Defective GALT can cause GALCT

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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 GALT can cause GALCT ↗

Stable identifier: R-HSA-5609978

Diseases: galactosemia

Galactose-1-phosphate uridylyltransferase (GALT) is one of the enzymes involved in galactose metabolism in the Leloir pathway. GALT catalyses the transfer of uridine monophosphate (UMP) from UDP-glucose (UDP-Glc) to galactose-1-phosphate (Gal1P) to form UDP-galactose (UDP-Gal) and glucose 1-phosphate. Defects in GALT can cause Galactosemia (GALCT; MIM:230400), an autosomal recessive disorder of galactose metabolism presenting in neonatals that causes jaundice, cataracts and mental retardation (Bosch 2006).

Literature references

Bosch, AM. (2006). Classical galactosaemia revisited. *J. Inherit. Metab. Dis., 29*, 516-25. ↗

Editions

2014-07-18	Authored, Edited	Jassal, B.
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Defective GALT does not transfer UMP to Gal1P ↗

Location: Defective GALT can cause GALCT

Stable identifier: R-HSA-5610038

Type: transition

Compartments: cytosol

Diseases: galactosemia