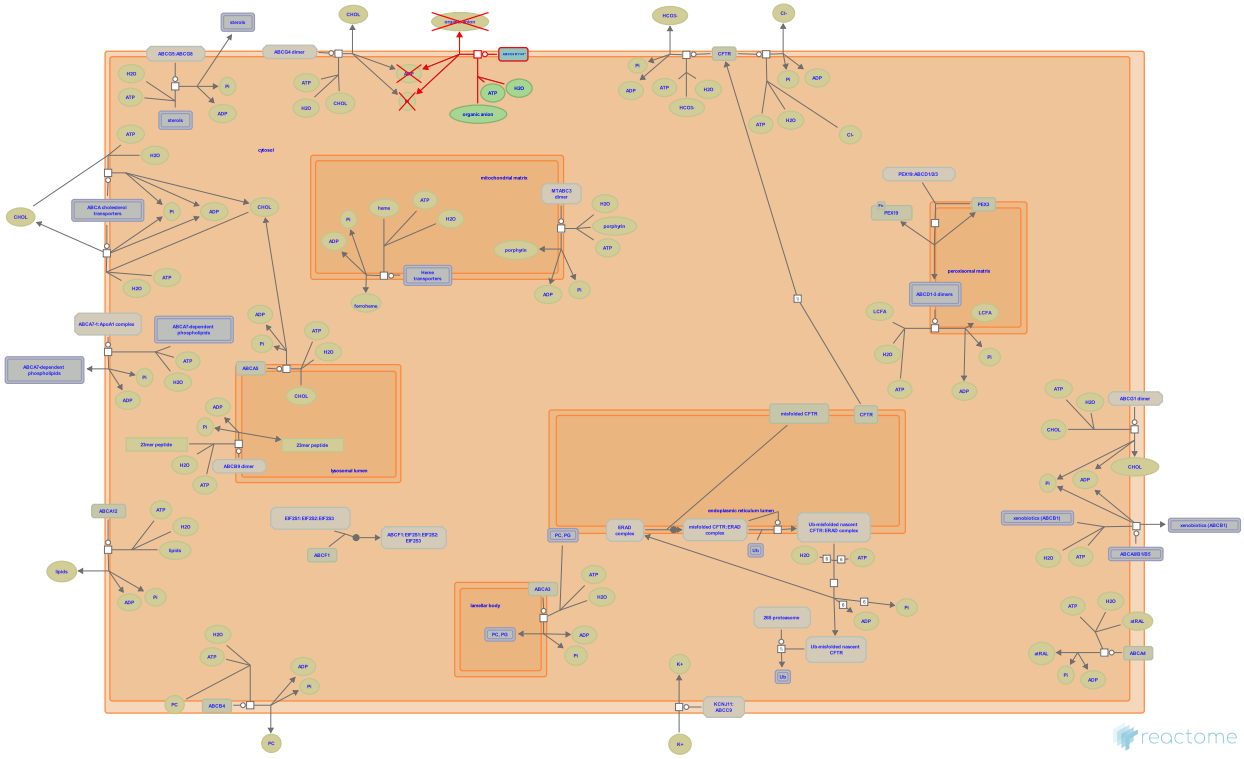
Defective ABCC6 causes PXE



Jassal, B., Shukla, S.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

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.

21/10/2024

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.
The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

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. ↗

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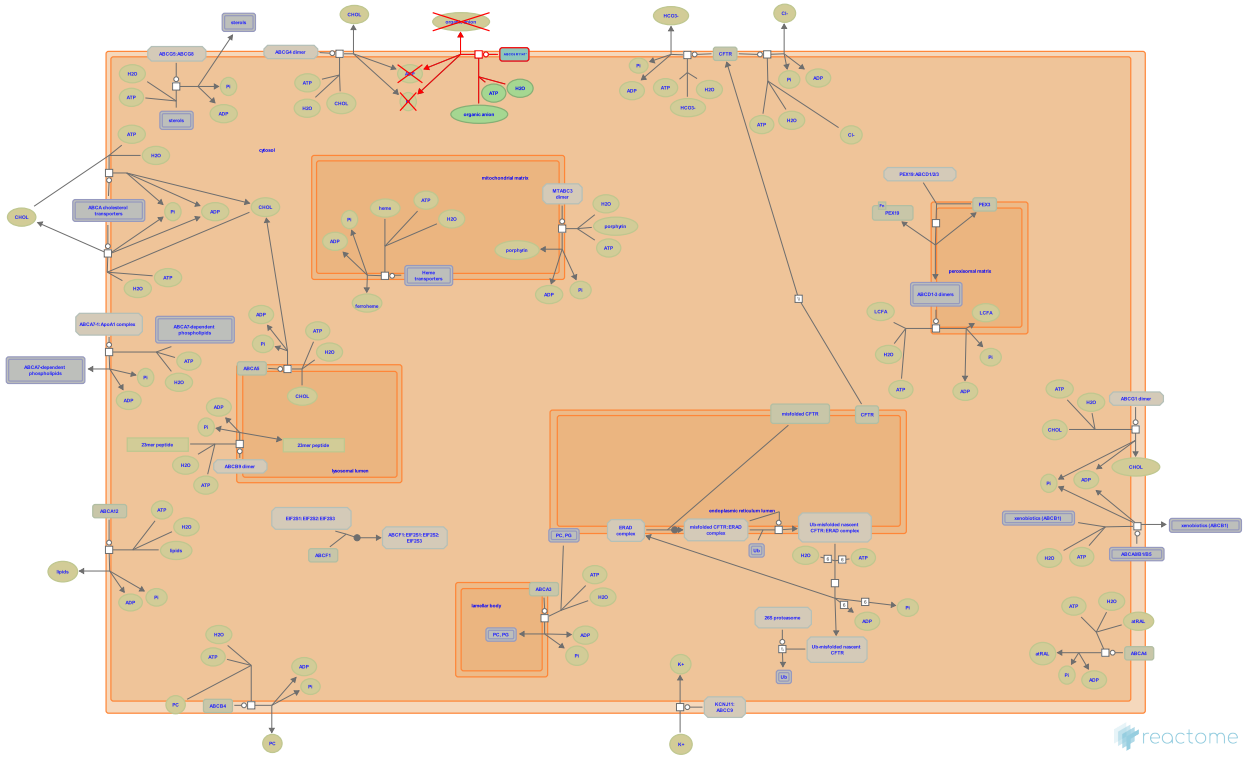
Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology, 14*, e1005968. ↗

Defective ABCC6 causes PXE ↗

Stable identifier: R-HSA-5690338

Diseases: pseudoxanthoma elasticum



The multidrug resistance associated protein (MRPs) subfamily of the ABC transporter family can transport a wide and diverse range of organic anions that can be endogenous compounds and xenobiotics and their metabolites. The multidrug resistance-associated protein 6 (ABCC6 aka MOAT-E) can actively transport organic anions. Defects in ABCC6 can cause pseudoxanthoma elasticum (PXE; MIM:264800), a rare multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vasculature and the Burch membrane of the eye (Finger et al. 2009).

Literature references

Szliska, C., Charbel Issa, P., Holz, FG., Scholl, HP., Ladewig, MS., Götting, C. et al. (2009). Pseudoxanthoma elasticum: genetics, clinical manifestations and therapeutic approaches. *Surv Ophthalmol, 54*, 272-85. ↗

Editions

2015-04-28	Authored, Edited	Jassal, B.
2015-09-15	Reviewed	Shukla, S.

Defective ABCC6 does not transport organic anion from cytosol to extracellular region ↗

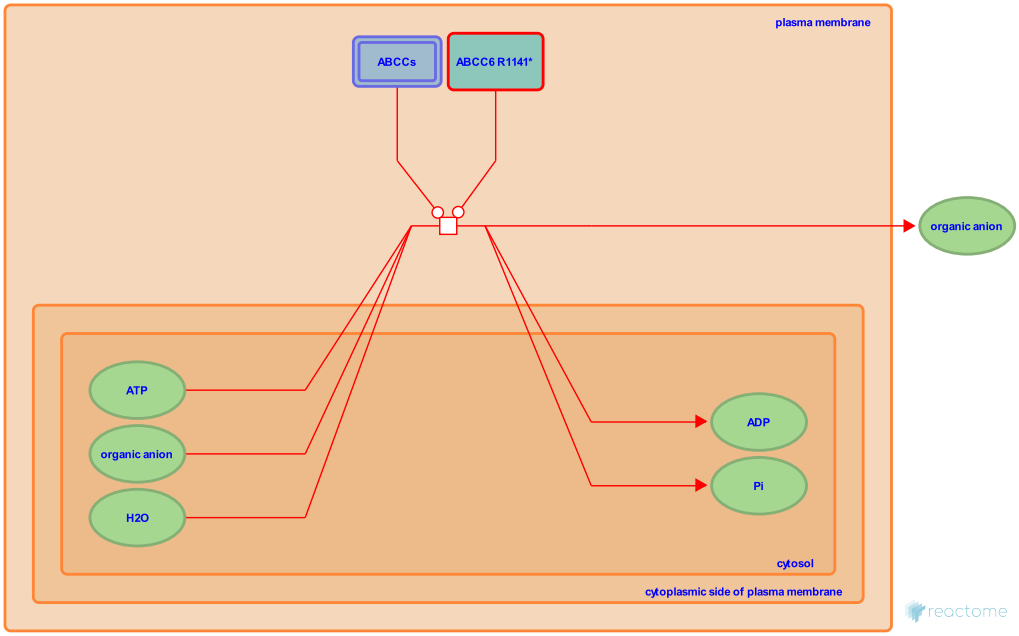
Location: Defective ABCC6 causes PXE

Stable identifier: R-HSA-5690340

Type: transition

Compartments: plasma membrane, cytosol

Diseases: pseudoxanthoma elasticum



The multidrug resistance associated protein (MRPs) subfamily of the ABC transporter family can transport a wide and diverse range of organic anions that can be endogenous compounds and xenobiotics and their metabolites. The multidrug resistance-associated protein 6 (ABCC6 aka MOAT-E) can actively transport organic anions. Defects in ABCC6 can cause pseudoxanthoma elasticum (PXE; MIM:264800), a rare multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vasculature and the Burch membrane of the eye. The most frequent mutation in ABCC6 causing PXE is R1141X (Le Saux et al. 2000, 2001, Hu et al. 2003).

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Scheffer, G., Peek, R., Leys, A., Plomp, A., ten Brink, Jt., de Jong, PT. et al. (2003). Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum. *Invest. Ophthalmol. Vis. Sci., 44*, 1824-9. ↗

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Silvestri, C., Le Saux, O., Pope, FM., Beck, K., De Paepe, A., Johnson, EW. et al. (2001). A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum. *Am. J. Hum. Genet., 69*, 749-64. ↗

Editions

2015-04-28	Authored, Edited	Jassal, B.
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