Phenylketonuria

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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.

27/10/2022
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)

https://reactome.org
Phenylketonuria

Stable identifier: R-HSA-2160456

Diseases: phenylketonuria

Phenylalanine hydroxylase (PAH) normally catalyzes the conversion of phenylalanine to tyrosine. In the absence of functional PAH, phenylalanine accumulates to high levels in the blood and is converted to phenylpyruvate and phenyllactate (Clemens et al. 1990; Langenbeck et al. 1992; Mitchell et al. 2011). The extent of these conversions is modulated by genetic factors distinct from PAH, as siblings with the identical PAH defect can produce different amounts of them (Treacy et al. 1996).

Both L-amino acid oxidase (Boulland et al. 2004) and Kynurenine–oxoglutarate transaminase 3 (Han et al. 2004) can catalyze the conversion of phenylalanine to phenylpyruvate and lactate dehydrogenase can catalyze the conversion of the latter molecule to phenyllactate (Meister 1950), in reactions not annotated here.

Literature references


Editions

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Defective PAH does not hydroxylate L-Phe to L-Tyr

Location: Phenylketonuria

Stable identifier: R-HSA-5649483

Type: transition

Compartments: cytosol

Diseases: phenylketonuria