Diseases associated with visual transduction

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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: 76

This document contains 3 pathways (see Table of Contents)
Diseases associated with visual transduction

Stable identifier: R-HSA-2474795

Diseases: retinal disease

The process of vision involves two stages; the retinoid cycle which supplies and regenerates the visual chromophore required for vision and phototransduction which propagates the light signal. Defects in the genes involved in the retinoid cycle cause degenerative retinal diseases. These defective genes are described here (for reviews see Travis et al. 2007, Palczewski 2010, Fletcher et al. 2011, den Hollander et al. 2008).

Literature references


Editions

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Retinoid cycle disease events

Location: Diseases associated with visual transduction

Stable identifier: R-HSA-2453864

Diseases: retinal disease

The gene defects which cause diseases related to the retinoid cycle are described here (Travis et al. 2007, Palczewski 2010, Fletcher et al. 2011, den Hollander et al. 2008).

Literature references


Editions

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Retinoid metabolism disease events

Location: Diseases associated with visual transduction

Stable identifier: R-HSA-6809583

Diseases: retinal disease

Retinol binding protein (RBP4) delivers all-trans-retinol (atROL) from liver stores to peripheral tissues. Defects in RBP4 cause retinol-binding protein deficiency (RBP deficiency, MIM:180250), causing night vision problems and a typical 'xerophthalmic fundus' with progressive atrophy of the retinal pigment epithelium (RPE) (Seeliger et al. 1999, Biesalski et al. 1999).

Literature references


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