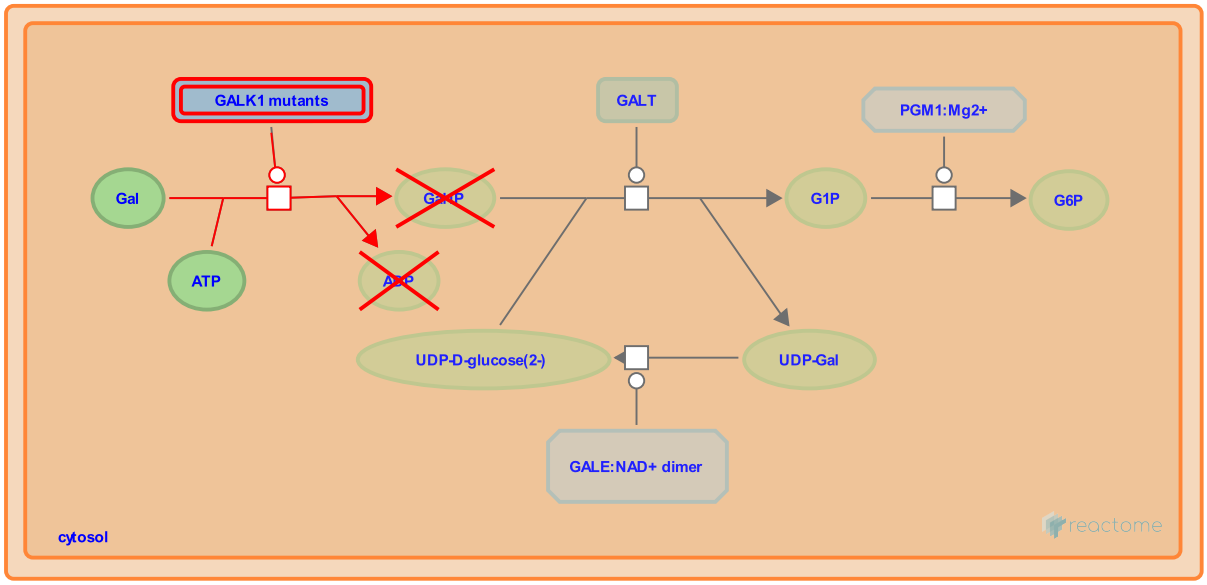
Defective GALK1 causes GALCT2



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04/04/2024

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).

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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. ↗

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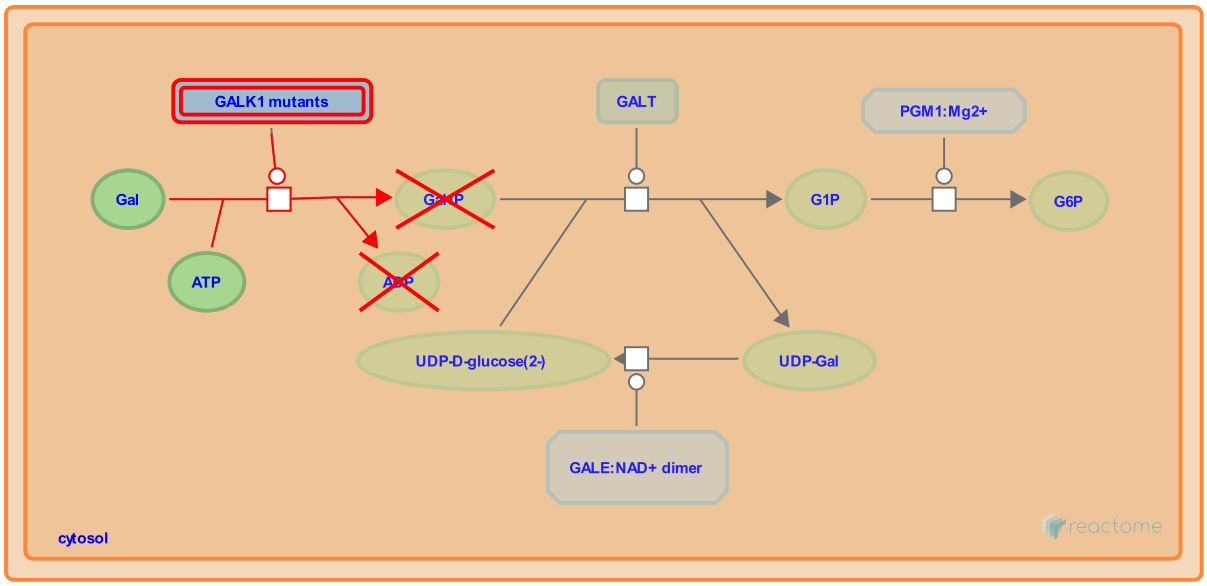
Defective GALK1 causes GALCT2

Stable identifier: R-HSA-5609976

Diseases: galactokinase deficiency



Cytosolic galactokinase (GALK1) catalyses the first committed step in the Leloir pathway of galactose metabolism. GALK1 catalyses the phosphorylation of D-galactose (Gal) to form D-galactose 1-phosphate (Gal1P). Defects in GALK1 can cause type II galactosemia (GALCT2; MIM:230200), an autosomal recessive deficiency characterised by congenital cataracts during infancy and presenile cataracts in the adult population. Galactitol accumulation in the lens is the cause of these cataracts (Bosch et al. 2002).

Literature references

Wijburg, FA., Bakker, HD., van Gennip, AH., Wanders, RJA., Bosch, AM., van Kempen, JV. (2002). Clinical features of galactokinase deficiency: a review of the literature. *J. Inherit. Metab. Dis., 25*, 629-34.

Editions

2014-07-18	Authored, Edited	Jassal, B.
2015-02-25	Reviewed	Timson, DJ.

Defective GALK1 does not phosphorylate Gal ↗

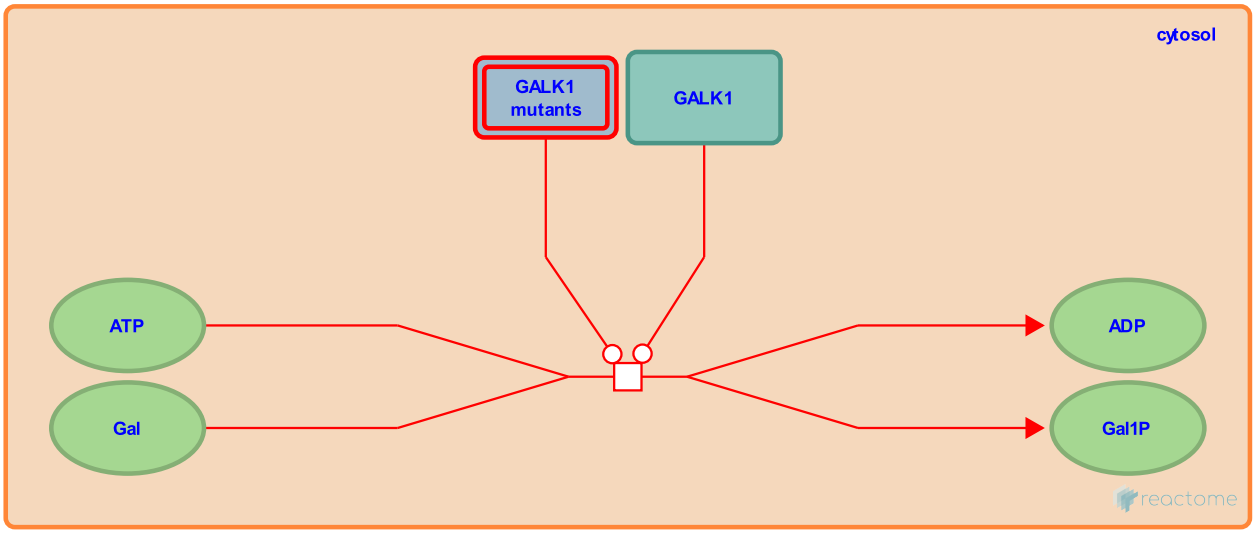
Location: Defective GALK1 causes GALCT2

Stable identifier: R-HSA-5610026

Type: transition

Compartments: cytosol

Diseases: galactokinase deficiency



Cytosolic galactokinase (GALK1) catalyses the first step in the Leloir pathway of galactose metabolism. GALK1 catalyses the phosphorylation of D-galactose (Gal) to form D-galactose 1-phosphate (Gal1P). Defects in GALK1 can cause Galactosemia II (GALCT2; MIM:230200), an autosomal recessive deficiency characterised by congenital cataracts during infancy and presenile cataracts in the adult population. Galactitol accumulation in the lens is the cause of these cataracts. Mutations causing GALCT2 include V32M, E80*, P28T, Q382* and R256W (Stambolian et al. 1995, Kalaydjieva et al. 1999, Kolosha et al 2000, Bayarchimeg et al. 2012). There is a wide spectrum of severity and age of onset. However, beyond cataract formation, no long-term complications have been documented (Bosch et al. 2002, Timson & Reece 2003).

Literature references

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Flanagan, SE., Ismail, D., Hussain, K., Burk, D., Hogler, W., Bayarchimeg, M. et al. (2012). Galactokinase deficiency in a patient with congenital hyperinsulinism. *JIMD Rep, 5*, 7-11. ↗

Editions

2014-07-21	Authored, Edited	Jassal, B.
2015-02-25	Reviewed	Timson, DJ.

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