid derivatives which may play an important physiological role in fatty acid signaling processes. Defects in CYP2U1 can cause Spastic paraplegia 56, autosomal recessive (SPG56; MIM:615030), a neurodegenerative disorder characterised by a slow, gradual, progressive weakness and spasticity of the lower limbs (Tesson et al. 2012, Fink 2013).

Literature references

Tesson, C., Yamashita, A., Gaussen, M., Nawara, M., Elmalik, SA., Zaki, MS. et al. (2012). Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *Am. J. Hum. Genet., 91*, 1051-64. ↗

Fink, JK. (2013). Hereditary spastic paraplegia: clinico-pathologic features and emerging molecular mechanisms. *Acta Neuropathol., 126*, 307-28. ↗

Editions

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

Defective CYP2U1 does not omega-hydroxylate ARA ↗

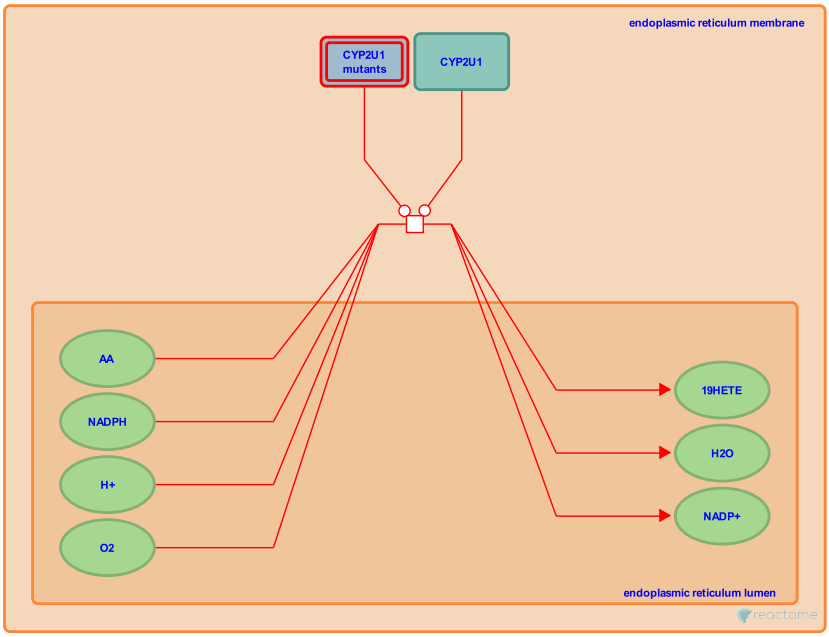
Location: Defective CYP2U1 causes SPG56

Stable identifier: R-HSA-5602242

Type: transition

Compartments: endoplasmic reticulum membrane, endoplasmic reticulum lumen

Diseases: hereditary spastic paraplegia



Cytochrome P450 2U1 (CYP2U1) catalyses the hydroxylation of arachidonic acid, docosahexaenoic acid and other long chain fatty acids, generating bioactive eicosanoid derivatives which may play an important physiological role in fatty acid signaling processes. Defects in CYP2U1 can cause Spastic paraplegia 56, autosomal recessive (SPG56; MIM:615030), a neurodegenerative disorder characterised by a slow, gradual, progressive weakness and spasticity of the lower limbs (Tesson et al. 2012, Fink 2013). CYP2U1 mutations that cause SPG56 are D316V, E380G, C262R, R488W and L21Wfs*19 (Tesson et al. 2012).

Literature references

Tesson, C., Yamashita, A., Gaussen, M., Nawara, M., Elmalik, SA., Zaki, MS. et al. (2012). Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *Am. J. Hum. Genet., 91,* 1051-64. ↗

Fink, JK. (2013). Hereditary spastic paraplegia: clinico-pathologic features and emerging molecular mechanisms. *Acta Neuropathol., 126,* 307-28. ↗

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