Defective GCK causes maturity-onset diabetes of the young 2 (MODY2)

Broer, S., Jassal, B.

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

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of Creative Commons Attribution 4.0 International (CC BY 4.0) License. For more information see our license.

08/05/2020
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


Reactome database release: 72

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective GCK causes maturity-onset diabetes of the young 2 (MODY2)

Stable identifier: R-HSA-5619073

Diseases: maturity-onset diabetes of the young

Cytosolic glucokinase (GCK) (and three isoforms of hexokinase) catalyse the irreversible reaction of alpha-D-glucose (Glc) and ATP to form alpha-D-glucose-6-phosphate (G6P) and ADP, the first step in glycolysis. In the body, GCK is found only in hepatocytes and pancreatic beta cells. GCK and the hexokinase enzymes differ in that GCK has a higher $K_m$ than the hexokinases and is less readily inhibited by the reaction product. As a result, GCK should be inactive in the fasting state when glucose concentrations are low but in the fed state should have an activity proportional to glucose concentration. These features are thought to enable efficient glucose uptake and retention in the liver, and to function as a sensor of glucose concentration coupled to insulin release in pancreatic beta cells. Defects in GCK are can cause maturity-onset diabetes of the young 2 (MODY2; MIM:125851), a heritable early onset form of type II diabetes (Hussain 2010, Osbak et al. 2009).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Author/Editor</th>
<th>Reviewed</th>
</tr>
</thead>
<tbody>
<tr>
<td>2014-08-22</td>
<td>Authored, Edited</td>
<td>Jassal, B.</td>
</tr>
<tr>
<td>2015-08-04</td>
<td>Reviewed</td>
<td>Broer, S.</td>
</tr>
</tbody>
</table>
Defective GCK does not phosphorylate Glc to form G6P ➤

Location: Defective GCK causes maturity-onset diabetes of the young 2 (MODY2)

Stable identifier: R-HSA-5621918