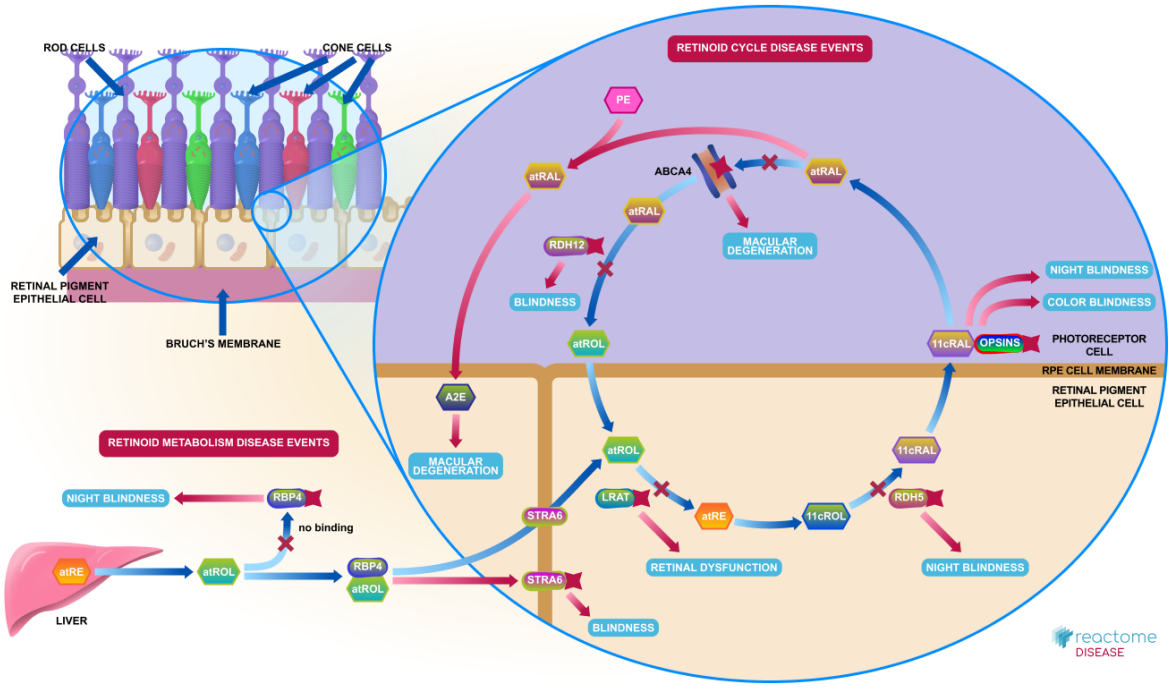
Diseases associated with visual transduction



Blaner, WS., Jassal, B.

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

07/09/2021

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

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology, 14*, e1005968. ↗

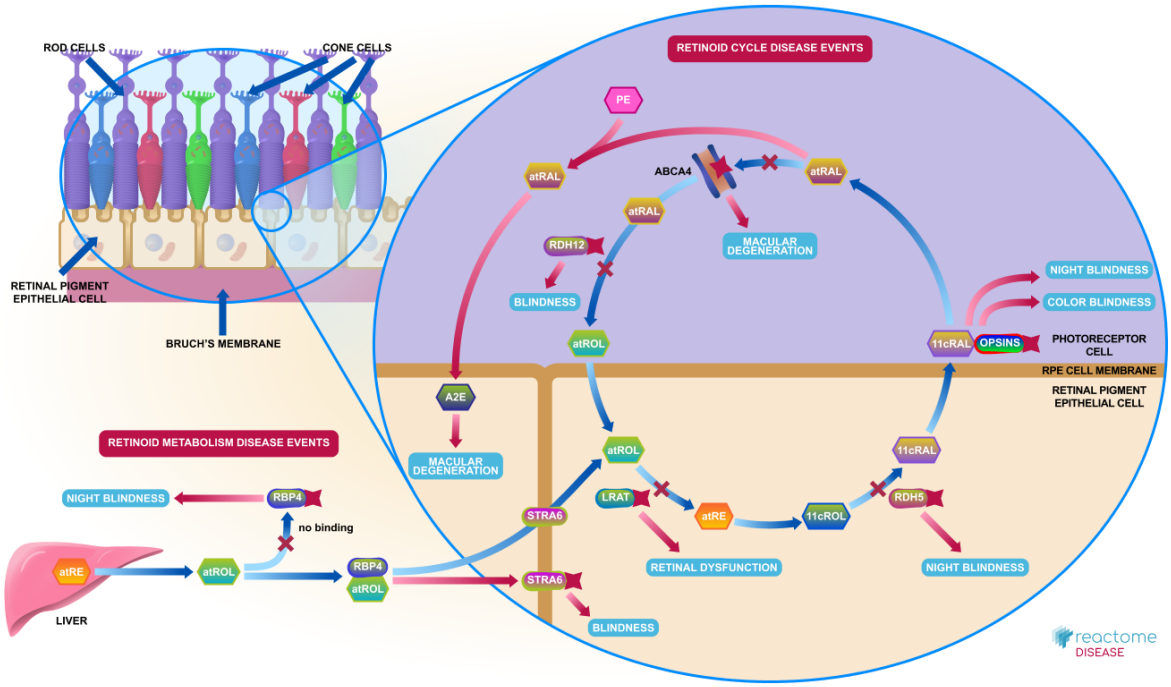
Reactome database release: 77

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

Travis, GH., Golczak, M., Moise, AR., Palczewski, K. (2007). Diseases caused by defects in the visual cycle: retinoids as potential therapeutic agents. *Annu. Rev. Pharmacol. Toxicol., 47*, 469-512.

Fletcher, EL., Jobling, AI., Vessey, KA., Luu, C., Guymer, RH., Baird, PN. (2011). Animal models of retinal disease. *Prog Mol Biol Transl Sci, 100*, 211-86.

den Hollander, AI., Roepman, R., Koenekoop, RK., Cremers, FP. (2008). Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog Retin Eye Res, 27*, 391-419.

Palczewski, K. (2010). Retinoids for treatment of retinal diseases. *Trends Pharmacol. Sci., 31*, 284-95.

Editions

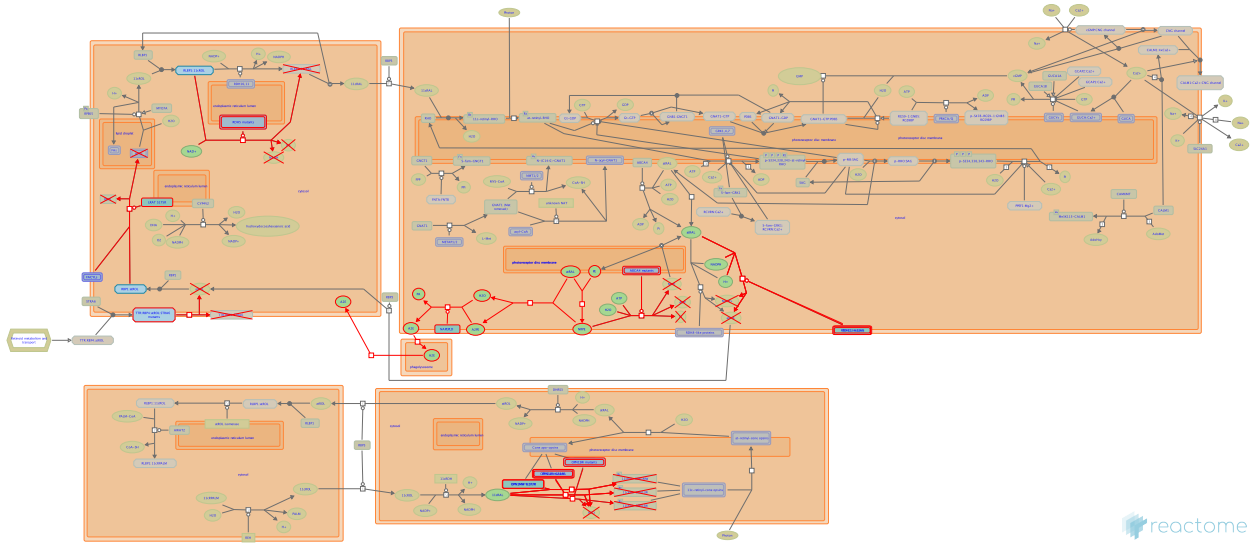
2012-09-25	Authored, Edited	Jassal, B.
2013-01-31	Reviewed	Blaner, WS.

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

Travis, GH., Golczak, M., Moise, AR., Palczewski, K. (2007). Diseases caused by defects in the visual cycle: retinoids as potential therapeutic agents. *Annu. Rev. Pharmacol. Toxicol., 47*, 469-512. ↗

Palczewski, K. (2010). Retinoids for treatment of retinal diseases. *Trends Pharmacol. Sci., 31*, 284-95. ↗

Fletcher, EL., Jobling, AI., Vessey, KA., Luu, C., Guymer, RH., Baird, PN. (2011). Animal models of retinal disease. *Prog Mol Biol Transl Sci, 100*, 211-86. ↗

den Hollander, AI., Roepman, R., Koenekoop, RK., Cremers, FP. (2008). Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog Retin Eye Res, 27*, 391-419. ↗

Editions

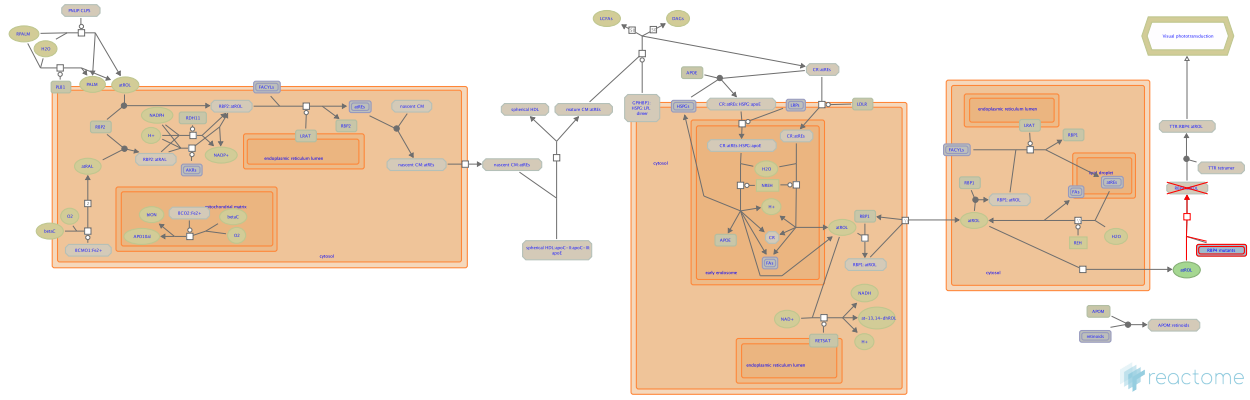
2012-08-17	Authored, Edited	Jassal, B.
2013-01-31	Reviewed	Blaner, WS.

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

Seeliger, MW., Biesalski, HK., Wissinger, B., Gollnick, H., Gielen, S., Frank, J. et al. (1999). Phenotype in retinol deficiency due to a hereditary defect in retinol binding protein synthesis. *Invest. Ophthalmol. Vis. Sci., 40*, 3-11. ↗

Biesalski, HK., Frank, J., Beck, SC., Heinrich, F., Illek, B., Reifen, R. et al. (1999). Biochemical but not clinical vitamin A deficiency results from mutations in the gene for retinol binding protein. *Am. J. Clin. Nutr., 69*, 931-6. ↗

Editions

2012-08-17	Authored, Edited	Jassal, B.
2013-01-31	Reviewed	Blaner, WS.

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