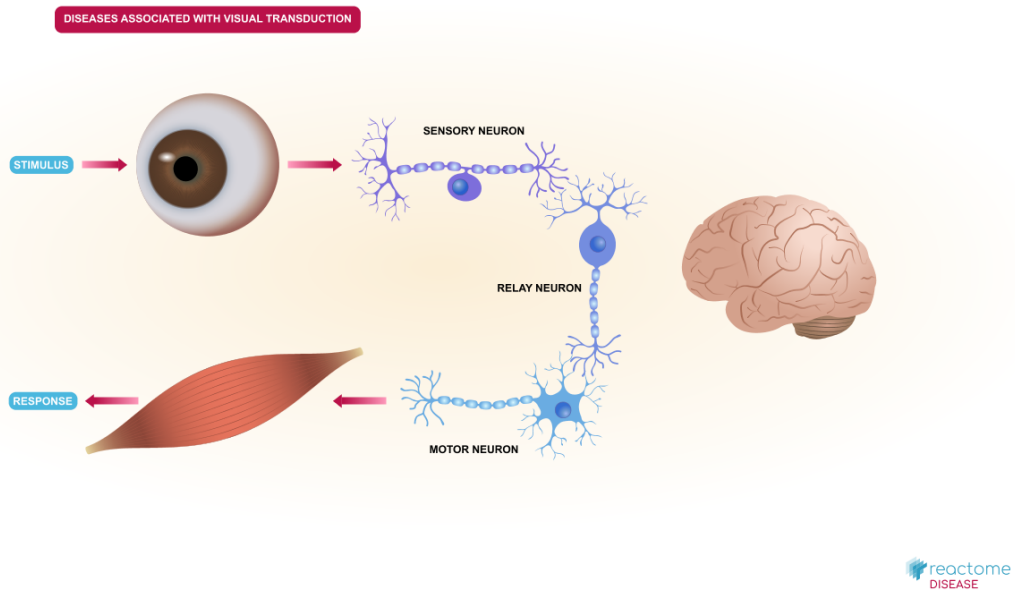
Diseases of the neuronal system



Blaner, WS., D'Eustachio, P., Jassal, B., Orlic-Milacic, M.

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.

16/09/2021

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. ↗

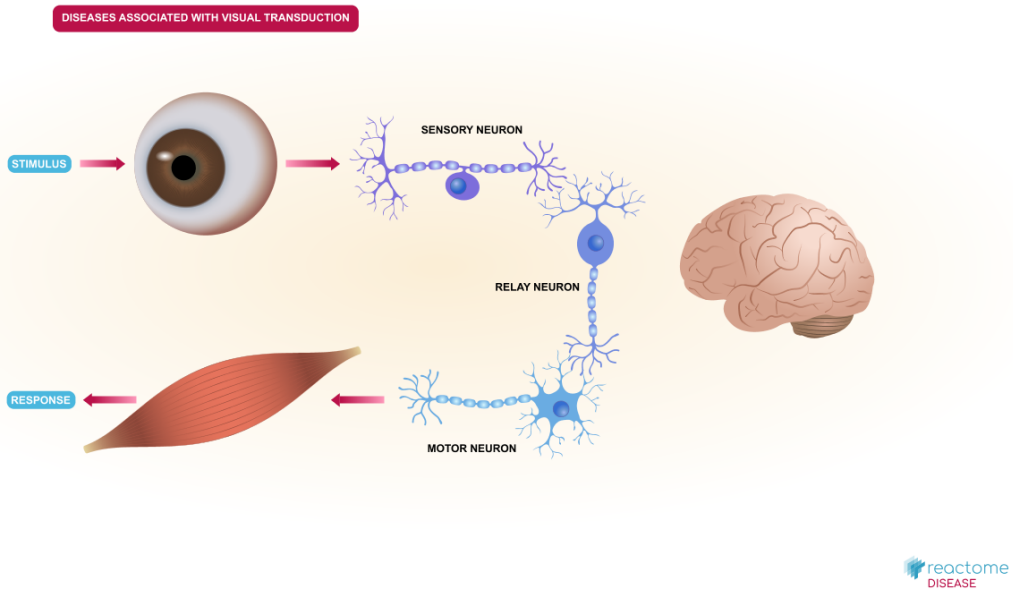
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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. ↗

Diseases of the neuronal system ↗

Stable identifier: R-HSA-9675143

Diseases: nervous system disease



Diseases of the neuronal system can affect sensory cells and transmission of signals between sensory cells and sensory neurons (Martemyanov and Sampath 2017), transmission of signals across electrical and chemical synapses in the nervous system (Picconi et al. 2012, Yin et al. 2012, Kida and Kato 2015), and transmission of signals between motor neurons and muscle cells (Sine 2012, Engel et al. 2015).

We have so far annotated diseases of visual phototransduction due to retinal degeneration caused by defects in the genes involved in the retinoid cycle (Travis et al. 2007, Palczewski 2010, Fletcher et al. 2011, den Hollander et al. 2008).

Literature references

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. ↗

Yin, DM., Chen, YJ., Sathyamurthy, A., Xiong, WC., Mei, L. (2012). Synaptic dysfunction in schizophrenia. *Adv. Exp. Med. Biol., 970*, 493-516. ↗

Engel, AG., Shen, XM., Selcen, D., Sine, SM. (2015). Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. *Lancet Neurol, 14*, 420-34. ↗

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. ↗

Picconi, B., Piccoli, G., Calabresi, P. (2012). Synaptic dysfunction in Parkinson's disease. *Adv. Exp. Med. Biol., 970*, 553-72. ↗

Editions

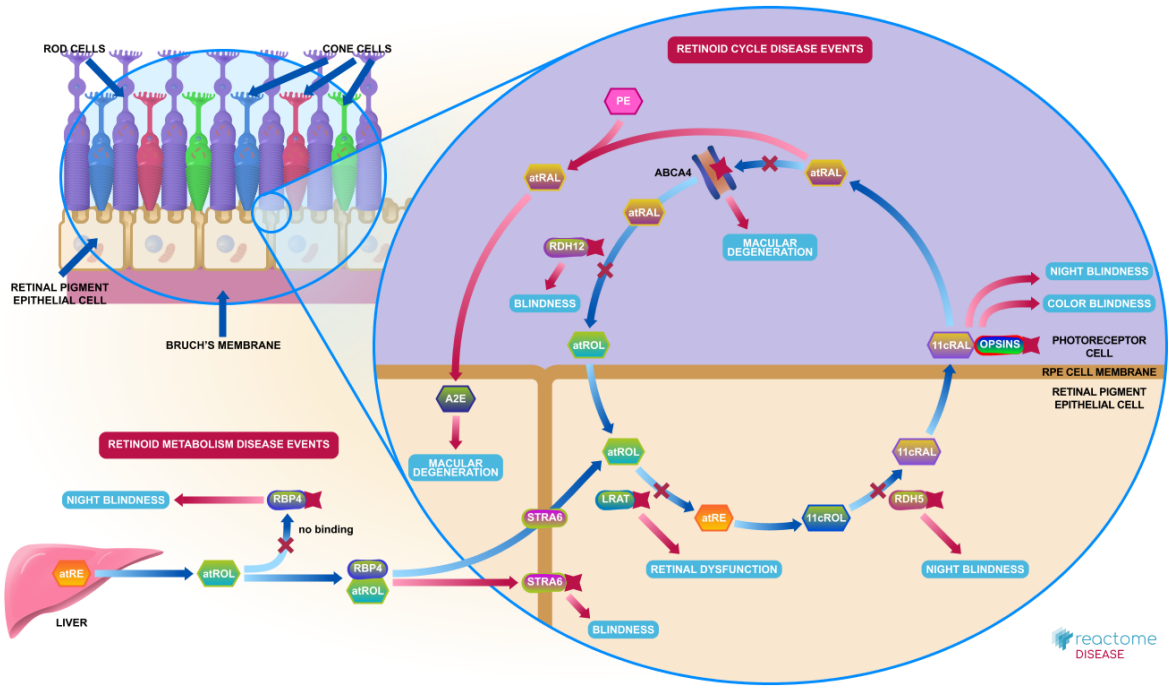
2020-02-21	Authored	Orlic-Milacic, M.
2020-02-24	Reviewed	D'Eustachio, P.
2020-02-24	Edited	Orlic-Milacic, M.

Diseases associated with visual transduction ↗

Location: Diseases of the neuronal system

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

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. ↗

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. ↗

Editions

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

Table of Contents

Introduction 1

Diseases of the neuronal system 2

Diseases associated with visual transduction 3

Table of Contents 4