

HOPE publication list

Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia

Nat Genet. 2015 Jul;47(7):757-65. Epub 2015 Jun 1

Kohl S, Zobor D, Chiang WC, Weisschuh N, Staller J, Menendez IG, Chang S, Beck SC, Garcia Garrido M, Sothilingam V, Seeliger MW, Stanzial F, Benedicenti F, Inzana F, Héon E, Vincent A, Beis J, Strom TM, Rudolph G, Roosing S, Hollander AI, Cremers FP, Lopez I, Ren H, Moore AT, Webster AR, Michaelides M, Koenekoop RK, Zrenner E, Kaufman RJ, Tsang SH, Wissinger B, Lin JH.

Peripherin-2 couples rhodopsin to the CNG channel in outer segments of rod photoreceptors

Hum Mol Genet. pii: ddu323. [Epub ahead of print Jun 24, 2014]

Becirovic E, Nguyen ON, Pappas C, Butz ES, Stern-Schneider G, Wolfrum U, Hauck SM, Ueffing M, Wahl-Schott C, Michalakakis S, Biel M (2014).

Disruption of the Retinitis Pigmentosa 28 gene Fam161a in mice affects the photoreceptor ciliary machinery and leads to retinal degeneration

Hum Mol Gene. 2014 pii: ddu242. [Epub ahead of print May 15]

Karlstetter M*, Soroush N*, Caramoy A, Dannhausen K, Scheiffert E, Mirza M, Aslanidis A, Boesl M, Stoehr H, Nagel-Wolfrum K, Tamm ER, Jäggle H, Wolfrum U, and Langmann T (*equal contributions).

Translational read-through as an alternative approach for ocular gene therapy of retinal dystrophies caused by in-frame nonsense mutations

Vis Neurosci. 10:1-8.

Nagel-Wolfrum K, Möller F, Penner I, and Wolfrum U (2014).

Identification of a Novel Neurotrophic Factor from Primary Retinal Müller Cells Using Stable Isotope Labeling by Amino Acids in Cell Culture (SILAC)

Mol Cell Proteomics. 13(9):2371-81

von Toerne C, Menzler J, Ly A, Senninger N, Ueffing M, Hauck SM (2014)

Identification of a Novel Neurotrophic Factor from Primary Retinal Müller Cells Using Stable Isotope Labeling by Amino Acids in Cell Culture (SILAC)

Phosphorylation of the Usher syndrome protein SANS controls Magi2-mediated endocytosis

Hum Mol Genet. 23(15):3923-42

Bauss K, Knapp B, Jores P, Roepman R, Kremer H, v. Wijk E, Maerker T, Wolfrum U

The progressive rod-cone degeneration (PRCD) protein is secreted through the conventional ER/Golgi-dependent pathway

Exp Eye Res. 125:217-25

Remez L, Zobor D, Kohl S, Ben-Yosef T (2014)

Mutation of POC1B in a Severe Syndromic Retinal Ciliopathy

Hum Mutat. 2014 Jul 17. doi: 10.1002/humu.22618. [Epub ahead of print]

Beck BB, Phillips JB, Bartram MP, Wegner J, Thoenes M, Pannes A, Sampson J, Heller R, Göbel H, Koerber F, Neugebauer A, Hedergott A, Nürnberg G, Nürnberg P, Thiele H, Altmüller J, Toliat MR, Staubach S, Boycott KM, Valente EM, Janecke AR, Eisenberger T, Bergmann C, Tebbe L, Wang Y, Wu Y, Fry AM, Westerfield M, Wolfrum U, Bolz HJ.

Cyr61 activates retinal cells and prolongs photoreceptor survival in rd1 mouse model of retinitis pigmentosa

Journal of Neurochemistry. 130(2):227-40

Kucharska J, Río PD, Arango-Gonzalez B, Gorza M, Feuchtinger A, Hauck SM, Ueffing M

Biallelic variants in TLL5, encoding a tubulin glutamylase, cause retinal dystrophy

Am J Hum Genet. 94:760-9.

Sergouniotis PI, Chakarova C, Murphy C, Becker M, Lenassi E, Arno G, Lek M, MacArthur DG, UCL-exomes consortium, Bhattacharya SS, Farooqi IS, Moore AT, Holder GE, Robson AG, Wolfrum U, Webster AR, Plagnol V

The neuroprotective potential of retinal Müller glial cells

Advances in Experimental and Medicinal Biology. 801:381-7

Hauck SM, von Toerne C, Ueffing M

Therapieansätze für Erbliche Netzhauterkrankungen: Von den Genen bis zum Chip.

Klin Monbl Augenheilkd. Accepted.

Arango-Gonzalez B, Leitritz M, Fischer D, Gerberding M, Paquet-Durand F und Ueffing M.

Mutations in IMPG1 Cause Vitelliform Macular Dystrophies.

Am J Hum Genet. 2013 Sep 5;93(3):571-8.

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Osteopontin inhibits osmotic swelling of retinal glial (M ller) cells by inducing release of VEGF.

Neuroscience. 2013 Aug 29;246:59-72. doi: 10.1016/j.neuroscience.2013.04.045. Epub 2013 Apr 30.

Wahl V, Vogler S, Grosche A, Pannicke T, Ueffing M, Wiedemann P, Reichenbach A, Hauck SM, Bringmann A.

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Identification of a novel mutation in the PRCD gene causing autosomal recessive retinitis pigmentosa in a Turkish family.

Mol Vis. 2013 Jun 13;19:1350-5. Print 2013.

Pach J, Kohl S, Gekeler F, Zobor D.

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Eur J Hum Genet. 2013 Apr 17. doi: 10.1038/ejhg.2013.72. [Epub ahead of print]

Gl ckle N, Kohl S, Mohr J, Scheurenbrand T, Sprecher A, Weisschuh N, Bernd A, Rudolph G, Schubach M, Poloschek C, Zrenner E, Biskup S, Berger W, Wissinger B, Neidhardt J.

Genetic Diagnostic Testing in Inherited Retinal Dystrophies.

Klin Monbl Augenheilkd. 2013 Mar;230(3):243-6

Kohl S, Biskup S (2013).

Successful subretinal delivery and monitoring of MicroBeads in mice.

PLoS One. 2013;8(1):e55173. doi: 10.1371/journal.pone.0055173. Epub 2013 Jan 28.

Fischer MD, Goldmann T, Wallrapp C, M hlfriedel R, Beck SC, Stern-Schneider G, Ueffing M, Wolfrum U, Seeliger MW.

The special electrophysiological signs of inherited retinal dystrophies.

Open Ophthalmol J. 2012;6:86-97. doi: 10.2174/1874364101206010086. Epub 2012 Oct 31.

Prokofyeva E, Troeger E, Zrenner E.

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Prokofyeva E, Zrenner E.

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Ophthalmic Physiol Opt. 2012 Jan;32(1):53-9. doi: 10.1111/j.1475-1313.2011.00883.x. Epub 2011 Nov 18.

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Glia. 2011 May;59(5):821-32. doi: 10.1002/glia.21155. Epub 2011 Feb 28.

Del Río P, Irmmler M, Arango-González B, Favor J, Bobe C, Bartsch U, Vecino E, Beckers J, Hauck SM, Ueffing M.

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Klin Monbl Augenheilkd. 2009 Dec;226(12):999-1011. doi: 10.1055/s-0028-1109684. Epub 2009 Sep 15.

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