Defective SI does not hydrolyze iMal

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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

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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 SI does not hydrolyze iMal ↗

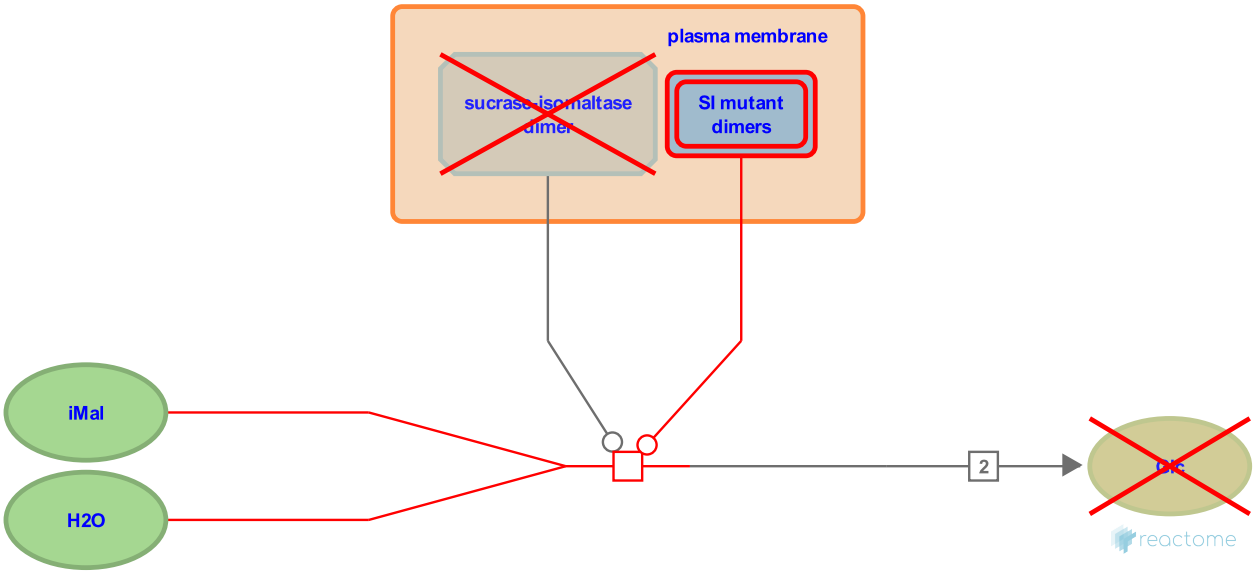
Stable identifier: R-HSA-5659879

Type: transition

Compartments: extracellular region, plasma membrane

Diseases: intestinal disaccharidase deficiency

Inferred from: Defective SI does not hydrolyze Mal (Homo sapiens)



Mutations that disrupt the catalytic activity or strongly interfere with proper folding, glycosylation and transport of SI (sucrase-isomaltase) are inferred to block the cleavage of isomaltose (iMal) to glucose, based on the experimentally demonstrated failure of these SI mutant proteins to hydrolyze maltose (e.g., Sander et al. 2006) and the broad substrate specificity of the normal enzyme (Sim et al. 2010).

Literature references

Sim, L., Willemsma, C., Mohan, S., Naim, HY., Pinto, BM., Rose, DR. (2010). Structural basis for substrate selectivity in human maltase-glucoamylase and sucrase-isomaltase N-terminal domains. *J. Biol. Chem., 285*, 17763-70. ↗

Sander, P., Alfalah, M., Keiser, M., Korponay-Szabo, I., Kovács, JB., Leeb, T. et al. (2006). Novel mutations in the human sucrase-isomaltase gene (SI) that cause congenital carbohydrate malabsorption. *Hum. Mutat., 27*, 119. ↗

Editions

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