

## REFERENCES

- (1) Berger W, Kloeckener-Gruissem B, Neidhardt J. The molecular basis of human retinal and vitreoretinal diseases. *Prog Retin Eye Res.* 2010;29(5):335-75.
- (2) Hartong DT, Berson EL, Dryja TP. Retinitis pigmentosa. *Lancet.* 2006; 368(9549):1795-809.
- (3) Rivolta C, Sharon D, DeAngelis MM, Dryja TP. Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. *Hum Mol.Genet.* 2002; 11(10):1219-1227.
- (4) Hamel C. Retinitis Pigmentosa. *Orphanet Journal of rare disease.* 2006; 1:1-12.
- (5) Dryja TP, Hahn LB, Kajiwara K, Berson EL. Dominant and digenic mutations in the peripherin/RDS and ROM1 genes in retinitis pigmentosa. *Invest Ophthalmol.Vis.Sci.* 1997; 38(10):1972-1982.
- (6) den Hollander AI, Black A, Bennet J, Cremers FP. Lighting a candle in the dark: advances in genetics and gene therapy of recessive retinal dystrophies *J Clin Invest.* 2010; 120(9):3042-3053.
- (7) Dryja TP, McGee TL, Reichel E, Hahn LB, Cowley GS, Yandell DW, et al. A point mutation of the rhodopsin gene in one form of retinitis pigmentosa. *Nature.* 1990;343: 364-366.

- (8) Cremers FPM, van de Pol TJR, van Kerkhoff EPM, Wieringa B, Ropers HH. Cloning of a gene that is rearranged in patients with choroideraemia. *Nature*. 1990; 347:674-677.
- (9) Resnikoff S, Pararajasegaram R. Blindness prevention programmes: past, present, and future. *Bull. World Health Organization*. 2003; 79:222-226.
- (10) Sitorus RS, Abidin MS, Prihartono J. Causes and temporal trends of childhood blindness in Indonesia: study at schools for the blind in Java. *Br J Ophthalmol*. 2007; 91(9):1109-1113.
- (11) Kartasasmita A, Fujiki K, Iskandar E, Sovani I, Fujimaki T, Murakami A. A novel nonsense mutation in rhodopsin gene in two Indonesian families with autosomal recessive retinitis pigmentosa. *Ophthalmic Genet*. 2011; 32(1):57-63.
- (12) Khan MI, Collin RWJ, Arimadyo K, Michael S, Azam M, Qureshi N, et al. Missense mutations at homologous positions in the fourth and fifth laminin A G-like domains of eyes shut homolog cause autosomal recessive retinitis pigmentosa. *Invest Ophthalmol. Vis. Sci*. 2010; 16: 2753-2759.
- (13) Siemiatkowska AM, Arimadyo K, Moruz LM, Astuti GDN, de Castro Miro M., Faradz SM, Zonneveld-Vrieling MN, et al. Molecular genetic analysis of retinitis pigmentosa in Indonesia using genome-wide homozygosity mapping. *Mol. Vis*. 2011;17:3013-24.
- (14) Eva PR, Whitcher JP. Vaughn and Aspbury's general ophthalmology. Fletcher CN, editor. *Retina: introduction*. Lange McGraw Hill's. 2007: 225-28.

- (15) Apte BA, Hollyfield JG. Developmental anatomy of the retinal and choroidal vasculature, *Encyclopedia of the eye*. Elsevier and Science Direct 2010; 48:9-15.
- (16) Kolb H, Fernandez E, Nelson R. Photoreceptors. *Webvision: The organization of retina and visual system*. Salt Lake City (UT) University of Utah Health Sciences Center; 1995-2005 (updated 2007 May 01).
- (17) Strauss O. The retinal pigment epithelium in visual function. *Physiol Rev*. 2005; 48:845-881.
- (18) Marc RE. Functional anatomy of the neural retina. Adkinson NF, editor. *Albert and Jakobiec's principles and practice of ophthalmology*. Elsevier Inc. 2008; 3:1-28.
- (19) Sircar S. *Principle of medical physiology*. Thieme. 2007; 735-736.
- (20) Friedman NJ, Kaiser PK. *Essentials of ophthalmology*. Elsevier Health Science. 2007; 13-15.
- (21) Kolb H. How retina works. *American Scientist*. 2003; 91:28-35.
- (22) Kahle W, Frotscher M. *Color atlas and textbook of human anatomy: nervous system and sensory organs*. Thieme. 3:350-351.
- (23) Rieke F. Mechanisms of single-photon detection in rod photoreceptors. *Methods enzymol*. 2000; 316:186-202.
- (24) Deng WT, Sakurai K, Liu J, Dinculescu A, Li J, Pang J, et al. Functional interchangeability of rod and cone transducin alpha-subunits. *Proc Natl Acad Sci U S A*. 2009; 106:17681-17686.

- (25) Germain F, Perez-Rico C, Vicente J, de la Villa P. Functional histology of the retina. *Formatex*. 2010; 914-925.
- (26) Dowling JE. *The Retina: An approachable part of the brain*. Harvard University Press. 1987;4:21-22.
- (27) Sung CH, Chuang JH. The cell biology of vision. *J.Cell Biol*. 2011;190: 953-963.
- (28) Fain GL, Matthews HF, Cornwall MC, Koutalus Y. Adaptation in vertebrate photoreceptors. *Physiol Rev*. 2001; 81:117-151.
- (29) Deterre P, Pfister C, Bigay J, Chabre M. The retinal phototransduction process: enzymatic cascade and regulation. *Biochemie*. 1987; 69:365-370.
- (30) Ripps H. Light to sight: milestones in phototransduction. *FASEB J*. 2010; 24:970-975.
- (31) Roberts JL. *From molecules to networks: an introduction to cellular and molecular neuroscience*. Academic press. 2004; 1:350-351.
- (32) Bhagavan NV. *Fat soluble vitamins, Medical biochemistry*. Academic press. 2002; 911.
- (33) Burns ME, Arshavsky VY. Beyond counting photons: Review trials and trends in vertebrate visual transduction. *Neuron*. 2005; 48:387-401.
- (34) Menon ST, May Han, Sakmar TP. Rhodopsin: Structural Basis of Molecular Physiology. *Physiol Rev*. 2001; 81:1659-1688.
- (35) Burns ME. Deactivation mechanisms of rod phototransduction: The Cogan Lecture. *Invest Ophthalmol.Vis.Sci*. 2010; 51:1282-1288.

- (36) Hims MM, Daiger SP, Inglehearn CF. Retinitis pigmentosa: genes, proteins and prospects. *Dev.Ophthalmol.* 2003; 37:109-125.
- (37) Szlyk JP, Seiple W, Fishman GA, Alexander KR, Grover S, Mahler CL. Perceived and actual performance of daily tasks: relationship to visual function tests in individuals with retinitis pigmentosa. *Ophthalmology.* 2001; 108:65-75.
- (38) Bovolenta P, Cisneros E. Retinitis pigmentosa: cone photoreceptors starving to death. *Nat.Neuroscience.* 2009; 12:44-52.
- (39) Jaissle GB., May CA, van de Pavert SA, Wenzel A, Claes May E, Giessel A, et al. Bone spicule pigment formation in retinitis pigmentosa: insights from a mouse model. *Graefes Arch Clin Exp Ophthalmol.* 2010; 248:1063-1070.
- (40) Milam AH, Li ZY, Fariss RN. Histopathology of the human retina in retinitis pigmentosa. *Prog.Retin.Eye Res.* 1998; 139:175-205.
- (41) Collin RWJ, Littink KW, Klevering BJ, van den Born LI, Koenekoop RK, Zonneveld MN, et al. Identification of a 2 Mb human ortholog of *Drosophila* eyes shut/spacemaker that is mutated in patients with retinitis pigmentosa. *Am J Hum Genet.* 2008; 83(5):594-603.
- (42) den Hollander AI, Koenekoop RK, Mohamed MD, Arts HH, Boldt K, Towns KV, et al. Mutations in *LCA5*, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. *Nat Genet.* 2007; 39(7):889-95.

- (43) den Hollander AI, Lopez I, Yzer S, Zonneveld MN, Janssen IM, Strom TM, et al. Identification of novel mutations in patients with Leber congenital amaurosis and juvenile RP by genome-wide homozygosity mapping with SNP microarrays. *Invest Ophthalmol Vis Sci.* 2007; 48(12):5690-8.
- (44) den Hollander AI, Koenekoop RK, Yzer S, Lopez I, Arends ML, Voeselek KE, et al. Mutations in the CEP290 (NPHP6) gene are a frequent cause of Leber congenital amaurosis. *Am J Hum Genet.* 2006; 79(3):556-61.
- (45) Thiadens AA, den Hollander AI, Roosing S, Nabuurs SB, Zekveld-Vroon RC, Collin RWJ, et al. Homozygosity mapping reveals PDE6C mutations in patients with early-onset cone photoreceptor disorders. *Am J Hum Genet.* 2009, 85(2):240-7.
- (46) Collin RW, van den Born LI, Klevering BJ, de Castro Miro M., Littink KW, Arimadyo K, et al. High-resolution homozygosity mapping is a powerful tool to detect novel mutations causative for autosomal recessive RP in the Dutch population. *Invest Ophthalmol.Vis.Sci.* 2011; 52: 2227-2239.
- (47) Yzer S, van den Born LI, Schuil J, Kroes HY, van Genderen MM, Boonstra FN, et al. A Tyr368His RPE65 founder mutation is associated with variable expression and progression of early onset retinal dystrophy in 10 families of a genetically isolated population. *J.Med.Genet.* 2003; 40: 709-713.

- (48) McQuillan R, Leutenegger AL, Abdel-Rahman R, Franklin CS, Pericic M, Barac-Lauc L, et al. Runs of Homozygosity in European Populations. *Am.J.Hum.Genet.* 2008; 83:359-372.
- (49) Omran H, Sasmaz G, Haffner K, Volz A, Olbrich H, Antignac C, et al. Identification of a gene Locus for Senior-Løken syndrome in the region of the nephronophthisis type 3 gene. *J Am.Soc.Nephrol.* 2002;13:75-79.
- (50) Massof RW, Fishman GA. How strong is the evidence that nutritional supplements slow the progression of retinitis pigmentosa. *Arch Ophthalmol.* 2010;128(4):493-5.
- (51) Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MA, Bender D, et al. PLINK: a tool set for whole-genome association and population-based linkage analyses. *Am J Hum Genet.* 2007; 81(3):559-75.
- (52) Rozen S, Skaletsky HJ. Primer3 on the WWW for general users and for biologist programmers. In: Krawetz S, Misener S, editors. *Bioinformatics Methods and Protocols: Methods in Molecular Biology.* Totowa, NJ: Humana Press. 2000; 365-86.
- (53) Davis J, Handford PA, Redfield C. The N1317H substitution associated with leber congenital amaurosis results in impaired interdomain packing in human CRB1 epidermal growth factor-like (EGF) domains. *J Biol Chem.* 2007;282:28807-14.
- (54) Liu Q, Zuo J, Pierce EA. The retinitis pigmentosa 1 protein is a photoreceptor microtubule-associated protein. *J Neurosci.* 2004;24:6427-36.

- (55) Pierce EA, Quinn T, Meehan T, McGee TL, Berson EL, Dryja TP. Mutations in a gene encoding a new oxygen-regulated photoreceptor protein cause dominant retinitis pigmentosa. *Nat Genet.* 1999 Jul;22(3):248-54.
- (56) Guillonneau X, Piriev NI, Danciger M, Kozak CA, Cicediyan AV, Jacobson SG, et al. A nonsense mutation in a novel gene is associated with retinitis pigmentosa in a family linked to the RP1 locus. *Hum Mol Genet.* 1999;8,:1541-1546.
- (57) Sullivan LS, Heckenlively JR, Bowne SJ, Zuo J, Hide WA, Gal A, et al. Mutations in a novel retina-specific gene cause autosomal dominant retinitis pigmentosa. *Nat Genet.* 1999;22:255-259.
- (58) Bowne SJ, Daiger SP, Hims MM, Sohocki MM, Malone KA, McKie AB, et al. Mutations in the RP1 gene causing autosomal dominant retinitis pigmentosa. *Hum Mol Genet.* 1999;8(11):2121-8.
- (59) Jacobson SG, Cicediyan AV, Iannaccone A, Weleber RG, Fishman GA, Maguire AM, et al. Disease expression of RP1 mutations causing autosomal dominant retinitis pigmentosa. *Invest Ophthalmol Vis Sci.* 2000; 41:1898-1908.
- (60) Payne A, Vithana E, Khaliq S, Hameed A, Deller J, Abu Safieh L, et al. RP1 protein truncating mutations predominate at the RP1 adRP locus. *Invest Ophthalmol Vis Sci.* 2000;41:4069-4073.



- (61) Baum L, Chan WM, Yeung KY, Lam DS, Kwok AK, Pang CP. RP1 in Chinese: Eight novel variants and evidence that truncation of the extreme C-terminal does not cause retinitis pigmentosa. *Hum.Mutat.* 2001;17: 436.
- (62) Berson EL, Grimsby JL, Adams SM, McGee TL, Sweklo E, Pierce EA, et al. Clinical features and mutations in patients with dominant retinitis pigmentosa-1 (RP1). *Invest Ophthalmol.Vis.Sci.* 2001;42:2217-2224.
- (63) Dietrich K, Jacobi FK, Tippmann S, Schmid R, Zrenner E, Wissinger B, et al. A novel mutation of the RP1 gene (Lys778ter) associated with autosomal dominant retinitis pigmentosa. *Br J Ophthalmol.* 2002;86,:328-332.
- (64) Kawamura M, Wada Y, Noda Y, Itabashi T, Ogawa S, Sato H, et al. Novel 2336-2337delCT mutation in RP1 gene in a Japanese family with autosomal dominant retinitis pigmentosa. *Am.J.Ophthalmol.* 2004;137: 1137-1139.
- (65) Gamundi MJ, Hernan I, Martinez-Gimeno M, Maseras M, Garcia-Sandoval B, Ayuso C, et al. Three novel and the common Arg677Ter RP1 protein truncating mutations causing autosomal dominant retinitis pigmentosa in a Spanish population. *BMC.Med.* 2006;7:35.
- (66) Sullivan LS, Bowne SJ, Birch DG, Hughbanks-Wheaton D, Heckenlively JR, Lewis RA, et al. Prevalence of disease-causing mutations in families with autosomal dominant retinitis pigmentosa: a screen of known genes in 200 families. *Invest Ophthalmol.Vis.Sci.* 2006;47:3052-3064.

- (67) Chen JL, Lai TYY, Tam POS, Zhang X, Lam S, Lai RYK, et al. C-compound heterozygosity of two novel truncation mutations in RP1 causing autosomal recessive retinitis pigmentosa. *Invest Ophthalmol Vis Sci.* 2010;51:2236-42.
- (68) Avedikian H. Marfan syndrome and retinopathia pigmentosa. *Klinische Monatsblätter für Augenheilkunde.* 1971;158:704-7.
- (69) Sharon D, Bruns GAP., McGee TL, Sandberg MA, Berson EL, Dryja TP. X-Linked retinitis pigmentosa: mutation spectrum of the RPGR and RP2 genes and correlation with visual function. *Invest Ophthalmol Vis Sci.* 2001;41:2712-21.
- (70) Ebenezer ND, Michaelides M, Jenkins SA., Webster A, Cheetham ME, Stockman A, et al. Identification of novel RPGR ORF15 mutations in X-linked progressive cone-rod dystrophy (XLCORD) families . *Invest Ophthalmol Vis Sci.* 2005;46:1891-8.
- (71) Acland GM, Aguirre GK, Bennet J. Long-term restoration of rod and cone vision by single dose rAAV-mediated gene transfer to the retina in a canine model of childhood blindness. *Mol Ther.* 2005;12:1072-82.