

## REFERENCES

1. Hamel C. Retinitis pigmentosa. *Orphanet J Rare Dis.* 2006;11:40
2. Liesegang TJ, Skuta GL, Cantor LB. Retinitis Pigmentosa. Retina and Vitreous. American Academy of Ophthalmology. Section 12. San Fransisco: 2006; 207-213.
3. Wright K, Spiegel P, Thompson L. Retinitis Pigmentosa and Associated Disorder. Handbook of Paediatric Retinal Disease. Springer ; 2006: 135-155
4. Pagon R, Daiger S. Retinitis Pigmentosa Overview. Cited 12 March 2009. Available in [www.genetests.org](http://www.genetests.org)
5. Sharma YR, Reddy RR, Singh DV. Retinitis Pigmentosa and Allied Disorders. *Centre for Ophthalmic Sciences.* Vol 6. 2004: 115-9
6. Bunker CH, Berson EL, Bromley WC, Hayes RP, Roderick TH. Prevalence of retinitis pigmentosa in Maine. *Am J ophtalmol.* 1984; 97 : 357-365
7. Biro pusat statistic Kotamadya Semarang 2006 . Cited 10 March 2009. [www.bps.go.id](http://www.bps.go.id)
8. Grondahl J. Estimation of prognosis and prevalence of retinitis pigmentosa and Usher syndrome in Norway. *Clin Genet* 1987; **31**: 255–64.
9. Novak-Lauš K, Suzana Kukulj S, Zoric-Geber M, Bastaic O. Primary tapetoretinal dystrophies as the cause of blindness and impaired vision in the republic of Croatia. *Acta Clin Croat* 2002; **41**: 23–27
10. Rivolta C, Berson EL, Dryja TP. Paternal uniparental heterodisomy with partial isodisomy of chromosome 1 in a patient with retinitis pigmentosa without hearing loss and a missense mutation in the Usher syndrome type II gene USH2A. *Arch Ophtalmol* 2002 ; 120:1566-71
11. Bunday S, Crews SJ. A study of retinitis pigmentosa in the City of Birmingham. I Prevalence. *J Med Genet.* 1984;21(6):417– 420.
12. Xu L, Hu L, Ma K, Li J, Jonas JB. Prevalence of retinitis pigmentosa in urban and rural adult Chinese: The Beijing Eye Study. *Eur J Ophthalmol.* 2006;16(6):865– 866

13. Budu, Hayasaka S, Syawal R, Muhiddin HS, Idris I, Yusuf I. Peripherin / RDS gene in Indonesian patients with retinitis pigmentosa : geographic comparison of polymorphic variations. *Hiroshima J Med Sci* 2005. Sep 54(3)73-6
14. Tsui I, Chao CL, Palmer N, Lin CS, Tsang SH. Phenotype-Genotype Correlations in Autosomal Dominant Retinitis Pigmentosa Caused by RHO, D190N. *Current Eye Research* 2008;33:11,1014-1022.
15. Gandra M, Anandula V, Authiappan V, Sundaramurthy S, Raman R, Bhattacharya S et al. Retinitis pigmentosa: mutation analysis of *RHO*, *PRPF31*, *RPI*, and *IMPDH1* genes in patients from India. *Molecular Vision* 2008;14:1105-1113
16. Coppieters F, Leroy BP, Beyten D, Hellemans J, De Bosscher K, Haegeman G et al. Recurrent Mutation in the First Zinc Finger of the Orphan Nuclear Receptor NR2E3 Causes Autosomal Dominant Retinitis Pigmentosa. *Am. J. Hum. Genet.* 2007;81:147-157.
17. Collin RWJ, Littink KW, Klevering J, 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 ;83:594-603.
18. Anatomy of the retina. Cited 10 January 2009. Available in [www.answersingenesis.org/tj/v13/i1/retina.asp](http://www.answersingenesis.org/tj/v13/i1/retina.asp)
19. Guyer DR, Schachat AP, Green WR. The Choroid : structural consideration .In : Schachat AP, editor. *Retina*. Philadelphia: Mosby, 2008 : 33-42
20. Henkind P, Hansen RI, Szalay J. Ocular circulation. *Physiology of the human eye and visual system*. New York: Harper & Row;1979:98-155
21. Anatomy of the choroid. Cited 14 January 2009. Available in: [http://www.accessexcellence.org/RC/VL/eye anatomy/human eye anatomy.php](http://www.accessexcellence.org/RC/VL/eye%20anatomy/human%20eye%20anatomy.php)
22. Birch DG, Anderson JL, Fish GE. Yearly rates of rod and cone functional loss in retinitis pigmentosa and cone-rod dystrophy. *Ophthalmology* 1999; **106**: 258-68.
23. Grover S, Fishman GA, Anderson RJ, Tozatti MS, Heckenlively JR, Weleber RG et al. Visual acuity impairment in patients with retinitis pigmentosa at age 45 years or older. *Ophthalmology* 1999; **106**: 1780-85.
24. Virgili G, Pierrottet C, Parmeggiani F, Pennino M, Giacomelli G, Steindler P et al. Reading performance in patients with retinitis pigmentosa: a study using the MNREAD charts. *Invest Ophthalmol Vis Sci* 2004; **45**: 3418-24.
25. Den Hollander DR, 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;39:889-895
26. Sheffield VC, Nishimura DY, Stone EM. Novel approaches to linkage mapping. *Curr Opin Genet Dev.* 1995;5:335-341
27. Lathrop GM, Lalouel JM, Julier C, Ott J. Strategies for multilocus linkage analysis in humans. *Proc Natl Acad Sci U S A.* 1984;81:3443-3446
28. **Berkeley Drosophila Genome Project** .Cited 10 January 2009. Available in: [http://www.fruitfly.org/seq\\_tools/splice.html](http://www.fruitfly.org/seq_tools/splice.html)
29. Mapped and Identified Retinal Disease 1980 - 2009. Cited 9 January 2009. Available in: <http://www.sph.uth.tmc.edu/RetNet/sum-dis.htm>
30. Bennett J. Gene therapy for Leber congenital amaurosis. *Novartis Found Symp.* 2004;255: 195-202

31. Chader GJ. Beyond basic research for inherited and orphan retinal diseases: successes and challenges. *Retina*. 2005;25:S15-S17
32. Hargrave PA. Rhodopsin structure, function, and topography: the Friedenwald lecture. *Invest Ophthalmol Vis Sci* 2001; **42**: 3–9.
33. Koutalos Y, Nakatani K, Yau KW. The cGMP-phosphodiesterase and its contribution to sensitivity regulation in retinal rods. *J Gen Physiol* 1995; **106**: 891–921.
34. Fung BK, Young JH, Yamane HK, Griswold-Prenner I. Subunit stoichiometry of retinal rod cGMP phosphodiesterase. *Biochemistry* 1990; **29**: 2657–64
35. Dhallan RS, Macke JP, Eddy RL, Shows TB, Reed RR, Yau KW et al. Human rod photoreceptor cGMP-gated channel: amino acid sequence, gene structure, and functional expression. *J Neurosci* 1992; **12**: 3248–56.
36. Korschen HG, Beyermann M, Muller F, Heck M, Vantler M, Koch KW et al. Interaction of glutamic-acid-rich proteins with the cGMP signalling pathway in rod photoreceptors. *Nature* 1999; **400**: 761–66.
37. Poetsch A, Molday LL, Molday RS. The cGMP-gated channel and related glutamic acid-rich proteins interact with peripherin-2 at the rim region of rod photoreceptor disc membranes. *J Biol Chem* 2001; **276**: 48 009–16.
38. Batra-Saff erling R, Abarca-Heidemann K, Korschen HG, Tziatzios C, Stoldt M, Budyak I et al. Glutamic acid-rich proteins of rod photoreceptors are natively unfolded. *J Biol Chem* 2006; **281**: 1449–60.
39. Palczewski K, McDowell JH, Jakes S, Ingebritsen TS, Hargrave PA. Regulation of rhodopsin dephosphorylation by arrestin. *J Biol Chem* 1989; **264**: 15770–73.
40. Weng J, Mata NL, Azarian SM, Tzekov RT, Birch DG, Travis GH. Insights into the function of Rim protein in photoreceptors and etiology of Stargardt's disease from the phenotype in abcr knockout mice. *Cell* 1999; **98**: 13–23.
41. Sun H, Nathans J. Mechanistic studies of ABCR, the ABC transporter in photoreceptor outer segments responsible for autosomal recessive Stargardt disease. *J Bioenerg Biomembr* 2001; **33**: 523–30.
42. Saari JC, Nawrot M, Kennedy BN, Garwin GG, Hurley JB, Huang J et al. Visual cycle impairment in cellular retinaldehyde binding protein (CRALBP) knockout mice results in delayed dark adaptation. *Neuron* 2001; **29**: 739–48.
43. Xue L, Gollapalli DR, Maiti P, Jahng WJ, Rando RR. A palmitoylation switch mechanism in the regulation of the visual cycle. *Cell* 2004; **117**: 761–71.
44. Moiseyev G, Chen Y, Takahashi Y, Wu BX, Ma JX. RPE65 is the isomerohydrolase in the retinoid visual cycle. *Proc Natl Acad Sci USA* 2005; **102**: 12 413–18.
45. Chen P, Hao W, Rife L, Wang XP, Shen D, Chen J et al. A photic visual cycle of rhodopsin regeneration is dependent on Rgr. *Nat Genet* 2001; **28**: 256–60.
46. Travis GH, Sutcliffe JG, Bok D. The retinal degeneration slow (rds) gene product is a photoreceptor disc membrane-associated glycoprotein. *Neuron* 1991; **6**: 61–70.
47. Connell G, Bascom R, Molday L, Reid D, McInnes RR, Molday RS. Photoreceptor peripherin is the normal product of the gene responsible for retinal degeneration in the rds mouse. *Proc Natl Acad Sci USA* 1991; **88**: 723–26.
48. Clarke G, Goldberg AF, Vidgen D, Collins L, Ploder L, Schwarz L et al. Rom-1 is required for rod photoreceptor viability and the regulation of disk morphogenesis. *Nat Genet* 2000; **25**: 67–73.

49. Saishin Y, Ishikawa R, Ugawa S, Guo W, Ueda T, Morimura H et al. Retinal fascin: functional nature, subcellular distribution, and chromosomal localization. *Invest Ophthalmol Vis Sci* 2000; **41**: 2087–95.
50. Tubb BE, Bardien-Kruger S, Kashork CD, Shaffer LG, Ramagli LS, Xu J et al. Characterization of human retinal fascin gene (FSCN2) at 17q25: close physical linkage of fascin and cytoplasmic actin genes. *Genomics* 2000; **65**: 146–56.
51. Xi Q, Pauer GJ, Marmorstein AD, Crabb JW, Hagstrom SA. Tubby-like protein 1 (TULP1) interacts with F-actin in photoreceptor cells. *Invest Ophthalmol Vis Sci* 2005; **46**: 4754–61.
52. Pellikka M, Tanentzapf G, Pinto M, Smith C, McGlade CJ, Ready DF et al. Crumbs, the *Drosophila* homologue of human CRB1/RP12, is essential for photoreceptor morphogenesis. *Nature* 2002; **416**: 143–49.
53. Liu Q, Zuo J, Pierce EA. The retinitis pigmentosa 1 protein is a photoreceptor microtubule-associated protein. *J Neurosci* 2004; **24**: 6427–36.
54. Q Zhang, F Zulfiqar, X Xiao, SA Riazuddin, Z Ahmad, R Caruso et al. Severe retinitis pigmentosa mapped to 4p15 and associated with a novel mutation in the PROM1 gene. *Hum. Genet.* 122:293-299 (2007).
55. Rice DS, Huang W, Jones HA, Hansen G, Ye GL, Xu N et al. Severe retinal degeneration associated with disruption of semaphorin 4A. *Invest Ophthalmol Vis Sci* 2004; **45**: 2767–77.
56. Boeda B, El-Amraoui A, Bahloul A, Goodyear R, Daviet L, Blanchard S et al. Myosin VIIa, harmonin and cadherin 23, three Usher I gene products that cooperate to shape the sensory hair cell bundle. *EMBO J* 2002; **21**: 6689–99.
57. Siemens J, Kazmierczak P, Reynolds A, Sticker M, Littlewood-Evans A, Muller U. The Usher syndrome proteins cadherin 23 and harmonin form a complex by means of PDZ-domain interactions. *Proc Natl Acad Sci USA* 2002; **99**: 14946–51.
58. Ahmed ZM, Riazuddin S, Ahmad J, Bernstein SL, Guo Y, Sabar MF et al. PCDH15 is expressed in the neurosensory epithelium of the eye and ear and mutant alleles are responsible for both USH1F and DFNB23. *Hum Mol Genet* 2003; **12**: 3215–23.
59. Reiners J, Van Wijk E, Marker T, Zimmermann U, Jürgens K, te Brinke H et al. Scaffold protein harmonin (USH1C) provides molecular links between Usher syndrome type 1 and type 2. *Hum Mol Genet* 2005; **14**: 3933–43.
60. Adato A, Vreugde S, Joensuu T, Avidan N, Hamalainen R, Belenkiy O et al. USH3A transcripts encode clarin-1, a four-transmembrane-domain protein with a possible role in sensory synapses. *Eur J Hum Genet* 2002; **10**: 339–50.
61. Chapple JP, Grayson C, Hardcastle AJ, Bailey TA, Matter K, Adamson P et al. Organization on the plasma membrane of the retinitis pigmentosa protein RP2: investigation of association with detergent-resistant membranes and polarized sorting. *Biochem J* 2003; **372**: 427–33.
62. NB Haider, SG Jacobson, AV Cideciyan, R Swiderski, LM Streb, C Searby et al. Mutation of a nuclear receptor gene, *NR2E3*, causes enhanced S cone syndrome, a disorder of retinal cell fate. *Nat. Genet.* 24:127-131 (2000).
63. Zhou Z, Licklider LJ, Gygi SP, Reed R. Comprehensive proteomic analysis of the human spliceosome. *Nature* 2002; **419**: 182–85.
64. Umen JG, Guthrie C. Prp16p, Slu7p, and Prp8p interact with the 3' splice site in two distinct stages during the second catalytic step of pre-mRNA splicing. *RNA* 1995; **1**: 584–97.
65. Lauber J, Plessel G, Prehn S, Will CL, Fabrizio P, Gröning K et al. The human U4/U6 snRNP contains 60 and 90kD proteins that are structurally homologous to the yeast splicing factors Prp4p and Prp3p. *RNA* 1997; **3**: 926–41.

66. Wang A, Forman-Kay J, Luo Y, Luo M, Chow YH, Plumb J et al. Identification and characterization of human genes encoding Hprp3p and Hprp4p, interacting components of the spliceosome. *Hum Mol Genet* 1997; **6**: 2117–26.
67. Maita H, Kitaura H, Keen TJ, Inglehearn CF, Ariga H, Iguchi-Arigo SM. PAP-1, the mutated gene underlying the RP9 form of dominant retinitis pigmentosa, is a splicing factor. *Exp Cell Res* 2004; **300**: 283–96.
68. Gibbs D, Azarian SM, Lillo C, Kitamoto J, Klomp AE, Steel KP et al. Role of myosin VIIa and Rab27a in the motility and localization of RPE melanosomes. *J Cell Sci* 2004; **117**: 6473–83.
69. Adato A, Michel V, Kikkawa Y, Reiners J, Alagramam KN, Weil D et al. Interactions in the network of Usher syndrome type 1 proteins. *Hum Mol Genet* 2005; **14**: 347–56.
70. Yen HJ, Tayeh MK, Mullins RF, Stone EM, Sheffield VC, Slusarski DC. Bardet-Biedl syndrome genes are important in retrograde intracellular trafficking and Kupffer's vesicle cilia function. *Hum Mol Genet* 2006; **15**: 667–77.
71. Nishimura DY, Fath M, Mullins RF, Searby C, Andrews M, Davis R et al. Bbs2-null mice have neurosensory deficits, a defect in social dominance, and retinopathy associated with mislocalization of rhodopsin. *Proc Natl Acad Sci USA* 2004; **101**: 16588–93.
72. Mykytyn K, Mullins RF, Andrews M, Chiang AP, Swiderski RE, Yang B et al. Bardet-Biedl syndrome type 4 (BBS4)-null mice implicate Bbs4 in flagella formation but not global cilia assembly. *Proc Natl Acad Sci USA* 2004; **101**: 8664–69.
73. Kim JC, Badano JL, Sibold S, Esmail MA, Hill J, Hoskins BE et al. The Bardet-Biedl protein BBS4 targets cargo to the pericentriolar region and is required for microtubule anchoring and cell cycle progression. *Nat Genet* 2004; **36**: 462–70.
74. Li JB, Gerdes JM, Haycraft CJ, Fan Y, Teslovich TM, May-Simera H et al. Comparative genomics identifies a flagellar and basal body proteome that includes the BBS5 human disease gene. *Cell* 2004; **117**: 541–52.
75. Fath MA, Mullins RF, Searby C, Nishimura DY, Wei J, Rahmouni K et al. Mkks-null mice have a phenotype resembling Bardet-Biedl syndrome. *Hum Mol Genet* 2005; **14**: 1109–18.
76. Blacque OE, Reardon MJ, Li C, McCarthy J, Mahjoub MR, Ansley SJ et al. Loss of *C elegans* BBS-7 and BBS-8 protein function results in cilia defects and compromised intraflagellar transport. *Genes Dev* 2004; **18**: 1630–42.
77. Ansley SJ, Badano JL, Blacque OE, Hill J, Hoskins BE, Leitch CC et al. Basal body dysfunction is likely cause of pleiotropic Bardet-Biedl syndrome. *Nature* 2003; **425**: 628–33.
78. Hong DH, Pawlyk B, Sokolov M, Strissel KJ, Yang J, Tulloch B et al. RPGR isoforms in photoreceptor connecting cilia and the transitional zone of motile cilia. *Invest Ophthalmol Vis Sci* 2003; **44**: 2413–21.
79. Khanna H, Hurd TW, Lillo C, Shu X, Parapuram SK, He S et al. RPGR-ORF15, which is mutated in retinitis pigmentosa, associates with SMC1, SMC3, and microtubule transport proteins. *J Biol Chem* 2005; **280**: 33 580–87.
80. Yang Z, Alvarez BV, Chakarova C, Jiang L, Karan G, Frederick JM et al. Mutant carbonic anhydrase 4 impairs pH regulation and causes retinal photoreceptor degeneration. *Hum Mol Genet* 2005; **14**: 255–65.
81. Vollrath D, Feng W, Duncan JL, Yasumura D, D'Cruz PM, Chappelow A et al. Correction of the retinal dystrophy phenotype of the RCS rat by viral gene transfer of Mertk. *Proc Natl Acad Sci USA* 2001; **98**: 12 584–89.

82. Bornancin F, Mechtcheriakova D, Stora S, **Graf C, Wlachos A, Dévay P** et al. Characterization of a ceramide v kinase-like protein. *Biochim Biophys Acta* 2005; **1687**: 31–43.
83. Bowne SJ, Sullivan LS, Blanton SH, Cepko CL, Blackshaw S, Birch DG et al. Mutations in the inosine monophosphate dehydrogenase 1 gene (IMPDH1) cause the RP10 form of autosomal dominant retinitis pigmentosa. *Hum Mol Genet* 2002; **11**: 559–68
84. Daiger SP, Sullivan LS, Bowne S. Retnet. Cited 10 January 2009 Available in: [www.sph.uth.tmc.edu/Retnet](http://www.sph.uth.tmc.edu/Retnet).
85. Daiger SP, Sullivan LS, Rodriguez JA. *Behav Brain Sci* 1995;18: 452-67
86. Geller AM, Sieving PA. Assessment of foveal cone photoreceptors in Stargardt's macular dystrophy using a small dot detection task. *Vision Res* 1993; 33: 1509–24.
87. **R.W.J. Collin, L.I. van den Born, B.J. Klevering, M. de Castro-Miró, K.W. Littink, K. Arimadyo et al.** High-Resolution Homozygosity Mapping Is a Powerful Tool to Detect Novel Mutations Causative of Autosomal Recessive RP in the Dutch Population. *IOVS* April 2011 52:2227-2239
88. Schorderet Daniel F. Escher Pascal. NR2E3 mutations in enhanced S-cone sensitivity syndrome (ESCS), Goldmann-Favre syndrome (GFS), clumped pigmentary retinal degeneration (CPRD), and retinitis pigmentosa (RP). *HUMAN MUTATION* Volume 30, Issue 11, November 2009, Pages: 1475–1485
89. Pascal E, Peter G, Raphaël R, Leila T, Sylvain B , Tania D et al. Mutations in NR2E3 Can Cause Dominant or Recessive Retinal Degenerations in the Same Family. *HUMAN MUTATION* Volume 30, Issue 3, March 2009, Pages: 342–351
90. Hartong DT, Terri L, McGee, Michael A., Sandberg, Eliot L. et al Search for a correlation between telomere length and severity of retinitis pigmentosa due to the dominant rhodopsin Pro23His mutation *Molecular Vision* 2009; 15:592-597
91. Tomas S. A, Artur V. C, Alexander S, Elizabeth AMW, Waldo H, D. Alan W, et al Retinal Lamina Architecture in Human Retinitis Pigmentosa Caused by *Rhodopsin* Gene Mutations. *Investigative Ophthalmology & Visual Science*, April 2008, Vol. 49, No. 4
92. Shahzadi A, Riazuddin SA, Ali S, Li D, Khan SN, Husnain T et al. Nonsense mutation in *MERTK* causes autosomal recessive retinitis pigmentosa in a consanguineous Pakistani family. *Br J Ophthalmol*. 2010 Aug;94(8):1094-9
93. Mackay DS, Henderson RH, Sergouniotis PI, Li Z, Moradi P, Holder GE et al. Novel mutations in *MERTK* associated with childhood onset rod-cone dystrophy. *Mol Vis*. 2010 Mar 9;16:369-77.
94. Ozgül RK, Durukan H, Turan A, Oner C, Ogiş A, Farber DB Molecular analysis of the *ABCA4* gene in Turkish patients with Stargardt disease and retinitis pigmentosa. *Hum Mutat*. 2004 May;23(5):523.
95. Burke<sup>1</sup> T R., Tsang S H. Tsang<sup>1,2</sup> Allelic and phenotypic heterogeneity in *ABCA4* mutations. *Ophthalmic Genetics*, 1–10, Early Online, 2011
96. Khan MI, Collin RWJ, Arimadyo K, Micheal 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. *Mol Vis*. 2010; 16: 2753–2759.
97. Radu RA, Yuan Q, Hu J, Peng JH, Lloyd M, Nusinowitz S, Bok D, Travis GH. Vitamin A supplementation accelerates lipofuscin accumulation in the retinal pigment epithelium of a

mouse model for ABCA4 mediated inherited retinal dystrophies. *Invest Ophthalmol Vis Sci* 2008; 49:3821-3829.

98. Thornton J, Edwards R, Mithchell P, Harrison RA, Buchan I, Kelly SP. Smoking and age-related macular degeneration: a review of association. *Eye* 2005;19:935-944.