Defective SLC6A3 causes Parkinsonism-dystonia infantile (PKDYS)

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.

09/05/2020
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 SLC6A3 causes Parkinsonism-dystonia infantile (PKDYS)

**Stable identifier:** R-HSA-5619081

**Diseases:** neurodegenerative disease

The human gene SLC6A3 encodes the sodium-dependent dopamine transporter DAT which mediates the Na-dependent re-uptake of dopamine (DA) from the synaptic cleft back into cells, thereby terminating the action of DA (Broer & Gether 2012, Schweikhard & Ziegler 2012). Defects in SLC6A3 can cause Parkinsonism-dystonia infantile (PKDYS; MIM:613135), a neurodegenerative disorder characterised by infantile onset of parkinsonism and dystonia (Kurian et al. 2011).

**Literature references**


**Editions**

<table>
<thead>
<tr>
<th>Date</th>
<th>Author/Editor</th>
<th>Reviewer</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 SLC6A3 does not cotransport DA, Na+ from extracellular region to cytosol

**Location:** Defective SLC6A3 causes Parkinsonism-dystonia infantile (PKDYS)

**Stable identifier:** R-HSA-5660706