Essential fructosuria

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


Reactome database release: 82

This document contains 1 pathway and 1 reaction (see Table of Contents)
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


Editions

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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**


**Editions**

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