

Publications Wolfgang Berger (peer reviewed, as of July 2022)

2022

Atac* D, Mohn* L, Feil S, Maggi K, Haenni D, Seebauer B, Koller S, Berger W (2022) Functional characterization of an in-frame deletion in the basic domain of the retinal transcription factor ATOH7. *Int J Mol Sci* 23:1053

Bryant D, Pauzuolyte V, Ingham NJ, Patel A, Pagarkar W, Anderson L, Smith KE, Moulding D, Leong YC, Jafree D, Long D, Al-Yassin A, Steel KP, Jagger DJ, Forge A, Berger W, Sowden JC, Bitner-Glindzicz M (2022) The timing of auditory sensory deficits in Norrie disease has implications for therapeutic intervention. *J Clin Invest Insight* 7:e148586

Kivrak Pfiffner F, Koller S, Ménétrey A, Graf U, Bähr L, Maspoli A, Hackenberg A, Kottke R, Gerth-Kahlert C, Berger W (2022) Homozygosity for a novel DOCK7 variant due to segmental uniparental isodisomy of chromosome 1 associated with early infantile epileptic encephalopathy (EIEE) and cortical visual impairment. *Int J Mol Sci* 23:7382

Neubauer J, Kissel CK, Billiger SA, Barbon D, Thali MJ, Clobber D, Bode PK, Kovacs B, Graf U, Maspoli A, Berger W, Sager AM, Haas C (2022) Benefits and outcomes of a new multidisciplinary approach for the management and financing of sudden unexplained death cases in a forensic setting in Switzerland. *Forensic Sci Int* 334:111240

Peters F, Ebner LJA, Atac D, Maggi J, Berger W, den Hollander AJ, Grimm C (2022) Regulation of ABCA1 by AMD-associated genetic variants and hypoxia in iPSC-RPE. *Int J Mol Sci* 23:31

*equal contribution; shared last authorship

2021

Costa S, Medeiros-Domingo A, Gasperetti A, Akdis D, Berger W, James CA, Ruschitzka F, Brunckhorst CB, Duru F, Saguner AM (2021) Impact of genetic variant reassessment on the diagnosis of arrhythmogenic right ventricular cardiomyopathy based on the 2010 task force criteria. *Circ Genom Precis Med* 14:e003047

Haug P, Koller S, Maggi J, Lang E, Silke Feil S, Włodarczyk A, Bähr L, Steindl K, Rohrbach M, *Gerth-Kahlert C, *Berger W (2021) Whole exome sequencing in coloboma/microphthalmia: Identification of novel and recurrent variants in seven genes. *Genes* 12:65

Maggi J, Koller S, Baehr L, Feil S, Kivrak Pfiffner F, Hanson JVM, Maspoli A, Gerth-Kahlert C, Berger W (2021) Long-range PCR-based NGS applications to diagnose Mendelian retinal diseases. *Int J Mol Sci* 22:1508

Rechsteiner D, Issler L, Koller S, Lang E, Bähr L, Feil S, Rüeggger C, Kottke R, Toelle S, Zweifel N, Steindl K, Josef P, Zweier M, Suter AA, Gogoll L, Haas C, *Berger W, *Gerth-Kahlert C (2021) Genetic analysis in a Swiss cohort of bilateral congenital cataract. *JAMA Ophthalmol* 139:691-700

2020

Atac D, Koller S, Hanson JVM, Feil S, Tiwari A, Bahr A, Baehr L, Magyar I, Kottke R, Gerth-Kahlert C, Berger W (2020) Atonal homolog 7 (ATOH7) loss-of-function mutations in predominant bilateral optic nerve hypoplasia. *Hum Mol Genet* 29:132-148

Kandaswamy DK, Prakash MVS, Graw J, Koller S, Magyar I, Tiwari A, Berger W, Santhiya ST (2020) Application of WES towards molecular investigation of congenital cataracts: Identification of novel alleles and genes in a hospital-based cohort of south India. *Int J Mol Sci* 21:9569

Klee K, Storti F, Maggi J, Todorova V, Karademir D, Berger W, Samardzija M, Grimm C (2020) The expression of DEPP is controlled by three distal consensus HRE elements in hypoxic retinal epithelial cells. *Genes* 11:111

Lang E, Koller S, Atac D, Pfäffli OA, Hanson JVM, Feil S, Bähr L, Bahr A, Josef P, Fasler K, Barthelmes D, Steindl K, Konrad D, Wille DA, *Berger W, *Gerth-Kahlert C (2020) Genotype-phenotype spectrum in isolated and syndromic nanophthalmos. *Acta Ophthalmol* 99:e594-e607

Lang E, Koller S, Bähr L, Töteberg-Harms M, Atac D, Roulez F, Bahr A, Steindl K, Feil F, *Berger W, *Gerth-Kahlert C (2020) Exome sequencing in a Swiss childhood glaucoma cohort reveals CYP1B1 and FOXC1 variants as most frequent causes. *Transl Vis Sci Technol* 9:47

Maggi J, Roberts L, Koller S, Rebello G, *Berger W, *Ramesar R (2020) De novo assembly-based analysis of RPGR exon ORF15 in an indigenous African cohort overcomes limitations of a standard next-generation sequencing (NGS) data analysis pipeline. *Genes* 11:800

Ulv Larsen SM, Landolt HP, Berger W, Nedergaard M, Knudsen GM, Holst SC (2020) Haplotype of the astrocytic water channel AQP4 is associated with slow wave energy regulation in human NREM sleep. *PLoS Biol* 18:e3000623

2019

Gerth-Kahlert C, Koller S, Hanson JVM, Baehr L, Tiwari A, Kivrak Pfiffner F, Bahr A, Berger W (2019) Genotype - phenotype analysis of a novel recessive and a recurrent dominant SNRNP200 variant causing retinitis pigmentosa. *Invest Ophthalmol Vis Sci* 60:2822-2835

Knöpfel EB, Vilches C, Camargo SMR, Errasti-Murugarren E, Stäubli A, Mayayo C, Munier FL, Miroshnikova N, Poncet N, Junza A, Bhattacharya SS, Prat E, Berry V, Berger W, Heon E, Moore AT, Yanes Ó, Nunes V, Palacin M, Verrey F, Kloeckener-Gruissem B (2019) Dysfunctional LAT2 amino acid transporter is associated with cataract in mouse and humans. *Front Physiol* 10:688

Park H, Yamamoto H, Mohn L, Ambühl L, Kanai K, Schmidt I, Kim KP, Fraccaroli A, Feil S, Junge HJ, Montanez E, Berger W, Adams RH (2019) Integrin-linked kinase controls retinal angiogenesis and is linked to Wnt signaling and exudative vitreoretinopathy. *Nat Commun* 10:5243

2018

Beck SC, Karlstetter M, Garcia Garrido M, Feng Y, Dannhausen K, Mühlfriedel R, Sohlingam V, Seebauer B, Berger W, Hammes HP, Seeliger MW, Langmann T (2018) Cystoid edema, neovascularization and inflammatory processes in the murine Norrin-deficient retina. *Sci Rep* 8:5970

Gerth-Kahlert C, Maggi J, Toteberg-Harms M, Tiwari A, Budde B, Nürnberg P, Koller S, Berger W (2018) Absence of goniodysgenesis in patients with chromosome 13q microdeletion-related microcoria. *Ophthalmol Glaucoma* 1:145-147

Gerth-Kahlert C, Tiwari A, Hauri-Hohl MM, Hanson JVM, Bahr A, Palmowski-Wolfe A, Güngör T, Berger W (2018) Unusual retinopathy in a child with severe combined immune deficiency. *Ophthalmic Genet* 39:92-94

Neubauer J, Lecca MR, Russo G, Bartsch C, Medeiros-Domingo A, Berger W, Haas C (2018) Exome analysis in 34 sudden unexplained death (SUD) victims mainly identified variants in channelopathy-associated genes. *Int J Legal Med* 132:1057-1065

Valomon A, Holst SC, Borrello A, Weigend S, Müller T, Berger W, Sommerauer M, Baumann CR, Landolt HP (2018) Effects of COMT genotype and tolcapone on lapses of sustained attention after sleep deprivation in healthy young men. *Neuropsychopharmacology* 43:1599-1607

2017

Beck S, Feng Y, Sohlingam V, Garcia Garrido M, Tanimoto N, Acar N, Seebauer B, Berger W, Hammes HP, Seeliger M (2017) Long-term consequences of developmental vascular defects on retinal vessel homeostasis and function in a mouse model of Norrie disease. *PLoS ONE* 12:e0178753

Gerth-Kahlert C, Tiwari A, Hanson JVM, Batmanabane V, Traboulsi E, Pennesi ME, Al-Qahtani AA, Lam BL, Heckenlively J, Zweifel SA, Vincent A, Fierz F, Barthelmes D, Branham K, Khan N, Bahr A, Baehr L, Magyar I, Koller S, Azzarello-Burri S, Niedrist D, Heon E, Berger W (2017) C2orf71 mutations as a frequent cause of autosomal-recessive retinitis pigmentosa: clinical analysis and presentation of 8 novel mutations. *Invest Ophthalmol Vis Sci* 58:3840-3850

Gerth-Kahlert G, Tiwari A, Hauri M, Hanson JVM, Bahr A, Palmowski-Wolfe A, Güngör T, Berger W (2017) Unusual retinopathy in a child with severe combined immuno deficiency. *Ophthalmic Genet* 39:92-94

Holst S, Müller T, Valomon A, Seebauer B, Berger W, Landolt HP (2017) Functional polymorphisms in dopaminergic genes modulate neurobehavioral and neurophysiological consequences of sleep deprivation. *Sci Rep* 7:45982

Medeiros-Domingo A, Saguner AM, Magyar I, Duru F, Bahr A, Akdis D, Brunckhorst C, Berger W (2017) Arrhythmogenic right ventricular cardiomyopathy versus dilated cardiomyopathy: Implications of next generation sequencing in appropriate diagnosis. *Europace* 19:1063-1069

Neubauer J, Lecca MR, Russo G, Bartsch C, Medeiros-Domingo A, Berger W, Haas C (2017) Post-mortem whole exome analysis in a large sudden infant death syndrome cohort with a focus on cardiovascular and metabolic genetic diseases. *Eur J Hum Genet* 25:404-409

Stäubli A, Fuhrer Y, Munier F, Schorderet D, Tiwari A, Verrey F, Heon E, Cheng CY, Wong TY, Berger W, Camargo S, Kloeckener-Gruissem B (2017) Abnormal creatine transport of mutations in monocarboxylate transporter 12 (MCT12) found in patients with age-related cataract can be partially rescued by exogenous chaperone CD147. *Hum Mol Genet* 26:4203-4214

Urry E, Jetter A, Holst SC, Berger W, Spinas GA, Langhans W, Landolt HP (2017) A case-control field study on the relationships among type 2 diabetes, sleepiness and habitual caffeine intake. *J Psychopharmacol.* 31:233-242

2016

Di Donato N, Neuhann T, Kahlert AK, Klink B, Hackmann K, Neuhann I, Novotna B, Schallner J, Krause C, Glass IA, Parnell SE, Benet-Pages A, Nissen AM, Berger W, Altmüller J, Thiele H, Weber BH, Schrock E, Dobyns WB, Bier A, Rump A (2016) Mutations in EXOSC2 are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. *J Med Genet* 53:419-425

Kloeckener-Gruissem B, Dours-Zimmermann MT, Skosyrski S, Brunner S, Mjaatvedt CH, Zimmermann DR, Rüther K, Berger W (2016) A potential mouse model for the erosive vitreoretinopathy of Wagner disease. *Matters* 20160500004

Neubauer J, Haas C, Bartsch C, Domingo-Medeiros A, Berger W (2016) Post-mortem whole-exome sequencing (WES) with a focus on cardiac disease-associated genes in five young sudden unexplained death (SUD) cases. *Int J Legal Med* 130:1011-1021

Tiwari A, Bahr, Baehr L, Fleischhauer J, Zinkernagel M, Winkler N, Barthelmes D, Berger L, Gerth-Kahlert C, Neidhardt J, Berger W (2016) Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies. *Sci Rep* 6:28755

Tiwari A., Lemke J, Altmueller J, Thiele H, Glaus E, Fleischhauer J, Nürnberg P, Neidhardt J, Berger W (2016) Identification of novel and recurrent disease-causing mutations in retinal dystrophies using whole exome sequencing (WES): Benefits and limitations. *PLoS ONE* 11:e0158692

Vincent A, Ng J, Gerth-Kahlert C, Tavares E, Maynes JT, Wright T, Tiwari A, Tumber A, Li S, Hanson JV, Bahr A, MacDonald H, Bähr L, Westall C, Berger W, Cremers FP, den Hollander AJ, Héon E (2016) Biallelic mutations in CRB1 underlie autosomal recessive familial foveal retinoschisis. *Invest Ophthalmol Vis Sci* 57:2637-2646

2015

Da Costa R, Glaus E, Tiwari A, Kloeckener-Gruissem B, Berger W, Neidhardt J (2015) Localizing the RPGR protein along the cilium: a new method to determine efficacies to treat RPGR mutations. *Gene Ther* 22:413-420

Gerth-Kahlert C, Seebauer B, Dold S, Hanson JVM, Wildberger H, Spörri A, van Waes H, Berger W (2015) Intra-familial phenotype variability in patients with Jalili syndrome. *Eye* 29:712-716

2014

Glöckle N, Kohl S, Mohr J, Scheurenbrand T, Sprecher A, Weisschuh N, Bernd A, Rudolph G, Schubach M, Poloscheck C, Zrenner E, Biskup S, Berger W, Wissinger B, Neidhardt J (2014) Panel-based next generation

sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. *Eur J Hum Genet* 22:99-104

Haghghi A, Tiwari A, Piri N, Nurnberg G, Saleh-Gohari N, Haghghi A, Neidhardt J, Nurnberg P, Berger W (2014) Homozygosity mapping and whole exome sequencing reveal a novel homozygous COL18A1 mutation causing Knobloch syndrome. *PLoS ONE* 9:e112747

Holst SC, Bersagliere A, Bachmann V, Berger W, Achermann P, Landolt HP (2014) Dopaminergic role in regulating neurophysiological markers of sleep homeostasis in humans. *J Neurosci* 8:566-573

Oczos J, Sutter I, Kloeckener-Gruissem B, Berger W, Riwanto M, Rentsch K, Hornemann T, von Eckardstein A, Grimm C (2014) Lack of paraoxonase 1 alters phospholipid composition, but not morphology and function of the mouse retina. *Invest Ophthalmol Vis Sci* 55:4714-4727

Valomon A, Holst SC, Bachmann V, Viola AU, Schmidt C, Zürcher J, Berger W, Cajochen C, Landolt HP (2014) Genetic polymorphisms of DAT1 and COMT differentially associate with actigraphy-derived sleep-wake cycles in young adults. *Chronobiol Int* 31:705-714

2013

Abplanalp J, Laczkó E, Philp NJ, Neidhardt J, Zuercher J, Braun P, Schorderet DF, Munier FL, Verrey F, Berger W, Camargo SMR, Kloeckener-Gruissem B (2013) The cataract and glucosuria associated monocarboxylate transporter MCT12 is a new creatine transporter. *Hum Mol Genet* 22:3218-26

Bukowy-Bieryło Z, Ziętkiewicz E, Loges NT, Wittmer M, Geremek M, Olbrich H, Fliegauf M, Voelkel K, Rutkiewicz E, Rutland J, Morgan LC, Pogorzelski A, Martin J, Haan EA, Berger W, Omran H, Witt M (2013) RPGR mutations might cause reduced orientation of respiratory cilia. *Pediatr Pulmonol* 48:352-363

Collin RWJ, Nikopoulos K, Dona M, Gilissen C, Hoischen A, Boonstra FN, Poulter JA, Kondo H, Berger W, Toomes C, Tahira T, Mohn LR, Blokland EA, Hetterschijt L, Ali M, Groothuisink J, Inglehearn CF, Sollfrank L, Strom TM, Uchio E, van Nouhuys CE, Kremer H, Veltman JA, van Wijk EAR, Cremers FPM (2013) ZNF408 is mutated in familial exudative vitreoretinopathy and crucial for the development of zebrafish retinal vasculature. *PNAS* 110:9856-9861

Kloeckener-Gruissem B, Neidhardt J, Magyar I, Plauchu H, Zech JC, Morlé L, Palmer-Smith SM, MacDonald MJ, Nas V, Fry AE, Berger W (2013) Novel VCAN mutations and evidence for unbalanced alternative splicing in the pathogenesis of Wagner syndrome. *Eur J Hum Genet* 21:352-356

Oczos J, Grimm C, Barthelmes D, Sutter F, Menghini M, Kloeckener-Gruissem B, Berger W. (2013) Regulatory regions of the paraoxonase 1 (PON1) gene are associated with neovascular age-related macular degeneration (AMD). *Age (Dordr)* 35:1651-1662

Schmid F, Hiller T, Korner G, Glaus E, Berger W, Neidhardt J (2013) A gene therapeutic approach to correct splice defects with modified U1 and U6 snRNPs. *Hum Gene Ther* 24:97-104

Thun GA, Imboden M, Berger W, Rochat T, Probst-Hensch NM (2013) The association of a variant in the cell cycle control gene CCND1 and obesity on the development of asthma in the Swiss SAPALDIA study. *J Asthma* 50:147-154

2012

Audo I, Bujakowska K, Orhan E, Poloschek CM, Defoort-Dhellemmes S, Drumare I, Kohl S, Luu TD, Lecompte O, Zrenner E, Lancelot ME, Antonio A, Germain A, Michiels C, Audier C, Letexier M, Saraiva JP, Leroy BP, Munier FL, Mohand-Saïd S, Lorenz B, Friedburg C, Preising M, Kellner U, Renner AB, Moskova-Doumanova V, Berger W, Wissinger B, Hamel CP, Schorderet DF, De Baere E, Sharon D, Banin E, Jacobson SG, Bonneau D, Zanlonghi X, Le Meur G, Casteels I, Koenekoop R, Long VW, Meire F, Prescott K, de Ravel T, Simmons I, Nguyen H, Dollfus H, Poch O, Léveillard T, Nguyen-Ba-Charvet K, Sahel JA, Bhattacharya SS, Zeitz C (2012) Whole-exome sequencing identifies mutations in GPR179 leading to autosomal-recessive complete congenital stationary night blindness. *Am J Hum Genet* 90:321-330

Bachmann V, Klaus F, Bodenmann S, Schäfer N, Brugger P, Huber S, Berger W, Landolt HP (2011) Functional ADA polymorphism increases sleep depth and reduces vigilant attention in humans. *Cereb Cortex* 22:962-970

Bachmann V, Klein C, Bodenmann S, Schäfer N, Berger W, Brugger P, Landolt HP (2012) The *BDNF* Val66Met polymorphism modulates sleep intensity: EEG frequency- and state-specificity. *Sleep* 35:335-344

Menghini M, Kloeckener-Gruissem B, Schindler C, Fleischhauer J, Kurz-Levin MM, Sutter FKP, Berger W, Barthelmes D (2012) Impact of loading phase, initial response and CFH genotype on the long-term outcome of treatment for neovascular age-related macular degeneration using ranibizumab. *PLoS One* 7:e42014

Zuercher J, Fritzsche M, Feil S, Mohn L, Berger W (2012) Norrin stimulates cell proliferation in the superficial retinal vascular plexus and is pivotal for the recruitment of mural cell. *Hum Mol Genet* 21:2619-2630

Zweifel SA, Maygar I, Berger W, Tschaud P, Becker M, Michels S (2012) Multimodal imaging of autosomal dominant drusen. *Klin Monbl Augenheilkd* 229:399-402

2011

Curjuric I, Zemp E, Dratva J, Ackermann-Liebrich U, Bridevaux PO, Bettschart RW, Brutsche M, Frey M, Gerbase MW, Knöpfli B, Künzli N, Pons M, Schwartz J, Schindler C, Rochat T; SAPALDIA team (2011) Determinants of change in airway reactivity over 11 years in the SAPALDIA population study. *Eur Respir J* 37:492-500

Gerbase MW, Keidel D, Imboden M, Gemperli A, Bircher A, Schmid-Grendelmeier P, Bridevaux PO, Berger W, Schindler C, Rochat T, Probst-Hensch N (2011) Effect modification of IgE-mediated atopy and rhinitis by GST genotypes in passive smokers. *Clin Exp Allergy* 41:1579-86

Glaus E, Schmid F, Da Costa R, Berger W, Neidhardt J (2011) Gene therapeutic approach using mutation-adapted U1 snRNA to correct a *RPGR* splice defect in patient-derived cells. *Mol Ther* 19:936-941

Kloeckener-Gruissem B, Barthelmes D, Labs S, Schindler C, Kurz-Levin M, Michels S, Fleischhauer J, Berger W, Sutter F, Menghini M (2011) Genetic association with response to intravitreal ranibizumab in patients with neovascular AMD. *Invest Ophthalmol Vis Sci* 52:4694-4702

Schmid F, Glaus E, Barthelmes D, Fliegauf M, Gaspar H, Nürnberg G, Nürnberg P, Omran H, Berger W, Neidhardt J (2011) U1 snRNA-mediated gene therapeutic correction of splice defects caused by an exceptionally mild BBS mutation. *Hum Mutat* 32:815-824

2010

Berger W, Kloeckener-Gruissem B, Neidhardt J (2010) The molecular basis of human retinal and vitreoretinal diseases. *Prog Retin Eye Res* 29:335-375

Brunner S, Skosyrski S, Kirschner-Schwabe R, Knobeloch KP, Neidhardt J, Feil S, Luhmann UFO, Rüther K, Berger W (2010) Cone versus rod disease in a mutant *Rpgr* mouse caused by different genetic backgrounds. *Invest Ophthalmol Vis Sci* 51:1106-1115

Curjuric I, Imboden M, Schindler C, Downs SH, Hersberger M, Liu SLJ, Mátyás G, Russi EW, Schwartz J, Thun GA, Postma DS, Rochat T, Probst-Hensch NM, and the SAPALDIA team (2010) HMOX1 and GST variants modify attenuation of FEF25-75-decline due to PM10 reduction. *Eur Respir J* 35:505-514

Hersberger M, Thun GA, Imboden M, Brandstätter A, Waechter V, Summerer M, Schmid-Grendelmeier P, Bircher A, Rohrer L, Berger W, Russi EW, Rochat T, Kronenberg F, Probst-Hensch N (2010) Association of STR polymorphisms in CMA1 and IL-4 with asthma and atopy: the SAPALDIA cohort. *Hum Immunol* 71:1154-1160

Poloschek CM, Bach M, Lagrèze WA, Glaus E, Lemke JR, Berger W, Neidhardt J (2010) ABCA4 and ROM1: Implications for modification of the *PRPH2*-associated macular dystrophy phenotype. *Invest Ophthalmol Vis Sci* 51:4253-4265

Schmid F, Glaus E, Cremers FPM, Kloeckener-Gruissem B, Berger W, Neidhardt J (2010) Mutation- and tissue-specific alterations of *RPGR* transcripts. *Invest Ophthalmol Vis Sci* 51:1628-1635

Zuercher J, Neidhardt J, Magyar I, Labs S, Moore AT, Tanner FC, Waseem N, Schorderet DF, Munier FL, Bhattacharya S, Berger W, Kloeckener-Gruissem B (2010) Alterations of the 5'untranslated leader region of *SLC16A12* lead to age-related cataract. *Invest Ophthalmol Vis Sci* 51:3354-3361

2009

Audo I, Kohl S, Leroy BP, Munier FL, Guillonneau X, Mohand-Saïd S, Bujakowska K, Nandrot E, Lorenz B, Preising M, Kellner U, Renner A, Bernd A, Antonio A, Moskova-Doumanova V, Lancelot ME, Poloschek CM, Drumare I, Defoort-Dhellemmes S, Wissinger B, Léveillard T, Hamel CP, Schorderet DF, De Baere E, Berger W, Jacobson SG, Zrenner E, Sahel JA, Bhattacharya SS, Zeitz C (2009) *TRPM1* is mutated in patients with complete autosomal recessive congenital stationary night blindness. ***Am J Hum Genet*** 85:720-729

Bodenmann S, Xu S, Luhmann UFO, Arand M, Berger W, Jung H, Landolt HP (2009) Pharmacogenetics of modafinil after sleep loss: Catechol-O-methyltransferase genotype modulates waking functions but not recovery sleep. ***Clin Pharmacol Ther*** 85:296-304

Castro-Giner F, Kogevinas M, Imboden M, de Cid R, Jarvis D, Mächler M, Berger W, Burney P, Franklin KA, Gonzalez JR, Heinrich J, Janson C, Omenaas E, Pin I, Rochat T, Sunyer J, Wjst M, Antó JM, Estivill X, Probst-Hensch NM (2009) Joint effect of obesity and TNFA variability on asthma: two international cohort studies. ***Eur Respir J*** 33:1003-1009

Imboden M, Schwartz J, Schindler C, Curjuric I, Berger W, Liu SL, Russi EW, Ackermann-Liebrich U, Rochat T, Probst-Hensch NM (2009) Decreased PM10 exposure attenuates age-related lung function decline: genetic variants in p53, p21, and CCND1 modify this effect. ***Environ Health Perspect*** 117:1420-1427

Leroy BP, Budde B, Wittmer M, De Baere E, Berger W, Zeitz C (2009) A common NYX mutation in Flemish patients with X-linked CSNB. ***Brit J Ophthalmol*** 93:692-696

Macas E, Mátyás G, Reuge P, Berger W, Imthurn B (2009) Polar body biopsy for Curschmann-Steinert disease and successful pregnancy following embryo vitrification. ***Reprod Biomed Online*** 18:815–820

Magyar I, Colman D, Arnold E, Baumgartner D, Bottani A, Fokstuen S, Addor M-C, Berger W, Carrel T, Steinmann B, Mátyás G (2009) Quantitative sequence analysis of *FBN1* premature termination codons provides evidence for incomplete NMD in leukocytes. ***Hum Mutat*** 30:1355-1364

Schäfer NF, Luhmann UF, Feil S, Berger W (2009) Differential gene expression in *Ndph* knockout mice in retinal development. ***Invest Ophthalmol Vis Sci*** 50:906-916

Tanner G, Glauß E, Barthelmes D, Ader M, Fleischhauer J, Pagani F, Berger W, Neidhardt J (2009) Therapeutic strategy to rescue mutation-induced exon skipping in rhodopsin by adaptation of U1 snRNA. ***Hum Mutat*** 30:255-263

Zeitz C, Labs S, Lorenz B, Forster U, Ueksti J, Kroes HY, De Baere E, Leroy BP, Cremers FP, Wittmer M, van Genderen MM, Sahel JA, Audo I, Poloschek CM, Mohand-Said S, Fleischhauer JC, Hueffmeier U, Moskova-Doumanova V, Levin AV, Hamel CP, Leifert D, Munier FL, Schorderet D, Zrenner E, Friedburg C, Wissinger B, Kohl S, Berger W (2009) Genotyping microarray for CSNB-associated genes. ***Invest Ophthalmol Vis Sci*** 50:5919-5926

2008

Brunner S, Colman D, Travis AJ, Luhmann UF, Shi W, Feil S, Imsand C, Nelson J, Grimm C, Rülicke T, Fundele R, Neidhardt J, Berger W (2008) Overexpression of RPGR leads to male infertility in mice due to defects in flagellar assembly. ***Biol Reprod*** 79:608-617

Castro-Giner F, Kogevinas M, Machler M, de Cid R, Steen KV, Imboden M, Schindler C, Berger W, Gonzalez JR, Franklin KA, Janson C, Jarvis D, Omenaas E, Burney P, Rochat T, Estivill X, Antó JM, Wjst M, Probst-Hensch NM (2008) TNFA-308 in two international population-based cohorts shows increased risk for asthma. ***Eur Respir J*** 32:350-361

Fröhlich BA, Zeitz C, Mátyás G, Alkadhi H, Tuor C, Berger W, Russi WE (2008) Novel mutations in the folliculin (*FLCN*) gene associated with spontaneous pneumothorax. ***Eur Respir J*** 32:1316-1320

Imboden M, Rochat T, Brutsche M, Schindler C, Downs SH, Gerbase MW, Berger W, Probst-Hensch NM (2008) Glutathione S-transferase genotype increases risk of progression from bronchial hyperresponsiveness to asthma in adults. *Thorax* 63:322-328

Kloeckener-Gruissem B, Vandekerckhove K, Nurnberg G, Neidhardt J, Zeitz C, Nurnberg P, Schipper I, Berger W (2008) Mutation of the solute carrier SLC16A12 associates with a syndrome combining autosomal dominant juvenile cataract with microcornea and glucosuria. *Am J Hum Genet* 82:772-779

Luhmann UF, Neidhardt J, Kloeckener-Gruissem B, Schaefer NF, Glaus E, Feil S, Berger W (2008) Vascular changes in the cerebellum of Norrin/*Ndph* knockout mice correlate with high expression of Norrin and Frizzled-4. *Eur J Neurosci* 27:2619-2628

Neidhardt J, Glaus E, Lorenz B, Netzer C, Li Y, Schambeck M, Wittmer M, Feil S, Kirschner-Schwabe R, Rosenberg T, Cremers FPM, Bergen AAB, Barthelmes D, Baraki H, Schmid F, Tanner G, Fleischhauer J, Orth U, Becker C, Wegscheider E, Nurnberg G, Nurnberg P, Bolz HJ, Gal A, Berger W (2008) Identification of novel mutations in X-linked retinitis pigmentosa families and implications for diagnostic testing. *Mol Vis* 14:1081-1093

Poloschek CM, Kloeckener-Gruissem B, Hansen LL, Bach M, Berger W (2008) Syndromic choroideremia - sublocalization of phenotypes associated with Martin-Probst deafness mental retardation syndrome (MPDMRS). *Invest Ophthalmol Vis Sci* 49:4096-4104

Probst-Hensch NM, Imboden M, Felber DD, Barthelemy JC, Ackermann-Liebrich U, Berger W, Gaspoz JM, Schwartz J (2008) Glutathione S-transferase polymorphisms, passive smoking, obesity, and heart rate variability in nonsmokers. *Environ Health Perspect* 116:1494-1499

Senn O, Russi EW, Schindler C, Imboden M, von Eckardstein A, Brandli O, Zemp E, Ackermann-Liebrich U, Berger W, Rochat T, Luisetti M, Probst-Hensch NM (2008) Circulating alpha1-antitrypsin in the general population: Determinants and association with lung function. *Respir Res* 9:35

Zeitz C, Gross AK, Leifert D, Kloeckener-Gruissem B, McAlear SD, Lemke J, Neidhardt J, Berger W (2008) Identification and functional characterization of a novel rhodopsin mutation associated with autosomal dominant CSNB. *Invest Ophthalmol Vis Sci* 49:4105-4114

Zorzetto M, Russi E, Senn O, Imboden M, Ferrarotti I, Tinelli C, Campo I, Ottaviani S, Scabini R, von Eckardstein A, Berger W, Brandli O, Rochat T, Luisetti M, Probst-Hensch N (2008) SERPINA1 gene variants in individuals from the general population with reduced alpha1-antitrypsin concentrations. *Clin Chem* 54:1331-1338

2007

Banin E, Mizrahi-Meissonnier L, Neis R, Silverstein S, Magyar I, Abeliovich D, Roepman R, Berger W, Rosenberg T, Sharon D (2007) A non-ancestral RPGR missense mutation in families with either recessive or semi-dominant X-linked retinitis pigmentosa. *Am J Med Genet A* 143:1150-1158

Cremers FP, Kimberling WJ, Kulm M, de Brouwer AP, van Wijk E, Te BH, Cremers CW, Hoefsloot LH, Banfi S, Simonelli F, Fleischhauer JC, Berger W, Kelley PM, Haralambous E, Bitner-Glindzic M, Webster AR, Saihan Z, De Baere E, Leroy BP, Silvestri G, McKay GJ, Koenekoop RK, Millan JM, Rosenberg T, Joensuu T, Sankila EM, Weil D, Weston MD, Wissinger B, Kremer H (2007) Development of a genotyping microarray for Usher syndrome. *J Med Genet* 44:153-160

Imboden M, Downs SH, Senn O, Matyas G, Brandli O, Russi EW, Schindler C, Ackermann-Liebrich U, Berger W, Probst-Hensch NM (2007) Glutathione S-transferase genotypes modify lung function decline in the general population: SAPALDIA cohort study. *Respir Res* 8:2

Matyas G, Alonso S, Patrignani A, Marti M, Arnold E, Magyar I, Henggeler C, Carrel T, Steinmann B, Berger W (2007) Large genomic fibrillin-1 (FBN1) gene deletions provide evidence for true haploinsufficiency in Marfan syndrome. *Hum Genet* 122:23-32

Neidhardt J, Glaus E, Barthelmes D, Zeitz C, Fleischhauer J, Berger W (2007) Identification and characterization of a novel RPGR isoform in human retina. *Hum Mutat* 28:797-807

Retey JV, Adam M, Khatami R, Luhmann UF, Jung HH, Berger W, Landolt HP (2007) A genetic variation in the adenosine A2A receptor gene (ADORA2A) contributes to individual sensitivity to caffeine effects on sleep. *Clin Pharmacol Ther* 81:692-698

Zeitz C, Forster U, Neidhardt J, Feil S, Kalin S, Leifert D, Flor PJ, Berger W (2007) Night blindness-associated mutations in the ligand-binding, cysteine-rich, and intracellular domains of the metabotropic glutamate receptor 6 abolish protein trafficking. *Hum Mutat* 28:771-780

2006

Bahadori R, Biehlmaier O, Zeitz C, Labhart T, Makhankov YV, Forster U, Gesemann M, Berger W, Neuhauss SC (2006) Nyctalopin is essential for synaptic transmission in the cone dominated zebrafish retina. *Eur J Neurosci* 24:1664-1674

Bartholdi D, Klein A, Weissert M, Koenig N, Baumer A, Boltshauser E, Schinzel A, Berger W, Matyas G (2006) Clinical profiles of four patients with Rett syndrome carrying a novel exon 1 mutation or genomic rearrangement in the MECP2 gene. *Clin Genet* 69:319-326

Imboden M, Nieters A, Bircher A, Brutsche M, Becker N, Wjst M, Ackermann-Liebrich U, Berger W, Probst-Hensch N (2006) Cytokine gene polymorphisms and atopic disease in two European cohorts. (ECRHS-Basel and SAPALDIA). *Clin Mol Allergy* 4:9

Imboden M, Nicod L, Nieters A, Glaus E, Matyas G, Bircher AJ, Ackermann-Liebrich U, Berger W, Probst-Hensch NM (2006) The common G-allele of interleukin-18 single-nucleotide polymorphism is a genetic risk factor for atopic asthma. The SAPALDIA Cohort Study. *Clin Exp Allergy* 36:211-218

Kloeckener-Gruissem B, Bartholdi D, Abdou MT, Zimmermann DR, Berger W (2006) Identification of the genetic defect in the original Wagner syndrome family. *Mol Vis* 12:350-355

Kurz DJ, Kloeckener-Gruissem B, Akhmedov A, Eberli FR, Buhler I, Berger W, Bertel O, Luscher TF (2006) Degenerative aortic valve stenosis, but not coronary disease, is associated with shorter telomere length in the elderly. *Arterioscler Thromb Vasc Biol* 26:e114-e117

Matyas G, Arnold E, Carrel T, Baumgartner D, Boileau C, Berger W, Steinmann B (2006) Identification and in silico analyses of novel TGFB1 and TGFB2 mutations in Marfan syndrome-related disorders. *Hum Mutat* 27:760-769

Neidhardt J, Barthelmes D, Farahmand F, Fleischhauer JC, Berger W (2006) Different amino acid substitutions at the same position in rhodopsin lead to distinct phenotypes. *Invest Ophthalmol Vis Sci* 47:1630-1635

Wollmann G, Lenzner S, Berger W, Rosenthal R, Karl MO, Strauss O (2006) Voltage-dependent ion channels in the mouse RPE: comparison with Norrie disease mice. *Vision Res* 46:688-698

Wycisk KA, Zeitz C, Feil S, Wittmer M, Forster U, Neidhardt J, Wissinger B, Zrenner E, Wilke R, Kohl S, Berger W (2006) Mutation in the auxiliary calcium-channel subunit CACNA2D4 causes autosomal recessive cone dystrophy. *Am J Hum Genet* 79:973-977

Wycisk KA, Budde B, Feil S, Skosyrski S, Buzzi F, Neidhardt J, Glaus E, Nurnberg P, Ruether K, Berger W (2006) Structural and functional abnormalities of retinal ribbon synapses due to Cacna2d4 mutation. *Invest Ophthalmol Vis Sci* 47:3523-3530

Zeitz C, Kloeckener-Gruissem B, Forster U, Kohl S, Magyar I, Wissinger B, Matyas G, Borruat FX, Schorderet DF, Zrenner E, Munier FL, Berger W (2006) Mutations in CABP4, the gene encoding the Ca²⁺-binding protein 4, cause autosomal recessive night blindness. *Am J Hum Genet* 79:657-667

2005

Hsieh M, Boerboom D, Shimada M, Lo Y, Parlow AF, Luhmann UF, Berger W, Richards JS (2005) Mice null for Frizzled4 (Fzd4^{-/-}) are infertile and exhibit impaired corpora lutea formation and function. *Biol Reprod* 73:1135-1146

Kloeckner-Gruissem B, Betts DR, Zankl A, Berger W, Gungor T (2005) A new and a reclassified ICF patient without mutations in DNMT3B and its interacting proteins SUMO-1 and UBC9. *Am J Med Genet A* 136:31-37

Luhmann UF, Meunier D, Shi W, Luttges A, Pfarrer C, Fundele R, Berger W (2005) Fetal loss in homozygous mutant Norrie disease mice: a new role of Norrin in reproduction. *Genesis* 42:253-262

Luhmann UF, Lin J, Acar N, Lammel S, Feil S, Grimm C, Seeliger MW, Hammes HP, Berger W (2005) Role of the Norrie disease pseudoglioma gene in sprouting angiogenesis during development of the retinal vasculature. *Invest Ophthalmol Vis Sci* 46:3372-3382

Ohlmann A, Scholz M, Goldwich A, Chauhan BK, Hudl K, Ohlmann AV, Zrenner E, Berger W, Cvekl A, Seeliger MW, Tamm ER (2005) Ectopic norrin induces growth of ocular capillaries and restores normal retinal angiogenesis in Norrie disease mutant mice. *J Neurosci* 25:1701-1710

Retey JV, Adam M, Honegger E, Khatami R, Luhmann UF, Jung HH, Berger W, Landolt HP (2005) A functional genetic variation of adenosine deaminase affects the duration and intensity of deep sleep in humans. *Proc Natl Acad Sci U S A* 102:15676-15681

Zeitz C, Minotti R, Feil S, Matyas G, Cremers FP, Hoyng CB, Berger W (2005) Novel mutations in CACNA1F and NYX in Dutch families with X-linked congenital stationary night blindness. *Mol Vis* 11:179-183

Zeitz C, van Genderen M, Neidhardt J, Luhmann UF, Hoeben F, Forster U, Wycisk K, Matyas G, Hoyng CB, Riemsdag F, Meire F, Cremers FP, Berger W (2005) Mutations in GRM6 cause autosomal recessive congenital stationary night blindness with a distinctive scotopic 15-Hz flicker electroretinogram. *Invest Ophthalmol Vis Sci* 46:4328-4335

2003

Pesch K, Zeitz C, Fries JE, Munscher S, Pusch CM, Kohler K, Berger W, 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

Zeitz C, Scherthan H, Freier S, Feil S, Suckow V, Schweiger S, Berger W (2003) NYX (nyctalopin on chromosome X), the gene mutated in congenital stationary night blindness, encodes a cell surface protein. *Invest Ophthalmol Vis Sci* 44:4184-4191

2002

Lenzner S, Prietz S, Feil S, Nuber UA, Ropers HH, Berger W (2002) Global gene expression analysis in a mouse model for Norrie disease: late involvement of photoreceptor cells. *Invest Ophthalmol Vis Sci* 43:2825-2833

Rehm HL, Zhang DS, Brown MC, Burgess B, Halpin C, Berger W, Morton CC, Corey DP, Chen ZY (2002) Vascular defects and sensorineural deafness in a mouse model of Norrie disease. *J Neurosci* 22:4286-4292

2001

Kirschner R, Erturk D, Zeitz C, Sahin S, Ramser J, Cremers FP, Ropers HH, Berger W (2001) DNA sequence comparison of human and mouse retinitis pigmentosa GTPase regulator (RPGR) identifies tissue-specific exons and putative regulatory elements. *Hum Genet* 109:271-278

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

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

Sudbrak R, Wieczorek G, Nuber UA, Mann W, Kirchner R, Erdogan F, Brown CJ, Wohrle D, Sterk P, Kalscheuer VM, Berger W, Lehrach H, Ropers HH (2001) X chromosome-specific cDNA arrays: identification of genes that escape from X-inactivation and other applications. *Hum Mol Genet* 10:77-83

Tonnes H, Toliat MR, Ramel C, Pape UF, Neitzel H, Berger W, Wiedenmann B (2001) Analysis of sporadic neuroendocrine tumours of the enteropancreatic system by comparative genomic hybridisation. *Gut* 48:536-541

2000

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

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

Roepman R, Bernoud-Hubac N, Schick DE, Maugeri A, Berger W, Ropers HH, Cremers FP, Ferreira PA (2000) The retinitis pigmentosa GTPase regulator (RPGR) interacts with novel transport-like proteins in the outer segments of rod photoreceptors. *Hum Mol Genet* 9:2095-2105

1999

Kimberland ML, Divoky V, Prchal J, Schwahn U, Berger W, Kazazian HH, Jr. (1999) Full-length human L1 insertions retain the capacity for high frequency retrotransposition in cultured cells. *Hum Mol Genet* 8:1557-1560

Kirschner R, Rosenberg T, Schultz-Heienbrok R, Lenzner S, Feil S, Roepman R, Cremers FP, Ropers HH, 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

Rosenberg T, Schwahn U, Feil S, Berger W (1999) Genotype-phenotype correlation in X-linked retinitis pigmentosa 2 (RP2). *Ophthalmic Genet* 20:161-172

1998

Functional implications of the spectrum of mutations found in 234 cases with X-linked juvenile retinoschisis. The Retinoschisis Consortium. *Hum Mol Genet* 7:1185-1192

Berger W (1998) Molecular dissection of Norrie disease. *Acta Anat (Basel)* 162:95-100

Richter M, Gottanka J, May CA, Welge-Lussen U, Berger W, Lutjen-Drecoll E (1998) Retinal vasculature changes in Norrie disease mice. *Invest Ophthalmol Vis Sci* 39:2450-2457

Schwahn U, Lenzner S, Dong J, Feil S, Hinzmann B, van Duijnhoven G, Kirschner R, Hemberger M, Bergen AA, Rosenberg T, Pinckers AJ, Fundele R, Rosenthal A, Cremers FP, Ropers HH, Berger W (1998) Positional cloning of the gene for X-linked retinitis pigmentosa 2. *Nat Genet* 19:327-332

Zaremba J, Feil S, Juszko J, Myga W, van Duijnhoven G, Berger W (1998) Intrafamilial variability of the ocular phenotype in a Polish family with a missense mutation (A63D) in the Norrie disease gene. *Ophthalmic Genet* 19:157-164

1997

Quaderi NA, Schweiger S, Gaudenz K, Franco B, Rugarli EI, Berger W, Feldman GJ, Volta M, Andolfi G, Gilgenkrantz S, Marion RW, Hennekam RC, Opitz JM, Muenke M, Ropers HH, Ballabio A (1997) Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. *Nat Genet* 17:285-291

Ruether K, van de PD, Jaissle G, Berger W, Tornow RP, Zrenner E (1997) Retinoschisislike alterations in the mouse eye caused by gene targeting of the Norrie disease gene. *Invest Ophthalmol Vis Sci* 38:710-718

Toliat MR, Berger W, Ropers HH, Neuhaus P, Wiedenmann B (1997) Mutations in the MEN I gene in sporadic neuroendocrine tumours of gastroenteropancreatic system. *Lancet* 350:1223

1996

Berger W, van de Pol D, Bachner D, Oerlemans F, Winkens H, Hameister H, Wieringa B, Hendriks W, Ropers HH (1996) An animal model for Norrie disease (ND): gene targeting of the mouse ND gene. *Hum Mol Genet* 5:51-59

Fuchs S, van de Pol D, Beudt U, Kellner U, Meire F, Berger W, Gal A (1996) Three novel and two recurrent mutations of the Norrie disease gene in patients with Norrie syndrome. *Hum Mutat* 8:85-88

Lenders JW, Eisenhofer G, Abeling NG, Berger W, Murphy DL, Konings CH, Wagemakers LM, Kopin IJ, Karoum F, van Gennip AH, Brunner HG (1996) Specific genetic deficiencies of the A and B isoenzymes of monoamine oxidase are characterized by distinct neurochemical and clinical phenotypes. *J Clin Invest* 97:1010-1019

Roepman R, van Duijnhoven G, Rosenberg T, Pinckers AJ, Bleeker-Wagemakers LM, Bergen AA, Post J, Beck A, Reinhardt R, Ropers HH, Cremers FP, Berger W (1996) Positional cloning of the gene for X-linked retinitis pigmentosa 3: homology with the guanine-nucleotide-exchange factor RCC1. *Hum Mol Genet* 5:1035-1041

Roepman R, Bauer D, Rosenberg T, van Duijnhoven G, van de Pol D, Platzer M, Rosenthal A, Ropers HH, Cremers FP, Berger W (1996) Identification of a gene disrupted by a microdeletion in a patient with X-linked retinitis pigmentosa (XLRP). *Hum Mol Genet* 5:827-833

Salenger PV, Hueber P, Speller PJ, van Duijnhoven G, Hoopes RR, Jr., Thakker RV, Berger W, Scheinman SJ (1996) A Pst I restriction fragment length polymorphism near the MAO locus on Xp. *Ann Hum Genet* 60:437

1995

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

1993

de Leeuw B, Berger W, Sinke RJ, Suijkerbuijk RF, Gilgenkrantz S, Geraghty MT, Valle D, Monaco AP, Lehrach H, Ropers HH, . (1993) Identification of a yeast artificial chromosome (YAC) spanning the synovial sarcoma-specific t(X;18)(p11.2;q11.2) breakpoint. *Genes Chromosomes Cancer* 6:182-189

Sinke RJ, de Leeuw B, Janssen HA, Weghuis DO, Suijkerbuijk RF, Meloni AM, Gilgenkrantz S, Berger W, Ropers HH, Sandberg AA, . (1993) Localization of X chromosome short arm markers relative to synovial sarcoma- and renal adenocarcinoma-associated translocation breakpoints. *Hum Genet* 92:305-308

1992

Berger W, van de Pol D, Warburg M, Gal A, Bleeker-Wagemakers L, de Silva H, Meindl A, Meitinger T, Cremers F, Ropers HH (1992) Mutations in the candidate gene for Norrie disease. *Hum Mol Genet* 1:461-465

Berger W, Meindl A, van de Pol TJ, Cremers FP, Ropers HH, Doerner C, Monaco A, Bergen AA, Lebo R, Warburg M, Zergollern L, Lorenz B, Gal A, Bleeker-Wagemakers EM, Meitinger T (1992) Isolation of a candidate gene for Norrie disease by positional cloning. *Nat Genet* 1:199-203

Berger W, Meindl A, de Leeuw B, de Roos A, van de Pol TJ, Meitinger T, van der Velde-Visser SD, Achatz H, Geurts vK, Cremers FP, . (1992) Generation and characterization of radiation reduced cell hybrids and isolation of probes from the proximal short arm of the human X chromosome. *Hum Genet* 90:243-246

Meindl A, Berger W, Meitinger T, van de Pol D, Achatz H, Dorner C, Haasemann M, Hellebrand H, Gal A, Cremers F, Ropers HH (1992) Norrie disease is caused by mutations in an extracellular protein resembling C-terminal globular domain of mucins. *Nat Genet* 2:139-143

1987-1990

Cremers FP, Brunsmann F, Berger W, van Kerkhoff EP, van de Pol TJ, Wieringa B, Pawlowitzki IH, Ropers HH (1990) Cloning of the breakpoints of a deletion associated with choroideremia. *Hum Genet* 86:61-64

Riess O, Michel A, Berger W, Nurnberg P, Epplen JT, Speer A, Coutelle C (1989) RFLP-discordance within the human phenylalanine hydroxylase locus. *Hum Genet* 83:199-201

Berger W, Hein J, Gedschold J, Bauer I, Speer A, Farrall M, Williamson R, Coutelle C (1987) Crossovers in two German cystic fibrosis families determine probe order for MET, 7C22 and XV-2c/CS.7. *Hum Genet* 77:197-199

Riethdorf S, Flunker G, Seidel W, Khairallah S, Berger W, Meixner M, Hecker M, Mach F, Dohner L (1987) [In vitro translation of mRNA from respiratory syncytial virus-infected cells and preparation of cDNA]. *Arch Exp Veterinarmed* 41:682-685