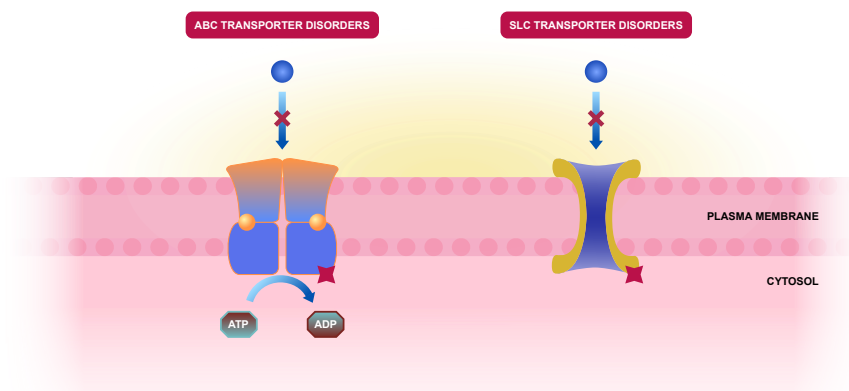


Disorders of transmembrane transporters



Broer, S., Jassal, B., Moitra, K., Shukla, S.

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

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of [Creative Commons Attribution 4.0 International \(CC BY 4.0\) License](https://creativecommons.org/licenses/by/4.0/). For more information see our [license](https://reactome.org/licenses/).

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/textbook/).

24/04/2024

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

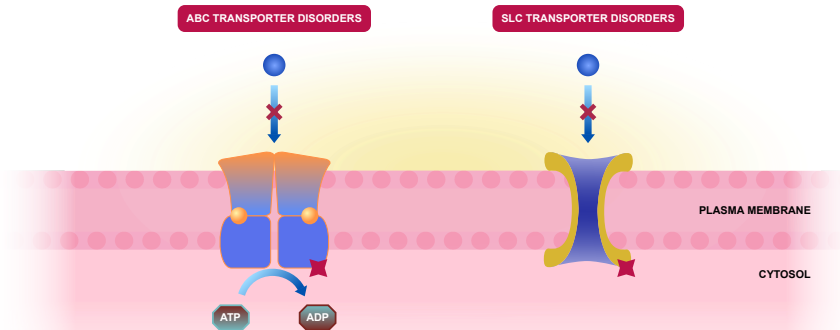
- Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics*, 18, 142. [↗](#)
- Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics*, 33, 3461-3467. [↗](#)
- 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. [↗](#)

Reactome database release: 88

This document contains 3 pathways ([see Table of Contents](#))

Disorders of transmembrane transporters ↗

Stable identifier: R-HSA-5619115



reactome
DISEASE

Proteins with transporting functions can be roughly classified into 3 categories: ATP hydrolysis-coupled pumps, ion channels, and transporters. Pumps utilize the energy released by ATP hydrolysis to power the movement of substrates across the membrane against their electrochemical gradient. Channels in their open state can transfer substrates (ions or water) down their electrochemical gradient at an extremely high efficiency (up to 10^8 s⁻¹). Transporters facilitate the movement of a specific substrate either against or with their concentration gradient at a lower speed (about 10^2 - 10^4 s⁻¹); as generally believed, conformational change of the transporter protein is involved in the transfer process. Diseases caused by defects in these transporter proteins are detailed in this section. Disorders associated with ABC transporters and SLC transporters are annotated here (Dean 2005).

Literature references

Dean, M. (2005). The genetics of ATP-binding cassette transporters. *Meth. Enzymol.*, 400, 409-29. ↗

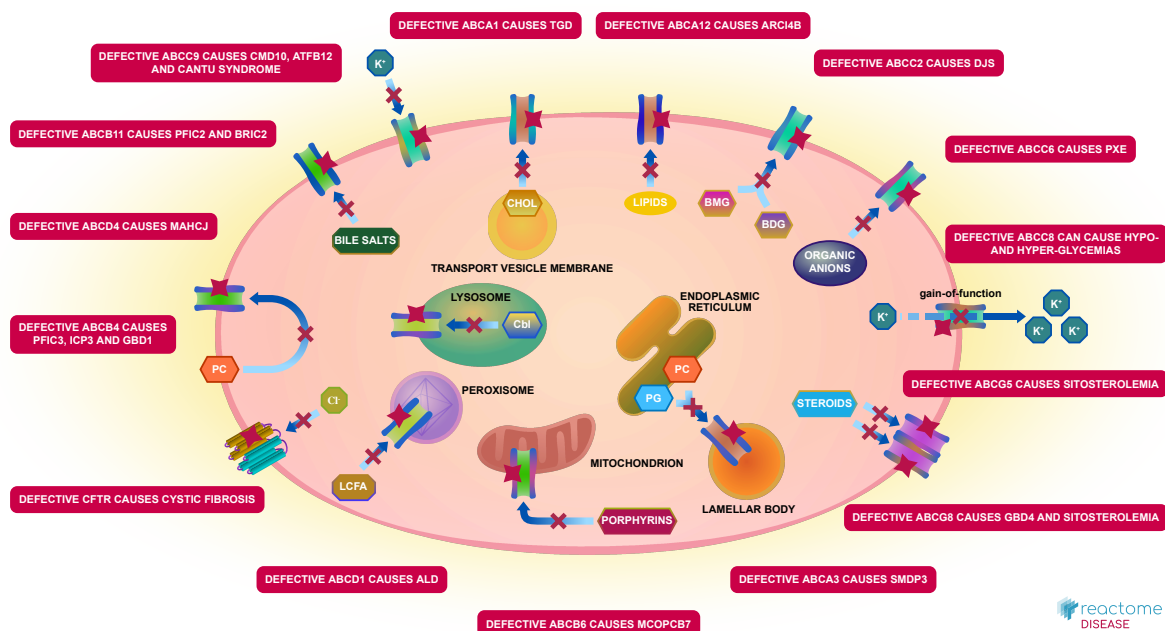
Editions

2014-08-22	Authored, Edited	Jassal, B.
2015-04-28	Reviewed	Moitra, K.

ABC transporter disorders ↗

Location: Disorders of transmembrane transporters

Stable identifier: R-HSA-5619084



The ATP-binding cassette (ABC) transporters form a large family of transmembrane proteins that utilise the energy from the hydrolysis of ATP to facilitate the movement of a wide variety of substrates against a concentration gradient across membrane bilayers. Substrates include amino acids, lipids, inorganic ions, peptides, saccharides, peptides for antigen presentation, metals, drugs, and proteins. Of the 48 known ABC transporters in humans, 15 are associated with a defined human disease (Tarling et al. 2013, Woodward et al. 2011, Dean 2005, Kemp et al. 2011, Ueda 2011, Chen & Tiwari 2011).

Literature references

- Chen, ZS., Tiwari, AK. (2011). Multidrug resistance proteins (MRPs/ABCCs) in cancer chemotherapy and genetic diseases. *FEBS J.*, 278, 3226-45. ↗
- Edwards, PA., de Aguiar Vallim, TQ., Tarling, EJ. (2013). Role of ABC transporters in lipid transport and human disease. *Trends Endocrinol. Metab.*, 24, 342-50. ↗
- Köttgen, M., Köttgen, A., Woodward, OM. (2011). ABCG transporters and disease. *FEBS J.*, 278, 3215-25. ↗
- Dean, M. (2005). The genetics of ATP-binding cassette transporters. *Meth. Enzymol.*, 400, 409-29. ↗
- Kemp, S., Wanders, RJA., Theodoulou, FL. (2011). Mammalian peroxisomal ABC transporters: from endogenous substrates to pathology and clinical significance. *Br. J. Pharmacol.*, 164, 1753-66. ↗

Editions

2014-08-22	Authored, Edited	Jassal, B.
2015-04-28	Reviewed	Moitra, K.
2015-09-15	Reviewed	Shukla, S.

SLC transporter disorders ↗

Location: Disorders of transmembrane transporters

Stable identifier: R-HSA-5619102



The solute-carrier gene (SLC) superfamily encodes membrane-bound transporters comprising 55 gene families with at least 362 putatively functional protein-coding genes. The gene products include passive transporters, symporters and antiporters and are located in all cellular and organelle membranes. Curated here is a list of SLCs, where mutations within them can result in disease (Hediger et al. 2013).

Literature references

Hediger, MA., Cl  men  on, B., Bruford, EA., Burrier, RE. (2013). The ABCs of membrane transporters in health and disease (SLC series): introduction. *Mol. Aspects Med.*, 34, 95-107. ↗

Editions

2014-08-22	Authored, Edited	Jassal, B.
2015-08-04	Reviewed	Broer, S.

Table of Contents

- Introduction 1
- ❖ Disorders of transmembrane transporters 2
 - ❖ ABC transporter disorders 3
 - ❖ SLC transporter disorders 4
- Table of Contents 5