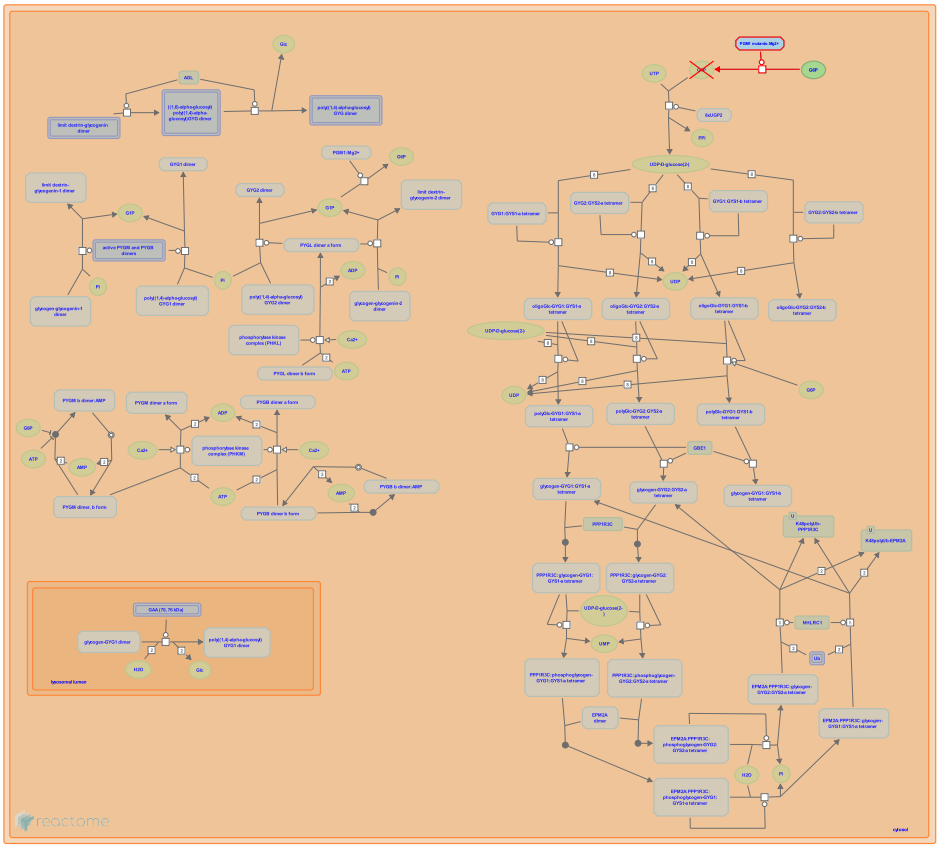
Defective PGM1 causes PGM1-CDG



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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.

19/09/2024

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. ↗

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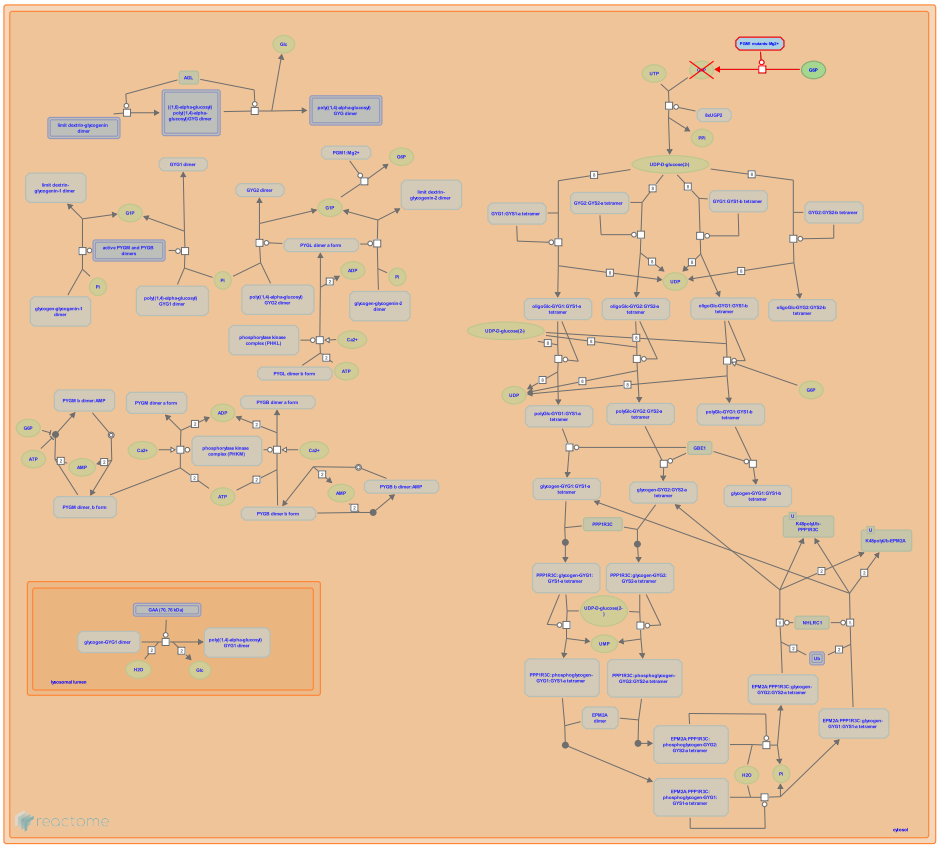
Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology, 14*, e1005968. ↗

Defective PGM1 causes PGM1-CDG ↗

Stable identifier: R-HSA-5609974

Diseases: congenital disorder of glycosylation



Phosphoglucomutases 1 and 2 (PGM1, 2) are involved in the cytosolic biosynthesis of nucleotide sugars needed for glycan biosynthesis, specifically, the isomerisation of glucose-6-phosphate (G6P) into glucose-1-phosphate (G1P). Defects in PGM1 can cause congenital disorder of glycosylation 1t (CDG1t, now known as PGM1-CDG; MIM:614921), a broad spectrum disorder characterised by under-glycosylated serum glycoproteins (Timal et al. 2012, Tegtmeyer et al. 2014). CDGs result in a wide variety of clinical features such as defects in nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency.

Literature references

Paprocka, J., Thiel, C., Morava, E., Rodenburg, RJ., Hoischen, A., Timal, S. et al. (2012). Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. *Hum. Mol. Genet., 21*, 4151-61. ↗

Tegtmeyer, LC., Debus, V., Seyyedi, S., Rymen, D., Ficicioglu, C., He, P. et al. (2014). Multiple phenotypes in phosphoglucomutase 1 deficiency. *N. Engl. J. Med., 370*, 533-42. ↗

Editions

2014-07-18	Authored, Edited	Jassal, B.
2015-04-30	Reviewed	Spillmann, D.

Defective PGM1 does not isomerise G6P to G1P ↗

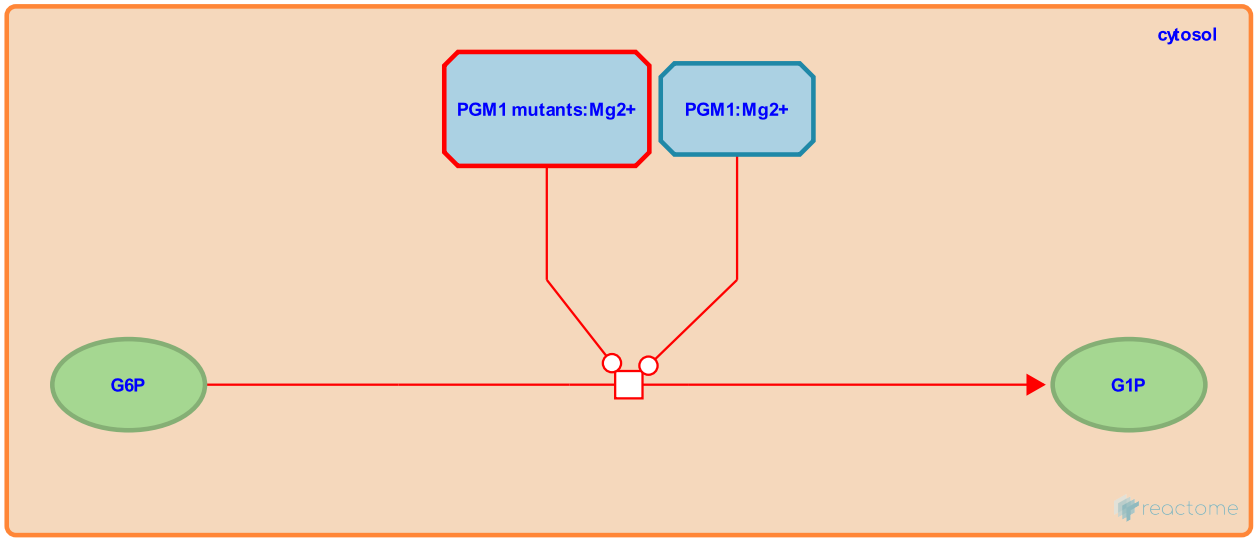
Location: Defective PGM1 causes PGM1-CDG

Stable identifier: R-HSA-5609939

Type: transition

Compartments: cytosol

Diseases: congenital disorder of glycosylation



Cytosolic phosphoglucomutase (PGM) catalyses the reversible conversion of glucose 6-phosphate (G6P) to glucose 1-phosphate (G1P), both precursor intermediates in glucose metabolism and protein glycosylation processes. Defects in PGM1 can cause Congenital disorder of glycosylation 1t (CDG1t, now known as PGM1-CDG; MIM:614921), a broad spectrum disorder characterised by under-glycosylated serum glycoproteins (Timal et al. 2012, Tegtmeyer et al. 2014). CDGs result in a wide variety of clinical features such as defects in nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. Mutations that cause almost complete loss of PGM1 activity include T115A, N38Y, D62H, D263Y and R221Vfs*13 (Tegtmeyer et al. 2014).

Literature references

Paprocka, J., Thiel, C., Morava, E., Rodenburg, RJ., Hoischen, A., Timal, S. et al. (2012). Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. *Hum. Mol. Genet., 21*, 4151-61. ↗

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Editions

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