

Birgit Lorenz

List of Publications by Year in descending order

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149
papers

9,031
citations

61857

43
h-index

48187

88
g-index

175
all docs

175
docs citations

175
times ranked

8541
citing authors

#	ARTICLE	IF	CITATIONS
1	Worldwide outcomes of nasal transposition of the split lateral rectus muscle for strabismus associated with 3rd-nerve palsy. <i>British Journal of Ophthalmology</i> , 2023, 107, 725-731.	2.1	6
2	Artificial Intelligence for Retinopathy of Prematurity. <i>Ophthalmology</i> , 2022, 129, e69-e76.	2.5	23
3	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. <i>Human Mutation</i> , 2022, 43, 832-858.	1.1	8
4	The landscape of submicroscopic structural variants at the <i>OPN1LW/OPN1MW</i> gene cluster on Xq28 underlying blue cone monochromacy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	2
5	Current Management of Inherited Retinal Degeneration Patients in Europe: Results of a Multinational Survey by the European Vision Institute Clinical Research Network. <i>Ophthalmic Research</i> , 2021, 64, 622-638.	1.0	8
6	Current Management of Patients with <i>RPE65</i> Mutation-Associated Inherited Retinal Degenerations in Europe: Results of a Multinational Survey by the European Vision Institute Clinical Research Network. <i>Ophthalmic Research</i> , 2021, 64, 740-753.	1.0	12
7	Inherited Retinal Degenerations in the Pediatric Population. , 2021, , 183-209.		0
8	Yokoyama procedure for esotropia associated with high myopia: real-world data from a large-scale multicentre analysis. <i>Acta Ophthalmologica</i> , 2021, 99, e1340-e1347.	0.6	2
9	Motor and Visual-spatial Cognitive Abilities in Children Treated for Infantile Esotropia. <i>Perceptual and Motor Skills</i> , 2021, 128, 1443-1463.	0.6	0
10	Biallelic mutations in l-dopachrome tautomerase (DCT) cause infantile nystagmus and oculocutaneous albinism. <i>Human Genetics</i> , 2021, 140, 1157-1168.	1.8	12
11	International Classification of Retinopathy of Prematurity, Third Edition. <i>Ophthalmology</i> , 2021, 128, e51-e68.	2.5	280
12	Analysis and management of intraoperative and early postoperative complications of bag-in-the-lens intraocular lens implantation in different age groups of paediatric cataract patients: report of the Giessen Paediatric Cataract Study Group. <i>Acta Ophthalmologica</i> , 2020, 98, e144-e154.	0.6	7
13	Recurrent heterozygous <i>PAX6</i> missense variants cause severe bilateral microphthalmia via predictable effects on DNA-protein interaction. <i>Genetics in Medicine</i> , 2020, 22, 598-609.	1.1	43
14	Cost Effectiveness of Voretigene Neparvovec for <i>RPE65</i> -Mediated Inherited Retinal Degeneration in Germany. <i>Translational Vision Science and Technology</i> , 2020, 9, 17.	1.1	16
15	Creation of different bioluminescence resonance energy transfer based biosensors with high affinity to VEGF. <i>PLoS ONE</i> , 2020, 15, e0230344.	1.1	3
16	New Scleral Depressor Marker for Retinal Detachment Surgery. <i>Ophthalmology Retina</i> , 2019, 3, 73-76.	1.2	2
17	High-dose Anderson operation for nystagmus-related anomalous head turn. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2019, 257, 2033-2041.	1.0	8
18	Biallelic mutation of human <i>SLC6A6</i> encoding the taurine transporter TAUT is linked to early retinal degeneration. <i>FASEB Journal</i> , 2019, 33, 11507-11527.	0.2	36

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19	Fundus-Controlled Dark Adaptometry in Young Children Without and With Spontaneously Regressed Retinopathy of Prematurity. <i>Translational Vision Science and Technology</i> , 2019, 8, 62.	1.1	8
20	Precision of bag-in-the-lens intraocular lens power calculation in different age groups of pediatric cataract patients: Report of the Giessen Pediatric Cataract Study Group. <i>Journal of Cataract and Refractive Surgery</i> , 2019, 45, 1372-1379.	0.7	8
21	Dynamic intraoperative optical coherence tomography for inverted internal limiting membrane flap technique in large macular hole surgery. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2019, 257, 1649-1659.	1.0	21
22	Splitting of the lateral rectus muscle with medial transposition to treat oculomotor palsy: a retrospective analysis of 29 consecutive cases. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2019, 257, 2005-2014.	1.0	12
23	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the <i>GNAT2</i> gene. <i>Human Mutation</i> , 2019, 40, 1145-1155.	1.1	15
24	Cone-Mediated Function Correlates to Altered Foveal Morphology in Preterm-Born Children at School Age. , 2019, 60, 1614.		8
25	Homozygous stop mutation in <i>AHR</i> causes autosomal recessive foveal hypoplasia and infantile nystagmus. <i>Brain</i> , 2019, 142, 1528-1534.	3.7	41
26	Optimizing Measurement of Vascular Endothelial Growth Factor in Small Blood Samples of Premature Infants. <i>Scientific Reports</i> , 2019, 9, 6744.	1.6	4
27	16p13.11 microdeletion uncovers loss of function of a <i>MYH11</i> missense variant in a patient with megacystis-intestinal hypoperistalsis syndrome. <i>Clinical Genetics</i> , 2019, 96, 85-90.	1.0	20
28	Where are the missing gene defects in inherited retinal disorders? Intronic and synonymous variants contribute at least to 4% of <i>CACNA1F</i> -mediated inherited retinal disorders. <i>Human Mutation</i> , 2019, 40, 765-787.	1.1	24
29	The Natural History of Inherited Retinal Dystrophy Due to Biallelic Mutations in the <i>RPE65</i> Gene. <i>American Journal of Ophthalmology</i> , 2019, 199, 58-70.	1.7	77
30	Structure-Function Correlation in Hemianopic Vision Loss in Children Aged 3-6 Years Using OCT and SVOP, and Comparison with Adult Eyes. <i>Ophthalmic Research</i> , 2018, 60, 221-230.	1.0	1
31	Choroidal Thickness with Swept-Source Optical Coherence Tomography versus Foveal Morphology in Young Children with a History of Prematurity. <i>Ophthalmic Research</i> , 2018, 60, 205-213.	1.0	12
32	Clinical history and management recommendations of the smooth muscle dysfunction syndrome due to <i>ACTA2</i> arginine 179 alterations. <i>Genetics in Medicine</i> , 2018, 20, 1206-1215.	1.1	50
33	Reply. <i>Retina</i> , 2018, 38, e65-e66.	1.0	0
34	Clinical Characterization of 66 Patients With Congenital Retinal Disease Due to the Deep-Intronic c.2991+1655A>G Mutation in <i>CEP290</i> . , 2018, 59, 4384.		21
35	OCT Angiography in Young Children with a History of Retinopathy of Prematurity. <i>Ophthalmology Retina</i> , 2018, 2, 972-978.	1.2	30
36	Detection of DNA Double Strand Breaks by γ H2AX Does Not Result in 53bp1 Recruitment in Mouse Retinal Tissues. <i>Frontiers in Neuroscience</i> , 2018, 12, 286.	1.4	10

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37	Development of a Reporter System to Explore MMEJ in the Context of Replacing Large Genomic Fragments. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 11, 407-415.	2.3	13
38	RETINAL VASCULAR DEVELOPMENT WITH 0.312 MG INTRAVITREAL BEVACIZUMAB TO TREAT SEVERE POSTERIOR RETINOPATHY OF PREMATURITY. <i>Retina</i> , 2017, 37, 97-111.	1.0	62
39	Two patients with the heterozygous R189H mutation in <i>ACTA2</i> and Complex congenital heart defects expands the cardiac phenotype of multisystemic smooth muscle dysfunction syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 959-965.	0.7	7
40	Spatially Resolved Spectral Sensitivities as a Potential Read-out Parameter in Clinical Gene Therapeutic Trials. <i>Ophthalmic Research</i> , 2017, 58, 194-202.	1.0	3
41	Retinal layer segmentation in multiple sclerosis: a systematic review and meta-analysis. <i>Lancet Neurology</i> , The, 2017, 16, 797-812.	4.9	397
42	Next-generation sequencing reveals the mutational landscape of clinically diagnosed Usher syndrome: copy number variations, phenocopies, a predominant target for translational read-through, and <i>PEX26</i> mutated in Heimler syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 531-552.	0.6	55
43	Evaluation of tolerance to lentiviral LV-RPE65 gene therapy vector after subretinal delivery in non-human primates. <i>Translational Research</i> , 2017, 188, 40-57.e4.	2.2	21
44	Ocular morphology and function in juvenile neuronal ceroid lipofuscinosis (CLN3) in the first decade of life. <i>Ophthalmic Genetics</i> , 2017, 38, 252-259.	0.5	26
45	InÂvivo genome editing as a potential treatment strategy for inherited retinal dystrophies. <i>Progress in Retinal and Eye Research</i> , 2017, 56, 1-18.	7.3	62
46	Outer Plexiform Layer Structures Are Not Altered Following AAV-Mediated Gene Transfer in Healthy Rat Retina. <i>Frontiers in Neurology</i> , 2017, 8, 59.	1.1	2
47	Detection of the Vascular Endothelial Growth Factor with a Novel Bioluminescence Resonance Energy Transfer Pair Using a Two-Component System. <i>Sensors</i> , 2017, 17, 145.	2.1	6
48	OCT-Based Macular Structureâ€“Function Correlation in Dependence on Birth Weight and Gestational Ageâ€”the Giessen Long-Term ROP Study. , 2016, 57, OCT235.		48
49	Rebound macular edema following oral acetazolamide therapy for juvenile X-linked retinoschisis in an Italian family. <i>Clinical Ophthalmology</i> , 2016, Volume 10, 2377-2382.	0.9	13
50	Genetic Analysis of â€PAX6-Negativeâ€™™ Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016, 11, e0153757.	1.1	54
51	Biallelic Mutations in <i>GNB3</i> Cause a Unique Form of Autosomal-Recessive Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2016, 98, 1011-1019.	2.6	49
52	Comprehensive genotyping reveals RPE65 as the most frequently mutated gene in Leber congenital amaurosis in Denmark. <i>European Journal of Human Genetics</i> , 2016, 24, 1071-1079.	1.4	69
53	Retinal Vessel Pathologies in a Rat Model of Periventricular Leukomalacia: A New Model for Retinopathy of Prematurity?. , 2015, 56, 1830.		12
54	Fundus-controlled two-color dark adaptometry with the Microperimeter MP1. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2015, 253, 965-972.	1.0	9

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55	Recessâ€resect surgery with myopexy of the lateral rectus muscle to correct esotropia with high myopia. <i>British Journal of Ophthalmology</i> , 2015, 99, 1702-1705.	2.1	4
56	Shared Decision-Making, Control Preferences and Psychological Well-Being in Patients with RPE65 Deficiency Awaiting Experimental Gene Therapy. <i>Ophthalmic Research</i> , 2015, 54, 96-102.	1.0	12
57	Bestrophin 1 â€ Phenotypes and Functional Aspects in Bestrophinopathies. <i>Ophthalmic Genetics</i> , 2015, 36, 193-212.	0.5	22
58	Functional Characterization of AAV-Expressed Recombinant Anti-VEGF Single-Chain Variable Fragments In Vitro. <i>Journal of Ocular Pharmacology and Therapeutics</i> , 2015, 31, 269-276.	0.6	5
59	Retinopathy of prematurity: recent developments in diagnosis and treatment. <i>Expert Review of Ophthalmology</i> , 2015, 10, 167-182.	0.3	7
60	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. <i>American Journal of Human Genetics</i> , 2015, 97, 754-760.	2.6	54
61	Retrobulbar Spot Sign Predicts Thrombolytic Treatment Effects and Etiology in Central Retinal Artery Occlusion. <i>Stroke</i> , 2015, 46, 2322-2324.	1.0	62
62	Optical Coherence Tomography (OCT) Device Independent Intraretinal Layer Segmentation. <i>Translational Vision Science and Technology</i> , 2014, 3, 1.	1.1	32
63	Immuno-Histochemical Analysis of Rod and Cone Reaction to RPE65 Deficiency in the Inferior and Superior Canine Retina. <i>PLoS ONE</i> , 2014, 9, e86304.	1.1	7
64	Mosaic synaptopathy and functional defects in Cav1.4 heterozygous mice and human carriers of CSNB2. <i>Human Molecular Genetics</i> , 2014, 23, 1538-1550.	1.4	38
65	Expansion of Ocular Phenotypic Features Associated With Mutations in <i>ADAMTS18</i> . <i>JAMA Ophthalmology</i> , 2014, 132, 996.	1.4	15
66	Clinical utility gene card for: Oculocutaneous albinism. <i>European Journal of Human Genetics</i> , 2014, 22, 1054-1054.	1.4	18
67	Normative values of peripheral retinal thickness measured with Spectralis OCT in healthy young adults. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2014, 252, 1195-1205.	1.0	20
68	Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel <i>LCA5</i> Mutations and New Genotype-Phenotype Correlations. <i>Human Mutation</i> , 2013, 34, 1537-1546.	1.1	32
69	Prevalence and Diagnostic Spectrum of Generalized Retinal Dystrophy in Danish Children. <i>Ophthalmic Epidemiology</i> , 2013, 20, 164-169.	0.8	12
70	Automated Segmentation of Pathological Cavities in Optical Coherence Tomography Scans. , 2013, 54, 4385.		29
71	Automated segmentation of retinal blood vessels in spectral domain optical coherence tomography scans. <i>Biomedical Optics Express</i> , 2012, 3, 1478.	1.5	32
72	Chromatic Pupillometry Dissects Function of the Three Different Light-Sensitive Retinal Cell Populations in RPE65 Deficiency. , 2012, 53, 5641.		50

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73	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2012, 90, 321-330.	2.6	121
74	RPGR: Role in the photoreceptor cilium, human retinal disease, and gene therapy. <i>Ophthalmic Genetics</i> , 2011, 32, 1-11.	0.5	38
75	Long-Term Follow-Up of the Human Phenotype in Three Siblings with Cone Dystrophy Associated with a Homozygous p.G461R Mutation of KCNV2. <i>Investigative Ophthalmology and Visual Science</i> , 2011, 52, 8621.		15
76	Interchangeability of macular thickness measurements between different volumetric protocols of Spectralis optical coherence tomography in normal eyes. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2011, 249, 1137-1145.	1.0	8
77	Large deletions of the <i>KCNV2</i> gene are common in patients with cone dystrophy with supernormal rod response. <i>Human Mutation</i> , 2011, 32, 1398-1406.	1.1	39
78	Comparison of Central Retinal Thickness in Healthy Children and Adults Measured with the Heidelberg Spectralis OCT and the Zeiss Stratus OCT 3. <i>Ophthalmologica</i> , 2011, 225, 27-36.	1.0	19
79	The effects of midazolam on intraocular pressure in children during examination under sedation. <i>British Journal of Ophthalmology</i> , 2011, 95, 1102-1105.	2.1	25
80	Screening of TYR, OCA2, GPR143, and MC1R in patients with congenital nystagmus, macular hypoplasia, and fundus hypopigmentation indicating albinism. <i>Molecular Vision</i> , 2011, 17, 939-48.	1.1	38
81	LACK OF ALTOFLUORESCENCE IN FUNDUS ALBIPUNCTATUS ASSOCIATED WITH MUTATIONS IN RDH5. <i>Retina</i> , 2010, 30, 1704-1713.	1.0	39
82	Superior oblique tucking with versus without additional inferior oblique recession for acquired trochlear nerve palsy. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2010, 248, 223-229.	1.0	9
83	Feasibility and outcome of automated kinetic perimetry in children. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2010, 248, 1493-1500.	1.0	17
84	Retinal Blinding Disorders and Gene Therapy - Molecular and Clinical Aspects. <i>Current Gene Therapy</i> , 2010, 10, 350-370.	0.9	11
85	Gene therapy for vision loss -- recent developments. <i>Discovery Medicine</i> , 2010, 10, 425-33.	0.5	22
86	Systematic Review of Digital Imaging Screening Strategies for Retinopathy of Prematurity. <i>Pediatrics</i> , 2009, 123, e360-e361.	1.0	4
87	Detection of Intact rAAV Particles up to 6 Years After Successful Gene Transfer in the Retina of Dogs and Primates. <i>Molecular Therapy</i> , 2009, 17, 516-523.	3.7	73
88	Wide-field digital imaging based telemedicine for screening for acute retinopathy of prematurity (ROP). Six-year results of a multicentre field study. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2009, 247, 1251-1262.	1.0	140
89	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2009, 85, 720-729.	2.6	207
90	Evolution of central corneal thickness in children with congenital glaucoma requiring glaucoma surgery. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2008, 246, 397-403.	1.0	21

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91	A Comprehensive Clinical and Biochemical Functional Study of a Novel RPE65 Hypomorphic Mutation. , 2008, 49, 5235.		73
92	Identification of novel mutations in X-linked retinitis pigmentosa families and implications for diagnostic testing. <i>Molecular Vision</i> , 2008, 14, 1081-93.	1.1	46
93	Novel causative mutations in patients with Nance-Horan syndrome and altered localization of the mutant NHS-A protein isoform. <i>Molecular Vision</i> , 2008, 14, 1856-64.	1.1	26
94	De novo double mutation in PAX6 and mtDNA tRNA Lys associated with atypical aniridia and mitochondrial disease. <i>Journal of Molecular Medicine</i> , 2007, 85, 163-168.	1.7	11
95	Electron microscopic findings in levator muscle biopsies of patients with isolated congenital or acquired ptosis. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2007, 245, 1533-1541.	1.0	18
96	Mutation analysis in a family with oculocutaneous albinism manifesting in the same generation of three branches. <i>Molecular Vision</i> , 2007, 13, 1851-5.	1.1	12
97	Pediatric Ophthalmology in Germany. <i>Journal of AAPOS</i> , 2006, 10, 1-3.	0.2	3
98	Assessment of Cortical Visual Field Representations with Multifocal VEPs in Control Subjects, Patients with Albinism, and Female Carriers of Ocular Albinism. , 2006, 47, 3195.		18
99	Role of SOX2 Mutations in Human Hippocampal Malformations and Epilepsy. <i>Epilepsia</i> , 2006, 47, 534-542.	2.6	85
100	Pediatric clinical visual electrophysiology: a survey of actual practice. <i>Documenta Ophthalmologica</i> , 2006, 113, 193-204.	1.0	28
101	Fundus autofluorescence in children and teenagers with hereditary retinal diseases. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2006, 244, 36-45.	1.0	59
102	Congenital cataract and macular hypoplasia in humans associated with a de novo mutation in CRYAA and compound heterozygous mutations in P. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2006, 244, 912-919.	1.0	65
103	Ependymal Tissue in Microphthalmia with Cyst. <i>Ophthalmic Genetics</i> , 2006, 27, 165-168.	0.5	9
104	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. <i>European Journal of Human Genetics</i> , 2005, 13, 302-308.	1.4	216
105	Longitudinal and cross-sectional study of patients with early-onset severe retinal dystrophy associated with RPE65 mutations. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2005, 243, 417-426.	1.0	75
106	Disturbed visual system function in methionine synthase deficiency. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2005, 243, 497-500.	1.0	25
107	Age matters? thoughts on a grading system for ABCA4 mutations. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2005, 243, 87-89.	1.0	10
108	Misrouting of the Optic Nerves in Albinism: Estimation of the Extent with Visual Evoked Potentials. , 2005, 46, 3892.		68

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109	Genotyping Microarray (Disease Chip) for Leber Congenital Amaurosis: Detection of Modifier Alleles. , 2005, 46, 3052.		153
110	Oculo-auriculo-vertebral spectrum (OAVS): clinical evaluation and severity scoring of 53 patients and proposal for a new classification. European Journal of Medical Genetics, 2005, 48, 397-411.	0.7	184
111	Heterozygous Mutations of OTX2 Cause Severe Ocular Malformations. American Journal of Human Genetics, 2005, 76, 1008-1022.	2.6	266
112	Fundus autofluorescence in carriers of X-linked recessive retinitis pigmentosa associated with mutations in RPGR, and correlation with electrophysiological and psychophysical data. Graefe's Archive for Clinical and Experimental Ophthalmology, 2004, 242, 501-511.	1.0	52
113	Unilateral adult malignant optic nerve glioma. Graefe's Archive for Clinical and Experimental Ophthalmology, 2004, 242, 741-748.	1.0	63
114	Bilateral Tonic Pupils with Evidence of Anti-Hu Antibodies as a Paraneoplastic Manifestation of Small Cell Lung Cancer. Ophthalmologica, 2004, 218, 141-143.	1.0	36
115	Lack of fundus autofluorescence to 488 nanometers from childhood on in patients with early-onset severe retinal dystrophy associated with mutations in RPE65. Ophthalmology, 2004, 111, 1585-1594.	2.5	144
116	Fourteen novel OPA1 mutations in autosomal dominant optic atrophy including two de novo mutations in sporadic optic atrophy. Human Mutation, 2003, 21, 656-656.	1.1	57
117	Analysis of three genes in Leber congenital amaurosis in Indonesian patients. Vision Research, 2003, 43, 3087-3093.	0.7	21
118	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 72, 478-487.	2.6	219
119	Phenotypic variability in three carriers from a family with choroideremia and a frameshift mutation 1388delCCinsG in the REP-1 gene. Ophthalmic Genetics, 2003, 24, 203-214.	0.5	31
120	Causes of blindness at the "Wiyata Guna" School for the Blind, Indonesia. British Journal of Ophthalmology, 2003, 87, 1065-1068.	2.1	17
121	X-linked Retinitis Pigmentosa:RPGRMutations in Most Families with Definite X Linkage and Clustering of Mutations in a Short Sequence Stretch of Exon ORF15. , 2003, 44, 1458.		103
122	Phenotype in two families with RP3 associated with RPGR mutations. Ophthalmic Genetics, 2003, 24, 89-101.	0.5	24
123	In search for increased prevalence rates of strabismus and microstrabismus in two Bavarian districts, Oberpfalz and Niederbayern, to spot populations for gene identification. Strabismus, 2002, 10, 163-168.	0.4	3
124	Genetics of isolated and syndromic strabismus: Facts and perspectives. Strabismus, 2002, 10, 147-156.	0.4	50
125	Mutations in the Cone Photoreceptor G-Protein $\hat{\pm}$ -Subunit Gene GNAT2 in Patients with Achromatopsia. American Journal of Human Genetics, 2002, 71, 422-425.	2.6	245
126	Identification and in vitro expression of novelCDH23 mutations of patients with Usher syndrome type 1D. Human Mutation, 2002, 19, 268-273.	1.1	40

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127	First evidence of an endogenous <i>Spiroplasma</i> sp. infection in humans manifesting as unilateral cataract associated with anterior uveitis in a premature baby. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2002, 240, 348-353.	1.0	33
128	Thirty distinct CACNA1F mutations in 33 families with incomplete type of XLCSNB and <i>Cacna1f</i> expression profiling in mouse retina. <i>European Journal of Human Genetics</i> , 2002, 10, 449-456.	1.4	75
129	BIGH3 mutation spectrum in corneal dystrophies. <i>Investigative Ophthalmology and Visual Science</i> , 2002, 43, 949-54.	3.3	142
130	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. <i>American Journal of Human Genetics</i> , 2001, 69, 722-737.	2.6	294
131	Mutation spectrum and splicing variants in the OPA1 gene. <i>Human Genetics</i> , 2001, 109, 584-591.	1.8	327
132	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. <i>Human Genetics</i> , 2001, 108, 51-54.	1.8	31
133	Mutations in the VMD2 gene are associated with juvenile-onset vitelliform macular dystrophy (Best) Tj ETQq1 1 0.784314 rgBT /Overl <i>Journal of Human Genetics</i> , 2000, 8, 286-292.	1.4	198
134	A Comprehensive Survey of Sequence Variation in the ABCA4 (ABCR) Gene in Stargardt Disease and Age-Related Macular Degeneration. <i>American Journal of Human Genetics</i> , 2000, 67, 800-813.	2.6	308
135	Assessment of RS1 in X-linked juvenile retinoschisis and sporadic senile retinoschisis. <i>Clinical Genetics</i> , 1999, 55, 461-465.	1.0	15
136	An L-type calcium-channel gene mutated in incomplete X-linked congenital stationary night blindness. <i>Nature Genetics</i> , 1998, 19, 260-263.	9.4	450
137	Tubby-like protein-1 mutations in autosomal recessive retinitis pigmentosa. <i>Lancet, The</i> , 1998, 351, 1103-1104.	6.3	74
138	IRPA Scientific Newsletter "A new website to integrate data on hereditary retinal disorders. <i>Ophthalmic Genetics</i> , 1998, 19, 213-214.	0.5	0
139	Constitutional RB1-Gene Mutations in Patients with Isolated Unilateral Retinoblastoma. <i>American Journal of Human Genetics</i> , 1997, 61, 282-294.	2.6	148
140	Positional cloning of the gene associated with X-linked juvenile retinoschisis. <i>Nature Genetics</i> , 1997, 17, 164-170.	9.4	446
141	Mutations in RPE65 cause autosomal recessive childhood-onset severe retinal dystrophy. <i>Nature Genetics</i> , 1997, 17, 194-197.	9.4	599
142	A tribute to Mette Warburg. <i>Ophthalmic Genetics</i> , 1996, 17, 123-125.	0.5	0
143	A gene (SRPX) encoding a sushi-repeat-containing protein is deleted in patients with X-linked retinitis pigmentosa. <i>Human Molecular Genetics</i> , 1995, 4, 2339-2346.	1.4	54
144	Aplasia of the optic nerve in two cases of partial trisomy 10q24. <i>Clinical Genetics</i> , 1995, 48, 183-187.	1.0	13

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145	Transscleral and indirect ophthalmoscope diode laser retinal photocoagulation : Experimental quantification of the therapeutic range for their application in the treatment of retinopathy of prematurity. Graefe's Archive for Clinical and Experimental Ophthalmology, 1993, 231, 378-383.	1.0	7
146	Ocular growth in infant aphakia. Ophthalmic Paediatrics and Genetics, 1993, 14, 177-188.	0.4	28
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