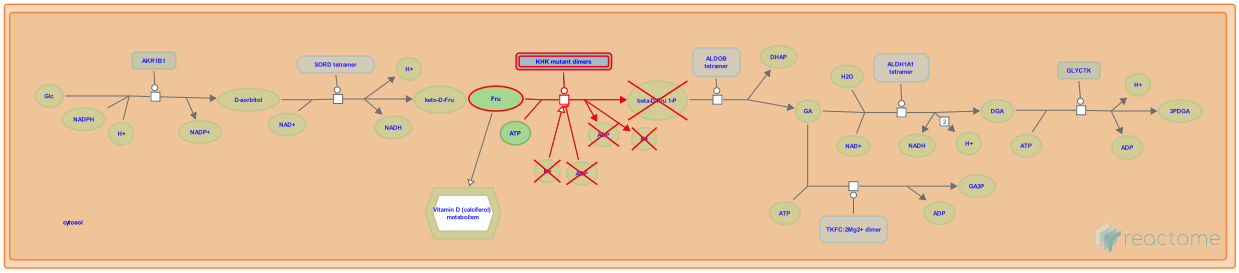
Essential fructosuria



D'Eustachio, P., Jassal, B., Timson, DJ., Tolan, DR.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

10/04/2024

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).

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. ↗

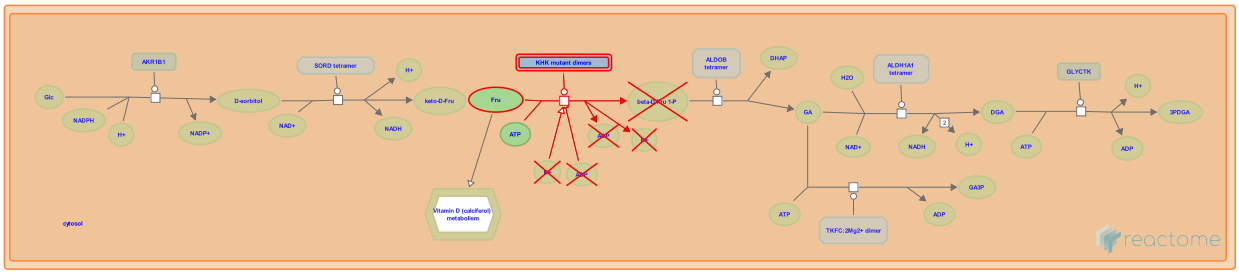
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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. ↗

Essential fructosuria ↗

Stable identifier: R-HSA-5657562

Diseases: carbohydrate metabolic disorder



Deficiencies in KHK (ketohexokinase) are associated with essential fructosuria (Bonthron et al. 1994).

Literature references

Donaldson, IA., Brady, N., Steinmann, B., Bonthron, DT. (1994). Molecular basis of essential fructosuria: molecular cloning and mutational analysis of human ketohexokinase (fructokinase). *Hum Mol Genet, 3*, 1627-31. ↗

Editions

2015-01-29	Authored, Edited	D'Eustachio, P.
2015-01-29	Reviewed	Jassal, B.
2015-02-17	Reviewed	Tolan, DR., Timson, DJ.

Defective KHK does not phosphorylate beta-D-fructose ↗

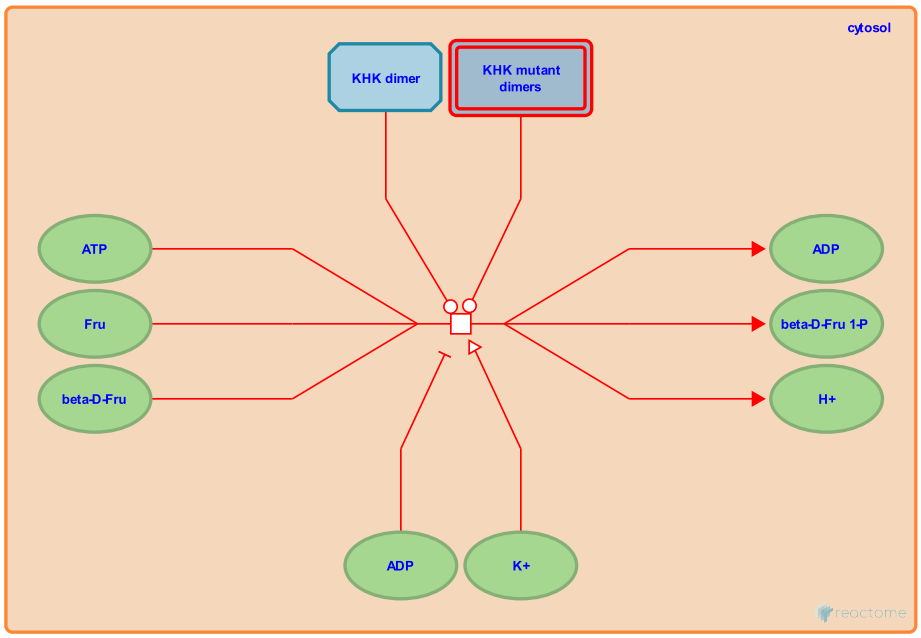
Location: Essential fructosuria

Stable identifier: R-HSA-5656459

Type: transition

Compartments: cytosol

Diseases: carbohydrate metabolic disorder



Variant KHK (ketohexokinase) protein fails to catalyze the phosphorylation of fructose to yield fructose 1-phosphate (Fru 1-P), the first step of fructose catabolism in the liver. This defect is associated with essential fructosuria, a rare benign condition characterized by elevated urinary fructose levels associated with consumption of fructose. Two missense mutant alleles have been identified in DNA sequencing studies of affected individuals (Bouthron et al. 1994). One, G40R, has no detectable activity. The second, A43T, encodes a protein whose liver ("A") isoform is inactive but whose peripheral ("C") isoform, though thermally unstable, retains some activity (Asipu et al. 2003).

Literature references

Hayward, BE., O'Reilly, J., Asipu, A., Bonthron, DT. (2003). Properties of normal and mutant recombinant human ketohexokinases and implications for the pathogenesis of essential fructosuria. *Diabetes, 52*, 2426-32. ↗

Donaldson, IA., Brady, N., Steinmann, B., Bonthron, DT. (1994). Molecular basis of essential fructosuria: molecular cloning and mutational analysis of human ketohexokinase (fructokinase). *Hum Mol Genet, 3*, 1627-31. ↗

Editions

2015-01-29	Authored, Edited	D'Eustachio, P.
2015-01-29	Reviewed	Jassal, B.
2015-02-17	Reviewed	Tolan, DR., Timson, DJ.

Table of Contents