Defective TALDO1 does not transform SH7P, GA3P to Fru(6)P, E4P

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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. ↗

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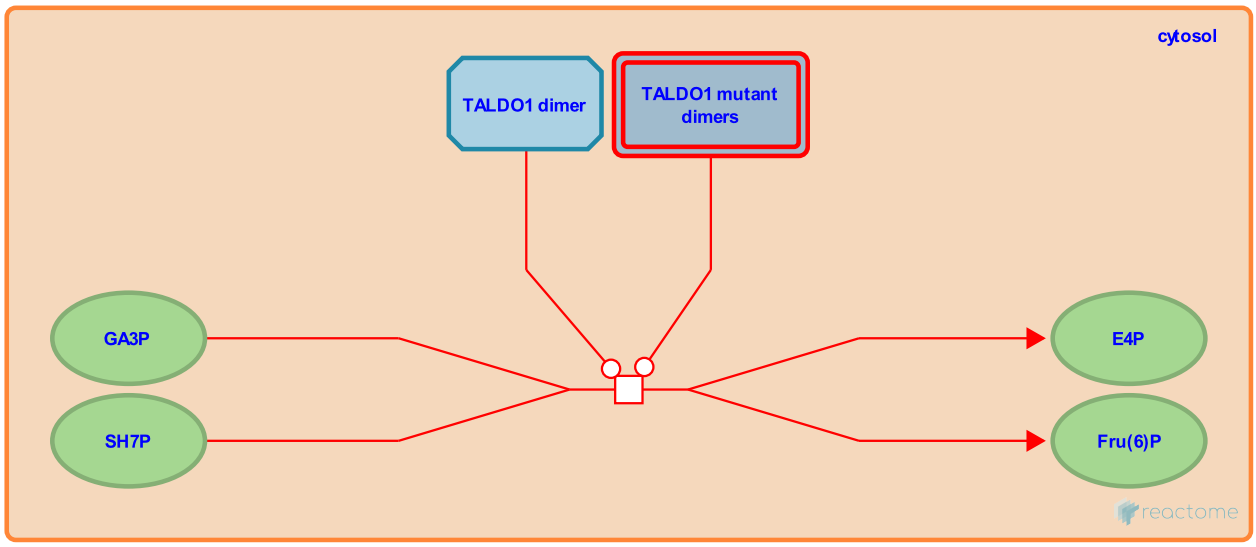
Stable identifier: R-HSA-5659989

Type: transition

Compartments: cytosol

Diseases: carbohydrate metabolic disorder



Defective TALDO1 (transaldolase 1) fails to transform sedoheptulose 7-phosphate (SH7P) and glyceraldehyde 3-phosphate (GA3P) to fructose 6-phosphate (Fru(6)P) and erythrose 4-phosphate (E4P). This defect has been associated with congenital liver disease and an array of other symptoms. The deficiency was first described by Verhoeven and colleagues (2001). Both the range and severity of these abnormalities are variable from patient to patient (Wamelink et al. 2008a; Eyaid et al. 2013). The three missense mutant alleles annotated here are associated with absence of detectable transaldolase activity in tissues from homozygous affected individuals (LeDuc et al. 2014; Verhoeven et al. 2005; Wamelink et al. 2008b).

Literature references

Verhoeven, NM., van der Knaap, MS., Salomons, GS., Struys, EA., Douwes, AC., Jakobs, C. et al. (2001). Transaldolase deficiency: liver cirrhosis associated with a new inborn error in the pentose phosphate pathway. *Am J Hum Genet, 68*, 1086-92.

Wilson, A., Wamelink, MM., Chung, WK., Shen, Y., Salomons, GS., Crouch, EE. et al. (2014). Novel association of early onset hepatocellular carcinoma with transaldolase deficiency. *JIMD Rep, 12*, 121-7.

Wamelink, MM., Struys, EA., Jakobs, C. (2008). The biochemistry, metabolism and inherited defects of the pentose phosphate pathway: a review. *J Inherit Metab Dis, 31*, 703-17.

Wamelink, MM., Fowler, D., Salomons, GS., Clayton, PT., Struys, EA., Jakobs, C. (2008). Transaldolase deficiency in a two-year-old boy with cirrhosis. *Mol. Genet. Metab., 94*, 255-8.

Al Balwi, M., Eyaid, W., Wamelink, MM., Alfadhel, M., Anazi, S., Al Harbi, T. et al. (2013). Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. *J. Inherit. Metab. Dis., 36*, 997-1004.

Editions

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