6 LITERATURVERZEICHNIS

Abbeg M., Hafezi F., Wenzel A., Grimm C. und Remé C. (2000) Therapieversuche bei RP; Licht am Ende des Tunnels? *Klin. Monatsbl. Augenheilkd*. **216**: 83-89.

Acland G. M., Aguirre D., Ray J., Zhang Q., Aleman T. S., Cideciyan A. V., Pearce-Kelling S. E., Anand V., Zeng Y., Magire A. M., Jacobson S. G., Hauswirth W. W. und Bennett J. (2001) Gene therapy restores vision in a canine model of childhood blindness. *Nature Genet.* **28**: 92-95.

Aldred M. A. und Wright A. F. (1991) PCR detection of excisting and new polymorphisms at the TIMP locus. *Nucleic acids res*. **19**: 1165.

Aldred M. A., Dry K. L., Sharp D. M., Van Dorp D. B., Brown J., Hardwick L. J., Lester D. H., Pryde F. E., Teague P. W., Jay M., Bird A. C., Jay, B. und Wright A. F. (1992) Linkage analysis in X-linked congenital stationary night blindness. *Genomics*. **14**: 99-104.

al-Jandal N., Farrar G. J., Kiang A.-S., Humphries M. M., Bannon N., Findlay J. B. C., Humphries P. und Kenna P. F. (1999) A novel mutation within the rhodopsin gene (thr94 to ile) causing autosomal dominant congenital stationary night blindness. *Hum. Mutat.* **13**: 75-81.

Allikmets R., Singh N., Sun H., Shroyer N. F., Hutchinson A., Chidambaram A., Gerrard B., Baird L., Stauffer D., Peiffer A., Rattner A., Smallwood P., Li Y., Anderson K. L., Lewis R. A., Nathans J., Leppert M., Dean M. und Lupski, J. R. (1997) A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Stargardt macular dystrophy. *Nature Genet.* **15**: 236-246.

Aputo B. L. und Guido M. E. (2002) Shedding light on the metabolism of phospholipids in the retina. *Biochim. Biophys. Acta* **1583**: 1-12.

Assink J. J. M., Tijmes N. T., ten Brink J. B., Oostra R.-J., Riemslag F. C., de Jong, P. T. V. M. und Bergen A. A. B. (1997) A gene for X-linked optic atrophy is closely linked to the Xp11.4-Xp11.2 region of the X chromosome. *Am. J. Hum. Genet.* **61**: 934-939.

Awatramani G. B. und Slaughter M. M. (2001) Intensity-dependent, rapid activation of presynaptic metabotropic glutamate receptor at a central synapse. *J. Neurosci.* **21**: 741-749.

Bareil C., Hamel C. P., Delague V., Arnaud B., Demaille J. und Claustres M. (2001) Segregation of a mutation in *CNGB1* encoding the beta-subunit of the rod cGMP-gated channel in a family with autosomal recessive retinitis pigmentosa. *Hum. Genet.* **108**: 328-334.

Barz W. P. und Walter P. (1999) Two endoplasmatic reticulum (ER) membrane proteins that facilate ER-to-Golgi transport of glycosylphosphatidyl-anchored proteins. *Mol.Cell. Biol.* **10**: 1043-1059.

Bayes M., Giordano M., Balcells S., Grinberg D., Viageliu L., Martinez I., Ayuso C., Benitez J., Ramos-Arroyo M.A., Chivelet P., Solans T., Valverde D., Amselem S., Goossens M., Baiget M., Gonzalezduarte R., und Besmond C. (1995) Homozygous tandem duplication within the gene encoding the beta-subunit of rod phosphodiesterase as a cause for autosomal retinitis pigmentosa. *Hum. Mut.* **5**: 228-234.

Bech-Hansen N. T., Field L. L., Schramm A. M., Reedyk M., Craig I. W., Fraser N. J.und Pearce W. G. (1990) A locus for X-linked congenital stationary night blindness is located on the proximal portion of the short arm of the X chromosome. *Hum. Genet.* **84**: 406-408.

Bech-Hansen N. T., Moore B. J. und Pearce W. G. (1992) Mapping of locus for X-linked congenital stationary night blindness (CSNB1) proximal to DXS7. *Genomics*. **12**: 409-411.

Bech-Hansen N. T. und Pearce W. G. (1993) Manifestations of X-linked congenital stationary night blindness in three daughters of an affected male: demonstration of homozygosity. *Am. J. Hum. Genet.* **52**: 71-77.

Bech-Hansen N. T., Pearce W. G., Musarella M. A., Weleber R. G., Maybaum T. A., Birch D. G., Miyake Y. und Boycott K. M. (1997). Evidence for genetic heterogeneity in X-linked congenital stationary night blindness. *Am. J. Hum. Genet.* **61** Suppl. A #1339

Bech-Hansen N. T., Taylor M. J., Maybaum T. A., Pearce W. G., Koop B., Fishman G. A., Mets M., Musarella M. A. und Boycott K. M. (1998) Loss-of-function mutations in a calcium-channel alpha-1-subunit gene in Xp11.23 cause incomplete X-linked congenital stationary night blindness. *Nature Genet.* **19**: 264-267.

Bech-Hansen N. T., Naylor M. J., Maybaum T. A., Sparkes R. L., Koop B., Birch D. G., Bergen A. A., Prinsen C. F., Polomeno R. C., Gal A., Drack A. V., Musarella M. A., Jacobson S. G., Young R. S. und Weleber R. G. (2000) Mutations in *NYX*, encoding the leucine-rich proteoglycan nyctalopin, cause X-linked complete congenital stationary night blindness. *Nature Genet.* **26**: 319-23.

Bech-Hansen N. T., Tobias R., Rosenberg T., Robitaille J., Unger K. D., Zaprzelski A., Farndon P. A., Schwart M., Birch D. G., Heckenlively J. R. und Weleber G. (2002) Genetics of X-linked congenital stationary night blindness (CSNB): summary of mutation analysis. Abstract. *Am. J. Hum. Gene*. **71**: 553.

Bengtsson E., Neame P. J., Heinegard D. und Sommarin Y. (1995) The primary structure of a basic leucine-rich repeat protein, PRELP, found in connective tissues. *J. Biol. Chem*. **270**: 25639-25644.

Bennett J., Zeng Y., Bajwa R., Klatt L., Li Y. und Maguire A. M. (1998) Adenovirus-mediated delivery of rhodopsin-promoted *bcl-2* results in a delay in photoreceptor cell death in the rd/rd mouse. *Gene Ther.* **5**: 1156-1164.

Bennett J. und Maguire A. M. (2000). Gene therapy for ocular disease. *Mol. Ther.* **1**: 501-505.

Bergen A. A., ten Brink J. B., Riemslage F., Schuurman E. J. und Tijmes N. (1995) Localization of a novel X-linked congenital stationary night blindness locus: close linkage to the RP3 type retinitis pigmentosa gene region. *Hum. Mol. Genet*. **4**: 931-935.

Bergen A. A., ten Brink J. B., Riemslage F., Schuurman E. J. Meire F., Tijmes N und de Jong P. T. (1996) Conclusive evidence for a distinct congenital stationary night blindness locus in Xp21.1. *J. Med. Genet*. **33**: 869-872.

Berger W., van Duijnhoven G., Pinckers A., Smits A., Ropers H.-H. und Cremers F. (1995) Linkage analysis in a Dutch family with X-linked congenital stationary night blindness (XL-CSNB). *Hum. Genet*. **95**: 67-70.

Blochberger T., Vergnes J., Hempel J. und Hassell J. (1992) cDNA to chick lumican (corneal keratan sulfate proteoglycan) reveals homology to the small interstitial proteoglycan gene family and expression in muscle and intestine. *J. Biol. Chem*. **267**: 347-352.

van Bokhoven H., van den Hurk J. A. J. M., Bogerd L., Philippe C., Gilgenkrantz S., de Jong P., Ropers H.-H. und Cremers F. P. M. (1994) Cloning and characterization of the human choroideremia gene. *Hum. Molec. Genet.* **3**: 1041-1046.

Boycott K. M., Pearce W. G., Musarella M. A., Weleber R. G, Maybaum T. A., Birch D. G., Miyake Y., Young R. S. und Bech-Hansen, N. T. (1998) Evidence for genetic heterogeneity in X-linked congenital stationary night-blindness. *J. Hum. Genet.* **62**: 865-875.

Boycott K. M., Maybaum T. A., Naylor M. J., Weleber R. G., Robitaille J., Miyake Y., Bergen A. B., Pierpont M. E., Pearce W. G. und Bech-Hansen N. T. (2001) A summary of 20 *CACNA1F* mutations identified in 36 families with incomplete X-linked congenital stationary night blindness, and characterization of splice variants. *Hum. Genet.* **108**: 91-97.

Brown D. A. und Rose J. K. (1992) Sorting of GPI-anchored proteins to glycolipid-enriched membrane subdomains during transport to the apical cell surface. *Cell*. **68**: 533-544.

Chaum E. und Hatton M. P. (2002) Gene theapy for genetic and acquired retinal disease. *Surv. Ophthalmol*. **47**: 449-469.

Cabot A., Rozet J.-M., Gerber S., Perrault I., Ducroq D., Smahi A., Souied E., Munnich A. und Kaplan J. (1999) A gene for X-linked idiopathic congenital nystagmus (*NYS1*) maps to chromosome Xp11.4-p11.3. *Am. J. Hum. Genet.* **64**: 1141-1146.

Candille S. I., Pardue M. T., McCall M. A., Peachey N. S. und Gregg R. G. (1999) Localization of the mouse nob (no b-wave) gene to the centromeric region of the X chromosome. *Invest. Ophthalmol. Vis. Sci.* **40**: 2748-2751.

Caputto B. E. und Guido M. E. (2002) Shedding light on the metabolism of phospholipids in theretina. *Biochim. Biophys. Acta*. **1583**: 1-12.

Chen J., Flannery J. G., LaVail M. M., Steinberg R. H., Xu J. und Simon M. I. (1996) *bcl-2* overexpression reduces apoptopic photoreceptor cell death in thre different retinal degenerations. *Proc. Natl. Acad. Sci. USA*. **93:** 7042-7047.

Clarke G., Heon H. und McInnes R. (2000). Recent advances in the molecular basis of inherited photoreceptor degeneration. *Clin. Genet.* **57**: 313-329.

Collins F. S. (1995) positional cloning moves from preditional to traditional (published erratum appears in Nat. Genet. 1995Sep; 11(1): 104). *Nature Genet.* **9**: 347-350.

Condie A., Eeles R., Borresen A. L., Coles C., Cooper C. und Prosser J (1993). Detection of pointmutations in the p53 gene: comparison of single-strand conformation polymorphism, constant denaturant gel electrophoresis, and hydroxylamine and osmium tetroxide techniques. *Hum. Mut.* **2**: 58-66.

Corpuz L. M., Funderburgh J. L., Funderburgh M. L., Bootomley G. S., Prakash S. und Conrad G. W. (1996) Molecular cloning and tissue distribution of keratocan. Bovine corneal keratan sulfate proteoglycan 37A. *J. Biol. Chem*. **271**: 9759-9763.

Cremers F. P., van de Pol D. J., van Driel M., den Hollander A. I., van Haren F. J., Knoers N. V., Tijmes N., Bergen A. A., Rohrschneider K., Blankenagel A., Pinckers A. J., Deutman A. F. und Hoyng C. B. (1998) Autosomal recessive retinitis pigmentosa and cone-rod dystrophy caused by splice site mutations in the Stargardt`s disease gene ABCR. *Hum. Mol. Genet.* **7**: 355-362.

Dacheux R. F. und Miller R. F. (1976) Photoreceptor –bipolar cell trasnsmission in the perfused retina eyecup of the mudpuppy. *Science*. **191**: 963-964.

Danciger M., Heilbron V., Gao Y. Q., Zhao D. Y., Jacobson S. G. und Farber D. B. (1996) A homozygous *PDE6B* mutaion in a family with autosomal recessive retinitis pigmentosa. *Mol. Vis.* **17**: 10.

Darnell J., Lodisch H. und Baltimore D. (1990) Molecular cell biology, sec. Edition. Scientific American Books. Distributed by W. H. Freeman and Company. New York.

Daw N. W., Jensen R. J. und Brunken W. J. (1990) Rod pathways in mammalian retinae. *Trends Neurosci.* **1**: 110-115.

Dhingra A., Luyubarsky A., Jiang M., Pugh E. N., Birnbaumer L., Sterling P. und Vardi N. (2000) The light response of ON bipolar neurons requires $G\alpha_0$. *J. Neurosci.* **20**: 9053-9058.

Dry K. L., Van Dorp D. B., Aldred M. A., Brown J., Hardwick L. J.und Wright A. F. (1993) Linkage analysis in a family with complete type congenital stationary night blindness with and without myopia. *Clin. Genet.* **43**: 250-254.

Dryja T. P., Finn J. T., Peng Y.-W., McGee T. L., Berson E. L., undYau, K.-W. (1995) Mutations in the gene encoding the alpha subunit of the rod cGMP-gated channel in autosomal recessive retinitis pigmentosa. *Proc. Nat. Acad. Sci.* **92**: 10177-10181.

Dryja T. P., Hahn L. B., Reboul T. und Arnaud B. (1996) Missense mutation in the gene encoding the alpha subunit of rod transducin in the Nougaret form of congenital stationary night blindness. *Nature Genet.* **13**: 358-365.

Dryja T. P., Adams S. M., Grimsby J. L., McGee T. L., Hong D.-H., Li T., Andreasson S. und Berson E. L. (2001) Null *RPGRIP1* alleles in patients with Leber congenital amaurosis. *Am. J. Hum. Genet.* **68**: 1295-1298.

Dunbar B. S. und Skinner S. M. (1990) Preparation of monoclonal antibodies. *Methods Enzymol.* **182**: 670-679.

Feinberg A. P. und Vogelstein B. (1983) a technique for radiolabeling restriction endonuclease fragments to high specific activity. *Anal. Biochem*. **132**: 6-13.

Fergueson M. A. (1992) Glycosy-phosphatidyinositol membrane anchors: the tale of a tail. *Biochem. Soc.Trans.* **20**: 243-256.

Fisher L. W., Termine J. D. und Young M. F. (1989) Deduced protein sequence of bone small proteoglycan I (biglycan) shows homology with proteoglycan II (decorin) and several nonconnective tissue proteins in a variety of species. *J. Biol. Chem.* **264**: 4571-4576.

Ford F. R. (1961) Diseases of the Nervous System in Infancy, Childhood and Adolescence. Springfield, Ill.: Charles C Thomas (pub.) Pp. 358-359.

Friedrichsom T. Kurzchalia T. V. (1998) Microdomains of GPI-anchored proteins in living cells revealed by crosslinking. *Nature*. **394**: 802-805.

Freund C. L., Wang Q.-L., Chen S., Muskat B. L., Wiles C. D., Sheffield V. C., Jacobson S. G., McInnes R. R., Zack D. J. und Stone E. M. (1998) De novo mutations in the *CRX* homeobox gene associated with Leber congenital amaurosis. *Nature Genet.* **18**: 311-312.

Funderburg J. L., Corpuz L. M., Roth M. R., Funderburgh M. L., Tasheva E. S. und Conrad G. W. (1997) Mimecan, the 25-kDa corneal keratan sulfate proteoglycan, is a product of the gene producing osteoglycin. *J. Biol. Chem*. **272**: 28089-28095.

Gaidano G., Ballerini P., Gong J. Z., Inghirami G., Neri A., Newcomb E. W., Magrath I. T., Knowles D. M. und Dalla-Favera R. (1991) p53 mutations in human lymphoid malignancies: association with Burkitt lymphoma and chronic lymphocytic leukemia. *Proc. Natl. Acad. Sci. USA* **88**: 5413-5417.

Gal A., Orth U., Schinzel A., Testa R., Maechler M., Neugebauer M. und Bleeker-Wagemakers, E. M. (1989) Gene of X-chromosomal congenital stationary night blindness is closely linked to DXS7 on Xp. (Abstract) *Cytogenet. Cell Genet.* **51**: 1001.

Gal A., Orth U., Baehr W., Schwinger E. und Rosenberg, T. (1994) Heterozygous missense mutation in the rod cGMP phosphodiesterase beta-subunit gene in autosomal dominant stationary night blindness. *Nature Genet.* **7**: 64-68.

Garriga P., Liu X. und Khorana H. G. (1996) Structure and function in rhodopsin: correct folding and misfolding in point mutants at and in proximity to the site of the retinitis pigmentosa mutation Leu-125→Arg in the transmembrane helix C. *Proc. Natl. Acad. Sci. USA* **93**: 4560-4564.

Gieser E. P. und Falls, H. F. (1961) Hereditary retinoschisis**.** *Am. J. Ophthal.* **51**: 1193-1200.

Glass I. A., Good P., Coleman M. P. Fullwood P., Giles M. G., Lindsay S., Nemeth A. H., Davies K. E,, Wilshaw H. A. und Fielder A. (1993) Genetic mapping of a cone and rod dysfunction (Aland Island eye disease) to the proximal short arm of the human X chromosome. *J. Med. Genet.* **30**: 1044-50.

Gonzalez-Fernandez F., Kurz D., Bao Y., Newman S., Conway B. P. Young J. E., Han, D. P. und Khani S. C. (1999) 11-cis Retinal dehydrogenase mutations as a major cause of the congenital night-blindness disorder known as fundus albipunctatus. *Molec. Vis.* **5**: 41.

Gregory-Evans K und Bhattacharya S. S. (1998) Genetic blindness: current concepts in the pathogenesis of human retinal dystrophies. *Trends Genet.* **14**: 103-108.

Gräf M., Halbach E. und Kaufmann H. (1996) Erblindungsursachen in Hessen. *Klin. Monatsbl. Augenheilkd.* **215**: 50-55.

Gregg R. G., Mukhopadhyay S., Candille S. I., Ball S. L., Pardue M. T., McCall M. A. und Peachey N. S. (2003) Identification of the gene and the mutation responsible for the mouse *nob* phenotype. *Invest. Ophthalmol. Vis. Sci.* **44**: 378-384.

Grover J., Chen X. N., Korenberg J. R. und Roughley P. J. (1997) The structure and chromosome location of the human chondroadherin gene (*CHAD*).*Genomics*. **45**: 379-385.

Guex N. und Peitsch M. C. (1997) SWISS-MODEL and Swiss-Pdb Viewer: an environment for comparative protein modelling. *Electrophoresis*. **18**: 2714-2723.

Halaban R., Svedine S., Cheng E., Smicun Y., Aron R. Und Hebert D. N. (2000) Endoplasmatic reticulum retention is a common defect associated with tyrosine-negative albinism. *Proc. Natl. Acad. Sci. USA* **97**: 5889-5894.

Hardcastle A. J., David-Gray Z. K., Jay M., Bird A. C. und Bhattacharya S. S. (1997) Localization of CSNBX (CSNB4) between the retinitis pigmentosa loci RP2 and RP3 on proximal Xp. *Invest. Ophthalmol. Vis. Sci.* **38**: 2750-2755.

Heon E. und Musarella M. A. Congenital stationary night blindness: a critcal review for molecular approaches*,* eds. Wright, A. F und Jay, B., *Molecular Genetics of Inherited Eye Disorders*, Chur, Switzerland, Harwood Academic, 1994: 277-301.

Heckenlively J. R., Deidre A., Martin B. S., Arthur L., Rosenbaum M. D. (1983) Loss of electroretinographic oscillatory potentials, optic atrophy, and dysplasia in congenital stationary night blindness. *Am J Ophthalmol*. **96**: 526-534.

Henry S. P., Takanosu M., Boyd T. C., Mayne P. M., Eberspaecher H., Zhou W., de Crombrugghe B., Hook M. und Mayne R. (2001) Expression pattern and gene characterization of asporin. a newly discovered member of the leucine-rich repeat protein family. *J. Biol. Chem*. **276**: 12212-12221.

Hirasawa H., Shiells R. und Yamada M. (2002). A metabotropic glutamate receptor regulates transmitter rlease from cone presynaptic terminals in carp retinal slices. *J. Gen. Physiol.* **119**: 55-68.

Hocking A., Shinomura T. und McQuillan D. (1998) Leucine-rich repeat glycoproteins of the extracellular matrix. *Matrix Biol*. **17**: 1-19.

den Hollander A. I., ten Brink J. B., de Kok Y. J. M., van Soest S., van den Born L. I., van Driel M. A., van de Pol D. J. R., Payne A. M., Bhattacharya S. S., Kellner U., Hoyng C. B., Westerveld A., Brunner H. G., Bleeker-Wagemakers E. M., Deutman A. F., Heckenlively J. R., Cremers F. P. M. und Bergen, A. A. B. (1999) Mutations in a human homologue of Drosophila crumbs cause retinitis pigmentosa (RP12). *Nature Genet.* **23**: 217-221.

Huang S. H., Pittler S. J., Huang X., Oliveira L., Berson E. L. und Dryja T. P. (1995). *Nature Genet.* **11**: 468-471.

Huang X. I., Mehren D., Wiese R., Lee S., Tam S. W., Daniel S., Gilmore J., Shi M. und Lashkari D. (2001) High-throughput genomic and proteomic analysis using microarray technology. *Clin. Chem.* **47**: 1912-1916.

Hübner C. A. und Jentsch T. (2002) Ion channel diseases. *Hum. Mol. Genet.* **11**: 2435- 2445. Huntington's disease collaborative research group (1993) A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell.* **72**: 971–983.

Ioannou P. A., Amemiya C. T., Garnes J., Kroisel P. M., Shizuya H., Chen C., Batzer M. A. und Jong P. J. de (1994) A new bacteriophage P1-derived vector for the propagation of large human DNA fragments. *Nature Genet.* **6**: 84–89.

Iozzo R. V. (1997) The family of the small leucine-rich proteoglycan: key regulators of matrix assembly and cellular growth. *Crit Rev Biochem Mol Biol*. **32**: 141-147.

Jacobi F. K., Broghammer M., Pesch K., Zrenner E., Berger W., Meindl A. und Pusch C. M. (2000) Physical mapping and exclusion of GPR34 as the causative gene for congenital stationary night blindness type 1. *Hum. Genet*. **107**: 89-91.

Jacobi F. K., Hamel C. P., Arnaud B., Blin N., Broghammer M., Jacobi P., Apfelstedt-Sylla E. und Pusch C. (2003) A novel *CACNA1F* mutation in a french family with the incomplete type of X-linked congenital stationary night blindness. *Am. J. Ophthal.* **135**: 733-736.

Jalkanen R., Demirci F. H., Tyynismaa H., Bech-Hansen T., Meindl A., Peippo M., Mäntyjärvi M., Gorin M. B. und Alitalo T. (2003) A new genetic locus for X-linked progressive cone-rod dystrophy. *J. Med. Genet*. **40**: 418-423.

Johnson H. J., Rosenberg L., Choi H. U., Garza S., Hook M. und Neame P. J. (1997) Characterization of epiphycan, a small proteoglycan with a leucine-rich repeat core protein. *J. Biol. Chem*. **272**: 18709-18717.

Jomary C., Vincent K. A., Grist J., Neal M. J. und Jones S. E. (1997) Rescue of photoreceptor function by AAV-mediated gene transfer in a mouse model of inherited retinal degeneration. *Gene Ther.* **4**: 683-690.

Jones M. H., Furlong R. A., Burkin H., Chalmers I. J., Brown G. M., Khwaja O. und Affara N. A. (1996) The Drosophila developmental gene fat facets has a human homologue in Xp11.4 which escapes X-inactivation and has related sequences on Yq11.2.*Hum. Mol. Genet.* **5**: 1695-701.

Joos K. und Chirmule N. (2003) Immunity to adenovirus and adeno-associated viral vectors: implications for gene therapy. *Gene Therapy.* **10**: 955-963.

Joseph R. M., und Li T. (1998) Overexpression of *Bcl-2* or *Bcl-XL* transgenes and photoreceptor degeneration. *Invest. Ophthalmol. Vis. Sci.* **37**: 2434-2446.

Kajiwara K., Hahn L. B., Mukai S., Travis G. H., Berson E. L. und Dryja T. P. Mutations in the human retinal degeneration slow gene in autosomal dominant retinitis pigmentosa. *Nature.* **354**: 480-483.

Kaupp U. B. und Koch K. W. (1992) Role of cGMP and Ca^{2+} in verterbrate photoreceptor excitation and adaption. *Annu. Rev. Physiol*. **54**: 153-175.

Kawamura S. (1995) Photransduction, excitation and adaptation. Neurobiology and Clinical Aspects of the Outer Retina (Eds. Djamgoz, M.B.A., Archer, S.N. and Vallerga, S.) Chapman & Hall, London, pp. 105-131.

Kenyon J. R. und Craig I. W. (1999) Analysis of the 5`regulatory region of the human Norrie`s disease gene: evidence that a non-translated CT dicnucleotide repeat in exon one has a role in controlling expression. *Gene*. **227**: 181-188.

Kim S. H., Bush R. A. und Sieving P. A. (1997) Increased phase lag of the fundamental harmonic component of the 30 Hz flicker ERG in Schubert-Bornschein complete type of CSNB. *Vision Res*. **37**: 2471-2475.

Kirschner R., Rosenberg T., Schultz-Heienbrok R., Lenzner S., Feil S., Roepman R., Cremers F. P. M., Ropers H.-H. und Berger W. (1999) RPGR transcription studies in mouse and human tissues reveal a retina–specific isoform that is disrupted in a patient with X-linked retinitis pigmentosa. *Hum. Mol. Genet.* **8**: 1571-1578.

Kishimoto M., Sakura H., Hayashi K., Akanuma Y., Yazaki Y., Kasuga M. und Kadowaki T. (1992) Detection of mutations in the human insulin gene by single-strand conformation polymorphisms. *J. Clin. Endocrinol. Metab.* **74**: 1027-1031.

Knapp A. G. und Mistler L. A. (1983) Response proberties of cells in rabbit`s lateral genicualte nucleus during reversible blockade of retinal on-center channel. *J. Neurophysiol.* **50**: 1236-1245.

Kobe B. und Deisenhofer J. (1994) The leucine-rich repeat: a versatile binding motif. *TIBS*. **19**: 415-421.

Kohl S., Marx T., Giddings I., Jagle H., Jacobson S. G., Apfelstedt-Sylla E. Zrenner E., Sharpe L. T. und Wissinger B. (1998) Total colourblindness is caused by mutations in the gene encoding the α-subunit of the cGMP-gated cation channel. *Nature Genet.* **19**: 257-259.

Kohl S., Baumann B., Broghammer M., Jagle H., Sieving P., Keller U., Spegal R., Anastasia M., Zrenner E., Sharpe L. T. und Wissinger B. (2000) Mutation analysis in the *CNGB3* gene encoding the β-subunit of the con photoreceptor cGMP-gated channel are responsible for achromatopsia (ACHM3) linked to chromosome 8q21. *Hum. Mol. Genet.* **9**: 2107-2116.

Kohler G. und Milstein C. (1975) Continuous cultures of fused cells secreting antibody of predefined specificity. *Nature.* **256**: 495-497.

Koutalos Y. und Yau K. W. (1996) Regulation of sensitivity in verterbrate rod photoreceptors by calcium. *Trends Neurosci.* **19**: 73-81.

Krantz D. E. und Zipursky S. L. (1990). Drosophila chaoptin, a member of the leucine-rich repeat family, is a phoreceptor cell-specific adhesion molecule. *EMBO J.* **6**: 1969-1977.

Krisch B., Feindt J. und Mentlein R. (1998) Immunoelectronmicroscopic analysis of the ligand-induced internalization of the somatostatin receptor subtype 2 in cultured human glioma cells. *J. Histochem. Cytochem.* **46**: 1233-1242.

Krumpaszky H. G., Lüdtke R., Mickler A., Klauss V. und Selbmann H. K. (1999) Blindness incidensce in Germany. *Ophthalmologica*. **213**: 176-182.

Krusius T. und Ruoslahti E. (1986). Primary structure of an extracellular matrix proteoglycan core protein deduced from cloned cDNA. *Proc. Natl. Acad. Sci. USA*. **83**: 7683-7687.

Ioshikhes I. P. und Zhang M. Q. (2000) Large-scale human promoter mapping using CpG islands. *Nature Genet.* **26**: 61-63.

Kyte J. und Doolittle R. F. (1982) A simple method for displaying the hydropathic character of a protein. *J. Mol. Biol.* **157**: 105-132.

Lacy S. E., Bonnemann C. G., Buzney E. A. und Kunkel L. M. (1999) Identification of FLRT1, FLRT2, and FLRT3: a novel family of transmembrane leucine-rich repeat proteins. *Genomics*. **62**: 417-426.

Lamb T. D. (1996) Gain and kinetics of activation in the G-protein cascade of phototransduction. *Proc. Natl. Acad. Sci. USA.* **93**: 566-570.

Leong S. R., Baxter R. C., Camerato T., Dai J. und Wood W. I. (1992) Structure and functional expression of the acid-labile subunit of the insulin-like growth factor-binding protein complex. *Mol Endocrinol.* **6**: 870-876.

Li Y., Bleeker-Wagenmakers E., Artlich A., Orth U., Wright A. F., Schinzel A. und Gal A. (1991) The locus for X-chromosomal congenital stationary night blindness is closely linked to TIMP in two families but not in a third one. *Cytogenet. Cell Genet*. **58**: 2075.

Linari M., Ueffing M., Manson F., Wright A., Meitinger T. und Becker J. (1999) The Retinitis Pigmentosa GTPase Regulator, *RPGR*, interacts with the Delta Subunit of Rod Cyclic GMP Phosphodiesterase. *Proc. Natl. Acad. Sci. USA*. **96**: 1315-1320.

Lindenov H. (1945). The Etiology of Deaf-mutism with Special Reference to Heredity. Copenhagen: E. Munksgaard (pub.).

Linn S. C., van den Rijn M. und Giaccone G. (2003) Novel approaches; improved diagnostics and therapeutics with DNA microarrays. *Ned. Tijdschr. Geneeskd.* **147**: 795-799.

Marchese A., Sawzdargo M., Nguyen T., Cheng R., Heng H. H. Q., Nowak T., Im D.-S., Lynch K. R., George S. R. und O'Dowd B. F. (1999) Discovery of three novel orphan G-protein-coupled receptors. *Genomics* **56**: 12-21.

Madisen L., Neubauer M., Plowman G., Rosen D., Segarini P., Dasch J., Thompson A., Ziman J., Bentz H. und Purchio A. F. (1990) Molecular cloning of a novel bone-forming compound: osteoinductive factor. *DNA Cell Biol*. **9**: 303-309.

Marlhens F., Bareil C., Griffoin J.-M., Zrenner E., Amalric P., Eliaou C., Liu S.-Y., Harris E., Redmond T. M., Arnaud B., Claustres M. und Hamel C. P. (1997) Mutations in *RPE65* cause Leber's congenital amaurosis. *Nature Genet.* **17**: 139-141.

Martinez-Mir A., Paloma E., Allikmets R., Ayuso C., del Rio T., Dean M., Vilageliu L., Gonzalez-Duarte R. und Balcells S. (1998) Retinitis pigmentosa caused by a homozygous mutation in the Stargardt disease gene *ABCR*. *Nature Genet.* **18**: 11-12.

Masu M., Iwakabe H., Tagawa Y., Miyoshi T., Yamashita M., Fukuda Y., Sasaki H., Hiroi K., Nakamura Y., Shigemoto R., Takada M., Nakamura K., Nakao K., Katsuki M. und Nakanishi S. (1995) Specific deficit of the ON response in visual transmission by targeted disruption of themGluR6 gene. *Cell.* **80**: 757-765.

Mayor S., Rothberg K. G. und Maxfield F. R. (1994) Sequestration of GPI-anchored proteins in caveolae triggered by cross-linking. *Science.* **264**: 1948-1951.

McDowell D. G., Burns N. A. und Parkes H. C. (1998) Localised sequence regions processing high melting temperatures prevent the amplification of a DNA mimic in competitive PCR. *Nucleic Acids Res.* **26**: 3340-3347.

McLaughlin M. E., Sandberg M. A., Berson E. L. und Dryja T. P. (1993) Recessive mutations in the gene encoding the beta-subunit of rod phosphodiesterase in patients with retinits pigmentosa. *Nature Genet.* **4**: 130-134.

Meindl A., Dry K., Hermann K., Manson F., Ciccodicola A., Edgar A., Carvaloh M. R. S., Achatz H., Hellbrand H., Lennon A., Migliaccio C., Porter K., Zrenner E., Bird A., Jay M., Lorenz B., Wittwer B., D`Urso M., Meitinger T. und Wright A. (1996) A gene (*RPGR*) with homology to the *RCC1* guanine nucleotide exchange factor is mutated in X-linked retinitis prgmentosa (RP3). *Naure. Genet.* **13**: 35-42.

Michaud J., Brody L. C., Steel G., Fontaine G., Martin L. S., Valle D. und Mitchell G. (1991) Strand-separating conformational polymorphism analysis: efficacy of detection of point mutations in the human ornithine δ-aminotransferase gene. *Genomics.* **13**: 389-394.

Milner N., Mir K. und Southern E. M. (1997) Selecting effective antisense reagents on combinatorial oligonucleotide arrays. *Nature Biotechnol.* **15**: 523-541.

Milstein C. (1980) Monoclonal antibodies. *Sci Am.* **243**: 66-74.

Miyake Y., Yagasaki K., Horiguchi M., Kawase Y. und Kanda T. (1986) Congenital stationary night blindness with negative electroretinogram: a new classification. *Arch. Ophthal.* **104**: 1013-1020.

Miyake Y., Horiguchi M., Ota I. und Shiroyama N. (1987). Characteristic ERG-flicker anomaly in incomplete congenital stationary night blindness. *Invest. Ophthalmol. Vis. Sci.* **28**: 1816-23.

Miyake Y., Horiguchi M., Terasaki H. und Konodo M. (1994) Scotopic treshold response in complete and incomplete types of congenital stationary night blindness. *Invest. Ophthalmol. Vis. Sci.* **35**: 3770-3775.

Molday R. S. (1998). Photoreceptor membrane proteins, phototransduction, and retinal degenerative diseases. The Friedenwald Lecture. *Invest. Ophthalmol. Vis. Sci*. **39**: 2491-2513.

Monaco A. P., Neve R. L., Colletti-Feener C., Bertelson C. J., Kurnit D. M. und Kunkel L. M. (1986) Isolation of candidate cDNAs for portions of the Duchenne muscular dystrophy gene. *Nature.* **323**: 646–650.

Morgan K. und Kalsheker N. A. (1997) Regultion of the serine proteinase inhibitor (SERPIN) gene a1-antitrypsin: a paradigm for other SERPINs. *Internat. J. Biochem. Cell. Biol.* **29**: 1501-1511.

Morimura H., Fishman G. A., Grover S. A., Fulton A. B., Berson E. L. und Dryja T. P. (1998) Mutations in the *RPE65* gene in patients with autosomal recessive retinitis pigmentosa or Leber congenital amaurosis. *Proc. Nat. Acad. Sci.* **95**: 3088-3093.

Morton A. S. (1893) Two cases of hereditary congenital night-blindness without visible fundus change. *Trans. Ophthal. Soc. U.K.* **13**: 147-150.

Musarella M. A., Weleber R. G., Murphey W. H., Young R. S. L., Anson-Cartwright L., Mets M., Kraft S. P., Polemeno R., Litt M. und Worton R. G. (1989) Assignment of the gene for complete X-linked congenital stationary night blindness (CSNB1) to human chromosome Xp11.3. *Genomics.* **5**: 727-737.

Nakamura M., Ito S., Terasaki H. und Miyake Y. (2001) Novel *CACNA1F* Mutations in Japanese Patients with incomplete congenital stationary night blindness. *Invest. Ophthalmol. Vis. Sci.* **42**: 1610-1616.

Nawy S. und Jahr C. E. cGMP-gated conductance in retinal bipoalr cells is supressed by the photoreceptor transmitter. *Neuron*. **7**: 677-683.

Neame P. J., Sommarin Y., Boynton R. E. und Heinegard D. (1994) The structure of a 38-kDa leucine-rich protein (chondroadherin) isolated from bovine cartilage. *J. Biol. Chem*. **269**: 21547-215544.

Nelson R. (1977) A comparison of electrical proberties of neurons in the Necturus retina. *J. Neurophysiol.* **36**: 519-535.

Nose A., Takeichi M. und Goodman C. S. (1994). Ectopic expression of connectin reveals a repulsive function during growth cone guidance and synapse formation. *Neuron.* **13**: 525-539.

Oldberg A., Antonsson P., Lindblom K. und Heinegard D. (1989) A collagen-binding 59-kd protein (fibromodulin) is structurally related to the small interstitial proteoglycans PG-S1 and PG-S2 (decorin). *EMBO*. **8**: 2601-2604.

Ollendorff V., Noguchi T., deLapeyriere O. und Birnbaum D. (1994) The GARP gene encodes a new member of the family of leucine-rich repeat-containing proteins. *Cell Growth Differ.* **5**: 213-219.

Osterberg, G. (1935) Topography of the layer of rods and cones in the human retina. *Acta Ophthal*., suppl. **6**: 1-103.

Palade G. (1975) Intracellular aspects of the process of protein secretion. *Science*. **189**: 347-358.

Park S. H., Lee S.-G., Kim Y. und Song K. (1998) Assignment of a human putative RNA helicase gene, *DDX3*, to human X chromosome bands p11.3-p11.23. *Cytogenet. Cell Genet.* **81**: 178-179.

Pardue M. T., McCall M. A., LaVail M. M., Gregg R. G. und Peachey N. S. (1998) A naturally occuring mouse modell of X-linked congenital stationary night blindness. *Invest. Ophthalmol. Vis. Sci.* **39**: 2443-2449.

Pearce W. G., Reedyk M. und Coupland S. G. (1990) Variable expressivity in X-linked congenital stationary night blindness. *Canad. J. Ophthal.* **25**: 3-10.

Peng C., Rich E. D. und Varnum M. D. (2003) Achromatopsia-associated mutation in the human cone photoreceptor cyclic nucleotide-gated channel subunit CNGB3 subunit alters the ligand sensitivity and pore properties of heteromeric channels. *J. Biol. Chem.* **278**: 34533-34540.

Pesch K., Zeitz C., Fries J., Münscher S., Pusch C. M., Kohler K., Berger W. und Wissinger B. (2003) Isolation of the mouse nyctalopin gene Nyx and expression studies in mouse and rat retina. *Invest. Ophthalmol. Vis. Sci.* **44**: 2260-2266.

Peitsch M. C., Wells T. N., Stampf D. R., Sussman J. L. (1995) The Swiss-3DImage collection and PDB-Browser on the World Wide Web. *Trend Biochem. Sci*. **20**: 82-84.

Peitsch M. C. (1996). ProMod and Swiss Model: Internet-based tools for automated comparative protein modelling. *Biochem. Soc. Trans*. **24**: 274-279.

Pietrobon D. (2002) Calcium channels and channelopathies of the central nervous system. *Mol. Neurobiol.* **25**: 31-50.

Polans A, Baehr W und Palczewski K. (1996) Turned on by Ca^{2+}! The physiology and pathologyof Ca^{2+}–binding proteins in the retina. *Trends Neurosci.* **19**: 547-554.

Pusch C., Zeitz C., Brandau O., Pesch K., Achatz H., Feil S., Scharfe C., Maurer J., Jacobi F. K., Pinckers A., Andreasson S., Hardcastle A. J., Wissinger B., Berger W. und Meindl A. (2000) The complete form of X-linked congenital stationary night blindness is caused by mutations in a gene encoding a leucin-rich repeat protein. *Nature Genet*. **26**: 324-327.

Pusch C. M., Maurer J., Ramser J., Tomiuk J., Achatz H., Pesch K., Lichtner P., Apfelstedt-Sylla E., Jacobi F. K., Berger W., Meindl A. und Wissinger B. (2001) Complete form of congenital stationary night blindness: refined mapping and evidence of genetic homogeneity. *Int. J. Mol. Med*. **7**: 155-161.

Raper S. E., Yudkoff M., Chirmule N., Gao G. P., Nunes F., Haskal Z. J., Furth E. E., Propert K. J., Robinson M. B., Magosin S., Simoes H., Speicher L., Hughes J., Tazelaar J., Wivel N. A., Wilson J. M. und Batshaw M. L. (2002). A pilot study of in vivo liver-directed gene transfer. *J. Virol.* **74**: 3555-3565.

Reardon A. L., Le Goff M., Briggs M. D., McLeod D., Sheehan J. K., Thornton D. J. und Bishop P. N. (2000) Identification in vitreous and molecular cloning of opticin, a novel member of the family of leucine-rich repeat proteins of the extracellular matrix. *J. Biol. Chem*. **275**: 2123-2329.

Rodgers W., Crise B. und Rose JK. (1994) Signals determining protein tyrosine kinase and glycosyl-phosphatidylinositol-anchored protein targeting to a glycolipid-enchriched membrane fraction. *Mol. Cell. Biol.* **14**: 5384-5391.

Roepman R., Bauer D., Rosenberg T., van Duijnhoven G., van de Vosse E., Platzer M., Rosenthal A., Ropers H.-H., Cremers F. P. M. und Berger W. (1996) Identification of a gene disrupted by a microdeletion in a patient with X-linked retinitis pigmentosa (XLRP). *Hum. Mol. Genet*. **9**: 2095-2105.

Rommens J. M., Jamuzzi M. C., Kerem B.–S., Drumm M. L., Melmer G., Dean M., Rozmahel R., Cole J. L., Kennedy D., Hidaka N, Zsiga M., Buchwald M., Riordan J. R., Tsui L.-C. und Collins F. S. (1989) Identification of the cystic fibrosis gene: chromosome walking and jumping. *Science.* **245**: 1059-1065.

Rothberg J. M., Hartley D. A., Walther Z. und Artavanis-Tsakonas S. (1988) slit: an EGF-homologous locus of D. melanogaster involved in the development of the embryonic central nervous system. *Cell.* **55**: 1047-1059.

Rothman J. E. (1994) Mechanisms of intracellular transport. *Nature*. **372**: 55-63.

Royer-Pokora B., Kunkel L. M., Monaco A. P., Goff S. C., Newburger P. E., Baehner R. L., Cole F. S., Curnutte J. T. und Orkin S. H. (1986) Cloning of the gene for an inherited human disorder-chronic granulomatous disease-on the basis on its chromosomal location. *Nature*. **322**: 32-38.

Rozzo C., Fossarello M., Galleri G., Miano G. M., Ciccodicola A., Sole G. und Piratsu M. (1999). Complete congenital stationary night blindness maps on Xp11.4 in a Sardinian family. *Eur. J. Hum. Genet.* **7**: 574-578.

Ruether K., Apfelstedt-Sylla E und Zrenner E. (1993) Clinical findings with congenital stationary night blindness of the Schubert-Bornschein type. *Ger. J. Ophthalmol*. **2**: 429-435.

Sambrook J., Fritsch E. F. und Maniatis T. (1989) Molecular Cloning – A Laboratory Manual. (Cold Spring Habour, New York. Cold Spring Habour Laboratory Press).

Sauer C. G., Gehrig A., Warneke-Wittstock R., Marquardt A., Ewing C. C., Gibson A., Lorenz B., Jurklies B. und Weber, B. H. F. (1997) Positional cloning of the gene associated with X-linked juvenile retinoschisis. *Nature Genet.* **17**: 164-170.

Schiller P. H. (1982) Central connections of the retinal ON and OFF pathways. *Nature* **297**: 580-583.

Schiller P. H. (1984) The connections of the retinal on and off pathways to the lateral geniculate nucleus of the monkey. *Vision Res.* **24**: 923-932.

Schoneberg T., Schulz A., Grosse R., Schade R., Henklein P., Schultz G. und Gudermann T. (1999) A novel subgroup of class I G-protein-coupled receptors. *Biochim. Biophys. Acta* **1446**: 57-70.

Schwahn U., Lenzner S., Dong J., Feil S., Hinzmann B., Duijnhoven G. v., Kirschner R., Hemberger M., Bergen A. A., Rosenberg T., Pinckers A. J. L. G., Fundele R., Rosenthal

A., Cremers F. P. M., Ropers H.-H. und Berger W. (1998) Positional cloning of the gene for retinitis pigmentosa 2. *Nature Genet*. **19**: 327-332.

Schwahn U., Paland N., Techritz S., Lenzner S. und Berger W. (2001) Mutations in the X-linked *RP2* gene cause intracellular misrouting and loss of the protein. *Hum. Mol. Gen*. **10**: 1177-1183.

Schwartz P. D., Zhang Z., Franzer K. A., Smit A., Riemer C., Bouck J., Gibbs R., Hardison R. und Miller W. (2000) PipMaker – a web server for aligning two genomic DNA sequences. *Nucleic Acids Res*. **23**: 1087-1088.

Sheffield V. C., Beck J. S., Kwitek A. E., Sandstrom D. W. und Stone E. M. (1993) The sensitivity of single-strand conformation polymorphism analysis for the detection of single base substitutions. *Genomics.* **16**: 325-332.

Shinomura T. und Kimata K. (1992). Proteoglycan-Lb, a small dermatan sulfate proteoglycan expressed in embryonic chick epiphyseal cartilage, is structurally related to osteoinductive factor. *J. Biol. Chem*. **267**: 1265-1270.

Shizuya H., Birren B., Kim U.-J., Mancino V., Slepak T., Tachiiri Y., Simon M. (1992) Cloning and stable maintenance of 300-kilobase-pair fragments of human DNA in *Escherichia coli* using an F-factor-based vector. *Proc. Natl. Acad. Sci. USA* **89**: 8794–8797.

Simon K. und Ikonen E. (1997) Functional rafts in membranes. *Nature*. **387**: 569-570.

Sohocki M. M., Bowne S. J., Sullivan L. S., Blackshaw S., Cepko C. L., Payne A. M., Bhattacharya S. S., Khaliq S., Mehdi S. Q., Birch D. G., Harrison W. R., Elder F. F. B., Heckenlively J. R. und Daiger S. P. (2000) Mutations in a new photoreceptor-pineal gene on 17p cause Leber congenital amaurosis. *Nature Genet.* **24**: 79-83.

Sohocki M. M., Daiger S. P., Bowne S. J., Rodriquez J. A., Northrup H., Heckenlively J. R., Birch D. G., Mintz-Hittner H., Ruiz R. S., Lewis R. A., Saperstein D. A. und Sullivan L. S. (2001) Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. *Hum. Mutat.* **17**: 42-51.

Sommarin Y., Wendel M., Shen Z., Hellman U. und Heinegard D. (1998) Osteoadherin, a cell-binding keratan sulfate proteoglycan in bone, belongs to the family of leucine-rich repeat proteins of the extracellular matrix. *J. Biol. Chem*. **273**: 16723-16729.

Southern E. M., Mir K., und Shchepinov M. (1999) Molecular interactions on microarrays. *Nature Genet.* **21**: 5-9.

Southern J. A., Young D. F., Heaney F. und Baumgartner W. (1991) Identification of an epitope on the P and V proteins of simian virus 5 that distinguishes between two isolates with different biological characteristics. *J. Gen. Virol*. **72**: 1551-1557.

Stargardt K. (1909) Über familiare, progressive Degeneration in der Makulagegend des Auges. *Albrecht von Graefes Arch. Klin. Exp. Ophthal.* **71**: 534-549.

Stevenson D., Laverty H. G., Wenwieser S., Douglas M. und Wilson J. B. (2000). Mapping and expression analysis of the human *CASK* gene. *Mammalian Genome* **11**: 934-937.

Strom T. M., Nyakatura G., Apfelstedt-Sylla E., Hellebrand H., Lorenz B., Weber B. H., Wutz K., Gutwillinger N., Ruther K., Drescher B., Sauer C., Zrenner E., Meitinger T., Rosenthal A. und Meindl A. (1998) An L-type channel gene is mutated in incomplete X-linked congenital stationary night blindness. *Nature Genet*. **19**: 260-263.

Stryer L. (1991) Visual excitation and recovery. *J. Biol. Chem.* **288**: 10711-10714.

Stulnig R., Berger M., Sigmund T., Stockinger H., Horejsi V. und Waldhäusl W. (1997) Signal transduction via glycosyl phosphatidylinositol-anchored proteins in T cells is inhibited by lowering cellular cholesterol. *J. Biol. Chem.* **272**: 19232-19247.

Suzuki Y., Orita M., Shiraishi M., Hayashi K. und Sekiya T. (1990) Detection of *ras* gene mutations in human lung cancers by single-strand conformation polymorphism analysis of polymerase chain reaction products. *Oncogene.* **5**: 1037-1043.

Toyoda, J. (1973). Membrane resistance changes underlying the bipolar cell response in the carp retina. *Vision Res*. **13**: 283-294.

Vardi N., Morigiwa K., Wang T.-L., Shi Y.-Y. und Sterling P. (1998) Neurochemistry of the mammalian cone synaptic complex. *Vis. Res.* **38**: 1359-1369.

Varma R. und Mayor S. (1998) GPI-anchored proteins are organized in submicron domains at the cell surfac. *Nature*. **394**: 798-801.

Vervoot R., Lennon A., Bird A. C., Tulloch B., Axton R., Milano M. G., Meindl A., Meitinger T., Ciccodicola A. und Wright A. F. (2000) Mutational hot spot within a new *RPGR* exon in X-linked retinitis pigmentosa. *Nature Genet*. **25**: 462-466.

Voth B. R., Kell B. L., Phalgun B. J., Ivens A. C. und McMaster W. R. (1998). Differntially expressed Leishmania major gp63 genes encode cell surface leishmanolysin with distinct signals forglycosylphosphatidylinositolattachment. *Mol. Biochem. Parasitol.* **93**: 31-41.

Walsh F. B. (1957) Clinical Neuro-ophthalmology. Baltimore: Williams and Wilkins (pub.) (2nd ed.). pp. 673-674.

Wells J., Wroblewski J., Keen J., Inglehearn C., Jubb C., Eckstein A., Jay M., Arden G., Bhattacharya S., Fitzke F. und Bird A. (1993) Mutations in the human retinal degeneration slow (*RDS*) gene can cause either retinitis pigmentosa or macular dystrophy. *Nature Genet.* **3**: 212-218.

White T. (1940) Linkage and crossing-over in the human sex chromosomes. *J. Genet*. **40**: 403-437.

Williams J. C., Case-Green S. C., Mir K. U. und Southern E. M. (1994) Studies of oligonucleotide interactions by hybridisation to arrays: the influence of dangling ends on duplex yield. *Nucleic Acids Res.* **22**: 1365-1357.

Wutz K., Sauer C., Zrenner E., Lorenz B., Alitalo T., Broghammer M., Hergersberg M., de La Chapelle A., Weber B. H. F., Wissinger A., Meindl A. und Pusch C. M. (2002) Thirty distinct *CACNA1F* mutations in 33 families with incomplete type of XLCSNB and *Cacna1f* expression profiling in mouse retina. *Europ. J. Hum. Genet.* **10**: 449-456.

Yamamoto S., Sippel K. C., Berson E. L. und Dryja T. P. (1997) Defects in the rhodopsin kinase gene in the Oguchi form of stationary night blindness. *Nature Genet.* **15**: 175-178.

Yang X. W., Model P. und Heintz N. (1997) Homologous recombination based modification in Escherichia coli and germline transmission in transgenic mice of a bacterial artifiical chromosome. *Nature Biotechnol*. **15**: 859-865.

Yau K. W. (1994) Photransduction mechanism in retinal rods and cones. The Friedenwald Lecture. *Invest. Ophthalmol. Vis. Sci.* **35**: 9-32.

Zeitz C., Scherthan H., Freier S., Feil S., Suckow V., Schweiger S. und Berger W. (2003) Functional characterization of protein motifs of NYX (nyctalopin on chromosome X) in mouse and man. *Invest Ophthalmol Vis Sci.* **44**: 4184-4191.

Zito I., Allen L. E., Patel R. J., Meindl A., Bradshaw K., Yates J. R., Bird A. C., Erksine L., Cheetham M E., Webster A. R., Poopalasundaram S., Moore A. T., Trump D. und Hardcastle A. J. (2003) Mutations in the *CACNA1F* and *NYX* Genes in British CSNBX families. *Hum Mut.* Online **#577**: 1-6.