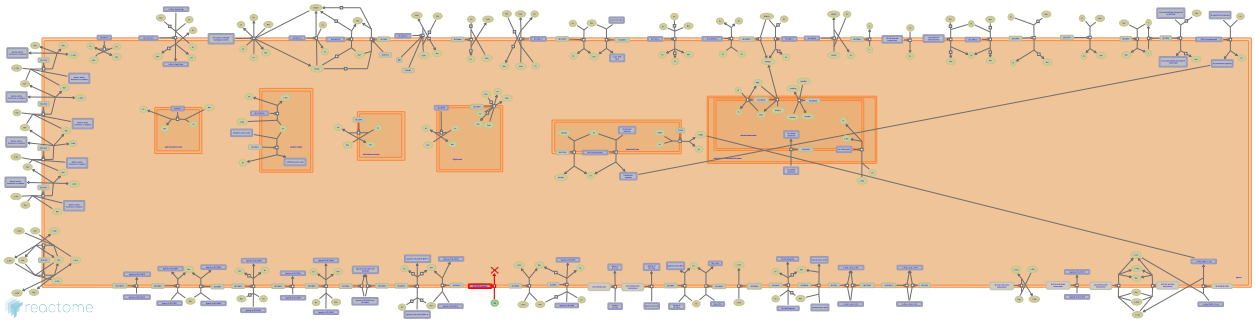
Defective SLC6A18 may confer susceptibility to iminoglycinuria and/or hyperglycinuria



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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. ↗

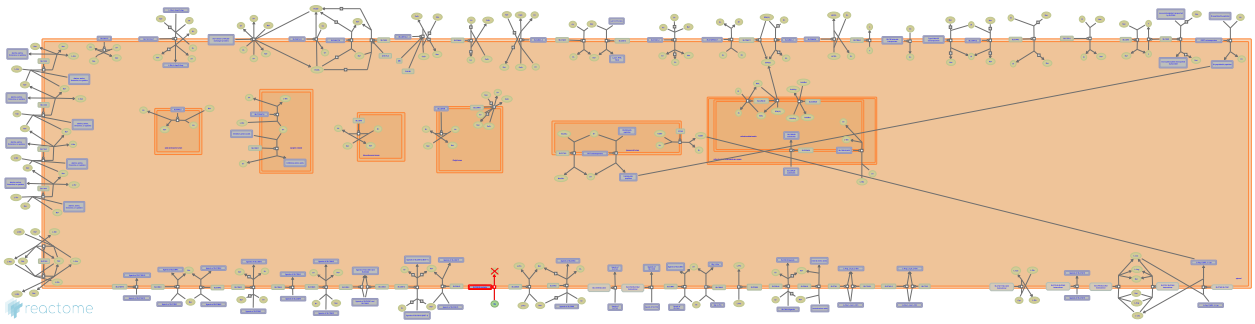
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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 SLC6A18 may confer susceptibility to iminoglycinuria and/or hyperglycinuria ↗

Stable identifier: R-HSA-5659729

Diseases: amino acid metabolic disorder



SLC6A18 encodes a neutral amino acid transporter B(0)AT3 which has preference for the amino acid glycine. It is abundantly expressed in the kidney, specifically the S2/3 segments of the kidney proximal tubule (Broer & Gether 2012, Schweikhard & Ziegler 2012). Iminoglycinuria (IG; MIM:242600) or hyperglycinuria (HG; MIM:138500) can arise from defects in SLC36A2, encoding a proton-coupled amino acid transporter 2 (PAT2), a high-affinity cotransporter of glycine and proline. Mutation in SLC6A18 may contribute to both IG and HG (Broer et al. 2008).

Literature references

Schweikhard, ES., Ziegler, CM. (2012). Amino acid secondary transporters: toward a common transport mechanism. *Curr Top Membr, 70*, 1-28. ↗

Broer, S., Bailey, CG., Kowalczuk, S., Ng, C., Vanslambrouck, JM., Rodgers, H. et al. (2008). Iminoglycinuria and hyperglycinuria are discrete human phenotypes resulting from complex mutations in proline and glycine transporters. *J. Clin. Invest., 118*, 3881-92. ↗

Editions

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

Defective SLC6A18 does not transport Gly from extracellular region to cytosol ↗

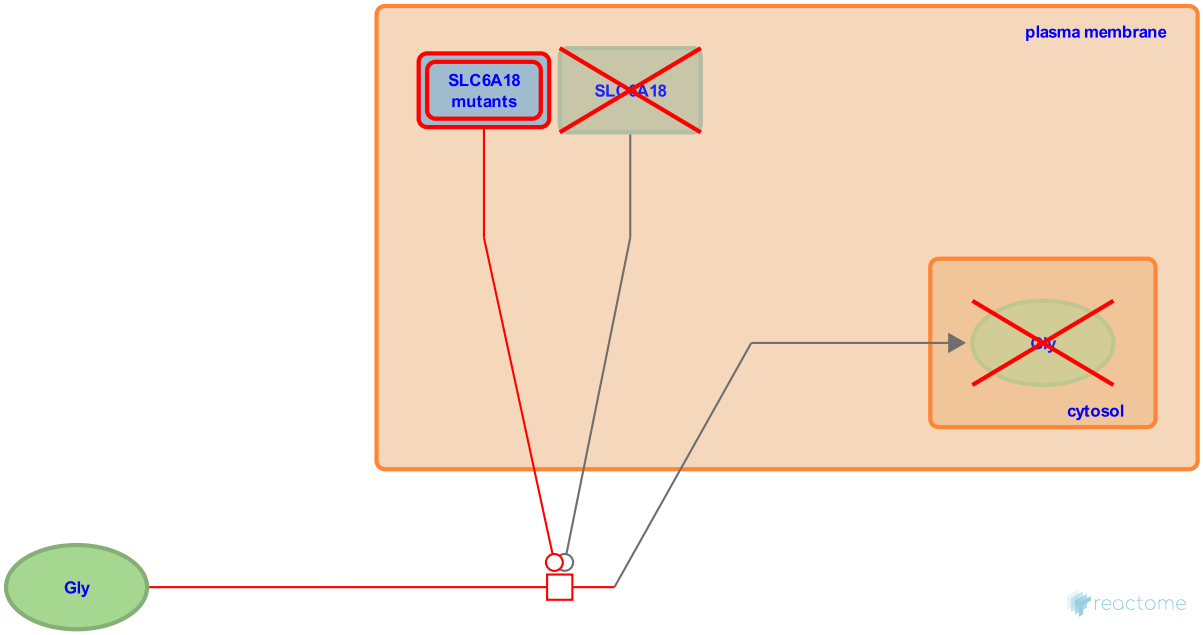
Location: Defective SLC6A18 may confer susceptibility to iminoglycinuria and/or hyperglycinuria

Stable identifier: R-HSA-5659755

Type: transition

Compartments: extracellular region, plasma membrane

Diseases: amino acid metabolic disorder



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Literature references

Broer, S., Bailey, CG., Kowalczuk, S., Ng, C., Vanslambrouck, JM., Rodgers, H. et al. (2008). Iminoglycinuria and hyperglycinuria are discrete human phenotypes resulting from complex mutations in proline and glycine transporters. *J. Clin. Invest., 118*, 3881-92. ↗

Editions

2014-12-23	Authored, Edited	Jassal, B.
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