Frans P M Cremers

List of Publications by Year
in descending order

301 papers	22,650 citations	6124 83 h-index	15253 130 g-index
313 all docs	313 docs citations	313 times ranked	15343 citing authors

#	Article	IF	Citations
1	BBS1 branchpoint variant is associated with non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2022, 59, 438-444.	1.5	13
2	CRB1-Associated Retinal Dystrophies: A Prospective Natural History Study in Anticipation of Future Clinical Trials. American Journal of Ophthalmology, 2022, 234, 37-48.	1.7	17
3	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	1.8	3
4	Stargardt disease: monitoring incidence and diagnostic trends in the Netherlands using a nationwide disease registry. Acta Ophthalmologica, 2022, 100, 395-402.	0.6	10
5	Personalized genetic counseling for Stargardt disease: Offspring risk estimates based on variant severity. American Journal of Human Genetics, 2022, 109, 498-507.	2.6	23
6	ABCA4c.859-25A>G, a Frequent Palestinian Founder Mutation Affecting the Intron 7 Branchpoint, Is Associated With Early-Onset Stargardt Disease. , 2022, 63, 20.		3
7	Identification of a Complex Allele in IMPG2 as a Cause of Adult-Onset Vitelliform Macular Dystrophy. , 2022, 63, 27.		13
8	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	1.7	5
9	A RIPOR2 in-frame deletion is a frequent and highly penetrant cause of adult-onset hearing loss. Journal of Medical Genetics, 2021, 58, 96-104.	1.5	14
10	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	1.6	71
11	Defining inclusion criteria and endpoints for clinical trials: a prospective crossâ€sectional study in CRB1â€associated retinal dystrophies. Acta Ophthalmologica, 2021, 99, e402-e414.	0.6	10
12	Clinical Phenotype of PDE6B-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 2374.	1.8	12
13	The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet Journal of Rare Diseases, 2021, 16, 142.	1.2	25
14	The Impact of Modern Technologies on Molecular Diagnostic Success Rates, with a Focus on Inherited Retinal Dystrophy and Hearing Loss. International Journal of Molecular Sciences, 2021, 22, 2943.	1.8	6
15	Antisense Oligonucleotide-Based Rescue of Aberrant Splicing Defects Caused by 15 Pathogenic Variants in ABCA4. International Journal of Molecular Sciences, 2021, 22, 4621.	1.8	30
16	Benchmarking deep learning splice prediction tools using functional splice assays. Human Mutation, 2021, 42, 799-810.	1.1	59
17	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	1.8	8
18	PRPH2 mutation update: In silico assessment of 245 reported and 7 novel variants in patients with retinal disease. Human Mutation, 2021, 42, 1521-1547.	1.1	13

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19	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.0	4
20	Retinal Degeneration Associated With RPGRIP1: A Review of Natural History, Mutation Spectrum, and Genotypeâ€“Phenotype Correlation in 228 Patients. Frontiers in Cell and Developmental Biology, 2021, 9, 746781.	1.8	9
21	Non-syndromic inherited retinal diseasesÂin Poland: Genes, mutations, and phenotypes. Molecular Vision, 2021, 27, 457-465.	1.1	3
22	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	1.7	27
23	A nationwide genetic analysis of inherited retinal diseases in Israel as assessed by the Israeli inherited retinal disease consortium (IIRDC). Human Mutation, 2020, 41, 140-149.	1.1	75
24	CEP290 Mutation Spectrum and Delineation of the Associated Phenotype in a Large German Cohort: A Monocentric Study. American Journal of Ophthalmology, 2020, 211, 142-150.	1.7	27
25	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	2.3	55
26	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	2.6	75
27	ABCA4-Associated Stargardt Disease. Klinische Monatsblatter Fur Augenheilkunde, 2020, 237, 267-274.	0.3	17
28	LONGITUDINAL STUDY OF RPE65-ASSOCIATED INHERITED RETINAL DEGENERATIONS. Retina, 2020, 40, 1812-1828.	1.0	12
29	Unique combination of clinical features in a large cohort of 100 patients with retinitis pigmentosa caused by FAM161A mutations. Scientific Reports, 2020, 10, 15156.	1.6	14
30	Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. JAMA Ophthalmology, 2020, 138, 1035.	1.4	31
31	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
32	Clinical spectrum, genetic complexity and therapeutic approaches for retinal disease caused by ABCA4 mutations. Progress in Retinal and Eye Research, 2020, 79, 100861.	7.3	173
33	In or Out? New Insights on Exon Recognition through Splice-Site Interdependency. International Journal of Molecular Sciences, 2020, 21, 2300.	1.8	8
34	Foveal Sparing in Central Retinal Dystrophies. , 2019, 60, 3456.		24
35	Identification of splice defects due to noncanonical splice site or deepâ€intronic variants in ABCA4. Human Mutation, 2019, 40, 2365-2376.	1.1	46
36	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	5.8	21

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37	Late-Onset Stargardt Disease Due to Mild, Deep-Intronic ABCA4 Alleles. , 2019, 60, 4249.		25
38	Highly Variable Disease Courses in Siblings with Stargardt Disease. Ophthalmology, 2019, 126, 1712-1721.	2.5	16
39	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. Genetics in Medicine, 2019, 21, 1761-1771.	1.1	111
40	Costâ€effective molecular inversion probeâ€based ABCA4 sequencing reveals deepâ€intronic variants in Stargardt disease. Human Mutation, 2019, 40, 1749-1759.	1.1	39
41	Intein-mediated protein trans-splicing expands adeno-associated virus transfer capacity in the retina. Science Translational Medicine, 2019, 11, .	5.8	109
42	The identification of a RNA splice variant in TULP1 in two siblings with earlyâ€onset photoreceptor dystrophy. Molecular Genetics & Genomic Medicine, 2019, 7, e660.	0.6	14
43	The absence of fundus abnormalities in Stargardt disease. Graefe's Archive for Clinical and Experimental Ophthalmology, 2019, 257, 1147-1157.	1.0	25
44	Chromatic Full-Field Stimulus Threshold and Pupillography as Functional Markers for Late-Stage, Early-Onset Retinitis Pigmentosa Caused by CRB1 Mutations. Translational Vision Science and Technology, 2019, 8, 45.	1.1	13
45	Genetic Spectrum of ABCA4-Associated Retinal Degeneration in Poland. Genes, 2019, 10, 959.	1.0	17
46	Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. Genetics in Medicine, 2019, 21, 1751-1760.	1.1	147
47	CLINICAL AND GENETIC CHARACTERISTICS OF MALE PATIENTS WITH RPGR-ASSOCIATED RETINAL DYSTROPHIES. Retina, 2019, 39, 1186-1199.	1.0	56
48	Identification and Analysis of Genes Associated with Inherited Retinal Diseases. Methods in Molecular Biology, 2019, 1834, 3-27.	0.4	12
49	Targeted next generation sequencing reveals genetic defects underlying inherited retinal disease in Iranian families. Molecular Vision, 2019, 25, 106-117.	1.1	10
50	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 614-624.	1.4	26
51	Antisense Oligonucleotide-Based Splice Correction of a Deep-Intronic Mutation in CHM Underlying Choroideremia. Advances in Experimental Medicine and Biology, 2018, 1074, 83-89.	0.8	33
52	Amelioration of Neurosensory Structure and Function in Animal and Cellular Models of a Congenital Blindness. Molecular Therapy, 2018, 26, 1581-1593.	3.7	19
53	Identification and Rescue of Splice Defects Caused by Two Neighboring Deep-Intronic ABCA4 Mutations Underlying Stargardt Disease. American Journal of Human Genetics, 2018, 102, 517-527.	2.6	105
54	ABCA4 midigenes reveal the full splice spectrum of all reported noncanonical splice site variants in Stargardt disease. Genome Research, 2018, 28, 100-110.	2.4	134

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55	EYSmutation update: In silico assessment of 271 reported and 26 novel variants in patients with retinitis pigmentosa. Human Mutation, 2018, 39, 177-186.	1.1	23
56	Molecular and clinical analysis of 27 German patients with Leber congenital amaurosis. PLoS ONE, 2018, 13, e0205380.	1.1	38
57	Author Response: Penetrance of theABCA4p.Asn1868Ile Allele in Stargardt Disease. , 2018, 59, 5566.		19
58	The CommonABCA4Variant p.Asn1868Ile Shows Nonpenetrance and Variable Expression of Stargardt Disease When Present intransWith Severe Variants. , 2018, 59, 3220.		67
59	Detection and quantification of a KIF11 mosaicism in a subject presenting familial exudative vitreoretinopathy with microcephaly. European Journal of Human Genetics, 2018, 26, 1819-1823.	1.4	9
60	Identification of Inherited Retinal Disease-Associated Genetic Variants in 11 Candidate Genes. Genes, 2018, 9, 21.	1.0	20
61	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
62	Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotypeâ€“Phenotype Correlations, and Inheritance Models. Genes, 2018, 9, 215.	1.0	58
63	Autosomal Recessive NRL Mutations in Patients with Enhanced S-Cone Syndrome. Genes, 2018, 9, 68.	1.0	35
64	Homozygous variants in KIAA1549, encoding a ciliary protein, are associated with autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2018, 55, 705-712.	1.5	13
65	The Spectrum of Structural and Functional Abnormalities in Female Carriers of Pathogenic Variants in theRPGRGene. , 2018, 59, 4123.		41
66	Putative digenic inheritance of heterozygousRP1L1andC2orf71null mutations in syndromic retinal dystrophy. Ophthalmic Genetics, 2017, 38, 127-132.	0.5	22
67	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	1.1	118
68	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	1.4	104
69	Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudocoloboma. Ophthalmology, 2017, 124, 992-1003.	2.5	37
70	Missense mutations in the WD40 domain ofAHI1cause non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2017, 54, 624-632.	1.5	21
71	Genotypic and Phenotypic Characteristics of CRB1 -Associated Retinal Dystrophies. Ophthalmology, 2017, 124, 884-895.	2.5	75
72	Deletions OverlappingVCANExon 8 Are New Molecular Defects for Wagner Disease. Human Mutation, 2017, 38, 43-47.	1.1	16

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73	A Rare Form of Retinal Dystrophy Caused by Hypomorphic Nonsense Mutations in CEP290. Genes, 2017, 8, 208.	1.0	25
74	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. Genes, 2017, 8, 381.	1.0	19
75	Homozygosity Mapping and Targeted Sanger Sequencing Identifies Three Novel CRB1 (Cumbs) Tj ETQq1 1 0.784314 rgBT /Overlock 10 294-302.	0.4	7
76	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1. , 2016, 57, 4806.		25
77	Asymmetric Inter-Eye Progression in Stargardt Disease. , 2016, 57, 6824.		17
78	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
79	Biallelic Mutations inCRB1Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
80	Mutations in the polyglutamylase geneTTLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. Human Molecular Genetics, 2016, 25, ddw282.	1.4	27
81	Mutations in CEP78 Cause Cone-Rod Dystrophy and Hearing Loss Associated with Primary-Cilia Defects. American Journal of Human Genetics, 2016, 99, 770-776.	2.6	44
82	Genetic and clinical characterization of Pakistani families with Bardet-Biedl syndrome extends the genetic and phenotypic spectrum. Scientific Reports, 2016, 6, 34764.	1.6	29
83	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	1.1	64
84	Photoreceptor Progenitor mRNA Analysis Reveals Exon Skipping Resulting from the ABCA4 c.5461-10Tât'C Mutation in Stargardt Disease. Ophthalmology, 2016, 123, 1375-1385.	2.5	96
85	Visual Prognosis in USH2A-Associated Retinitis Pigmentosa Is Worse for Patients with Usher Syndrome Type IIa Than for Those with Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2016, 123, 1151-1160.	2.5	76
86	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	9.4	50
87	Comprehensive genotyping reveals RPE65 as the most frequently mutated gene in Leber congenital amaurosis in Denmark. European Journal of Human Genetics, 2016, 24, 1071-1079.	1.4	69
88	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
89	A Nonsense Mutation inFAM161AIs a Recurrent Founder Allele in Dutch and Belgian Individuals With Autosomal Recessive Retinitis Pigmentosa. , 2015, 56, 7418.		9
90	Homozygosity Mapping and Targeted Sanger Sequencing Reveal Genetic Defects Underlying Inherited Retinal Disease in Families from Pakistan. PLoS ONE, 2015, 10, e0119806.	1.1	27

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91	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765.	9.4	183
92	Cerebral visual impairment and intellectual disability caused by PGAP1 variants. European Journal of Human Genetics, 2015, 23, 1689-1693.	1.4	15
93	Cerebral visual impairment, autism, and pancreatitis associated with a 9â€‰Mbp deletion on 10p12. Clinical Dysmorphology, 2015, 24, 34-37.	0.1	1
94	Non-syndromic retinitis pigmentosa due to mutations in the mucopolysaccharidosis type IIIC gene, heparan-alpha-glucosaminide N-acetyltransferase (HGSNAT). Human Molecular Genetics, 2015, 24, 3742-51.	1.4	47
95	Early-Onset Stargardt Disease. Ophthalmology, 2015, 122, 335-344.	2.5	127
96	Heterozygous Deep-Intronic Variants and Deletions inABCA4in Persons with Retinal Dystrophies and One ExonicABCA4Variant. Human Mutation, 2015, 36, 43-47.	1.1	68
97	Mutations in MFSD8, Encoding a Lysosomal Membrane Protein, Are Associated with Nonsyndromic Autosomal Recessive Macular Dystrophy. Ophthalmology, 2015, 122, 170-179.	2.5	60
98	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	1.4	136
99	Foveal Sparing in Stargardt Disease. , 2014, 55, 7467.		60
100	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
101	The RD5000 Database: Facilitating Clinical, Genetic, and Therapeutic Studies on Inherited Retinal Diseases. , 2014, 55, 7355.		27
102	Comprehensive Registration of DNA Sequence Variants Associated with Inherited Retinal Diseases in Leiden Open Variation Databases. Human Mutation, 2014, 35, 147-148.	1.1	5
103	Prenylation defects in inherited retinal diseases. Journal of Medical Genetics, 2014, 51, 143-151.	1.5	26
104	Nonpenetrance of the Most Frequent Autosomal Recessive Leber Congenital Amaurosis Mutation inNMNAT1. JAMA Ophthalmology, 2014, 132, 1002.	1.4	28
105	Retinal gene therapy in patients with choroideremia: initial findings from a phase 1/2 clinical trial. Lancet, The, 2014, 383, 1129-1137.	6.3	689
106	Oral 9-cis retinoid for childhood blindness due to Leber congenital amaurosis caused by RPE65 or LRAT mutations: an open-label phase 1b trial. Lancet, The, 2014, 384, 1513-1520.	6.3	91
107	A missense mutation in the splicing factor geneDHX38is associated with early-onset retinitis pigmentosa with macular coloboma. Journal of Medical Genetics, 2014, 51, 444-448.	1.5	48
108	Disruption of the Basal Body Protein POC1B Results in Autosomal-Recessive Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 95, 131-142.	2.6	65

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109	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
110	Low vision due to cerebral visual impairment: differentiating between acquired and genetic causes. BMC Ophthalmology, 2014, 14, 59.	0.6	29
111	Involvement of LCA5 in Leber Congenital Amaurosis and Retinitis Pigmentosa in the Spanish Population. Ophthalmology, 2014, 121, 399-407.	2.5	20
112	Genomic Approaches For the Discovery of Genes Mutated in Inherited Retinal Degeneration. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a017137-a017137.	2.9	10
113	Chromosomal aberrations in cerebral visual impairment. European Journal of Paediatric Neurology, 2014, 18, 677-684.	0.7	20
114	Causes and consequences of inherited cone disorders. Progress in Retinal and Eye Research, 2014, 42, 1-26.	7.3	127
115	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. Ophthalmology, 2014, 121, 1620-1627.	2.5	44
116	The Molecular Basis of Retinal Dystrophies in Pakistan. Genes, 2014, 5, 176-195.	1.0	20
117	Novel compound heterozygous NMNAT1 variants associated with Leber congenital amaurosis. Molecular Vision, 2014, 20, 753-9.	1.1	29
118	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	2.6	71
119	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	1.1	303
120	Mutations in the Mevalonate Kinase (MVK) Gene Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2013, 120, 2697-2705.	2.5	56
121	Maternal Uniparental Isodisomy of Chromosome 6 Reveals a TULP1 Mutation as a Novel Cause of Cone Dysfunction. Ophthalmology, 2013, 120, 1239-1246.	2.5	36
122	Mutations in RAB28, Encoding a Farnesylated Small GTPase, Are Associated with Autosomal-Recessive Cone-Rod Dystrophy. American Journal of Human Genetics, 2013, 93, 110-117.	2.6	85
123	Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel LCA5 Mutations and New Genotype-Phenotype Correlations. Human Mutation, 2013, 34, 1537-1546.	1.1	32
124	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	3.3	144
125	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. PLoS ONE, 2013, 8, e51622.	1.1	16
126	Unexpected CEP290 mRNA Splicing in a Humanized Knock-In Mouse Model for Leber Congenital Amaurosis. PLoS ONE, 2013, 8, e79369.	1.1	55

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127	Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. Molecular Vision, 2013, 19, 644-53.	1.1	26
128	Antisense Oligonucleotide (AON)-based Therapy for Leber Congenital Amaurosis Caused by a Frequent Mutation in CEP290. Molecular Therapy - Nucleic Acids, 2012, 1, e14.	2.3	116
129	Non-syndromic retinal ciliopathies: translating gene discovery into therapy. Human Molecular Genetics, 2012, 21, R111-R124.	1.4	117
130	BBS1 Mutations in a Wide Spectrum of Phenotypes Ranging From Nonsyndromic Retinitis Pigmentosa to Bardet-Biedl Syndrome. JAMA Ophthalmology, 2012, 130, 1425.	2.6	106
131	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
132	A Homozygous Frameshift Mutation in LRAT Causes Retinitis Punctata Albescens. Ophthalmology, 2012, 119, 1899-1906.	2.5	41
133	Clinical Course, Genetic Etiology, and Visual Outcome in Cone and Coneâ€“Rod Dystrophy. Ophthalmology, 2012, 119, 819-826.	2.5	115
134	Clinical and Genetic Characteristics of Late-onset Stargardt's Disease. Ophthalmology, 2012, 119, 1199-1210.	2.5	162
135	A Nonsense Mutation in PDE6H Causes Autosomal-Recessive Incomplete Achromatopsia. American Journal of Human Genetics, 2012, 91, 527-532.	2.6	148
136	Expression of Wild-Type Rp1 Protein in Rp1 Knock-in Mice Rescues the Retinal Degeneration Phenotype. PLoS ONE, 2012, 7, e43251.	1.1	22
137	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2012, 53, 4458.		81
138	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. American Journal of Human Genetics, 2012, 90, 102-109.	2.6	82
139	The Power of Homozygosity Mapping: Discovery of New Genetic Defects in Patients with Retinal Dystrophy. Advances in Experimental Medicine and Biology, 2012, 723, 345-351.	0.8	15
140	Identification and Analysis of Inherited Retinal Disease Genes. Methods in Molecular Biology, 2012, 935, 3-23.	0.4	16
141	Ocular and extra-ocular features of patients with Leber congenital amaurosis and mutations in CEP290. Molecular Vision, 2012, 18, 412-25.	1.1	40
142	A nonsense mutation in S-antigen (p.Glu306*) causes Oguchi disease. Molecular Vision, 2012, 18, 1253-9.	1.1	18
143	Identification of recurrent and novel mutations in TULP1 in Pakistani families with early-onset retinitis pigmentosa. Molecular Vision, 2012, 18, 1226-37.	1.1	17
144	Novel mutations in RDH5 cause fundus albipunctatus in two consanguineous Pakistani families. Molecular Vision, 2012, 18, 1558-71.	1.1	15

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145	A novel crumbs homolog 1 mutation in a family with retinitis pigmentosa, nanophthalmos, and optic disc drusen. Molecular Vision, 2012, 18, 2447-53.	1.1	27
146	Identification of a novel nonsense mutation in RP1 that causes autosomal recessive retinitis pigmentosa in an Indonesian family. Molecular Vision, 2012, 18, 2411-9.	1.1	16
147	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2011, 118, 1444-1448.	2.5	61
148	Late Onset Retinitis Pigmentosa. Ophthalmology, 2011, 118, 2523-2524.	2.5	6
149	IQCB1Mutations in Patients with Leber Congenital Amaurosis. , 2011, 52, 834.		107
150	Exome Sequencing and cis-Regulatory Mapping Identify Mutations in MAK, a Gene Encoding a Regulator of Ciliary Length, as a Cause of Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 89, 253-264.	2.6	95
151	Large deletions of theKCNV2gene are common in patients with cone dystrophy with supernormal rod response. Human Mutation, 2011, 32, 1398-1406.	1.1	39
152	High-Resolution Homozygosity Mapping Is a Powerful Tool to Detect Novel Mutations Causative of Autosomal Recessive RP in the Dutch Population. , 2011, 52, 2227.		67
153	The ciliopathy-associated protein homologs RPGRIP1 and RPGRIP1L are linked to cilium integrity through interaction with Nek4 serine/threonine kinase. Human Molecular Genetics, 2011, 20, 3592-3605.	1.4	60
154	Autosomal Recessive Stickler Syndrome in Two Families Is Caused by Mutations in theCOL9A1Gene. , 2011, 52, 4774.		40
155	Identification of Novel Mutations in Pakistani Families With Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2011, 129, 1377.	2.6	15
156	Disruption of intraflagellar protein transport in photoreceptor cilia causes Leber congenital amaurosis in humans and mice. Journal of Clinical Investigation, 2011, 121, 2169-2180.	3.9	94
157	Lighting a candle in the dark: advances in genetics and gene therapy of recessive retinal dystrophies. Journal of Clinical Investigation, 2011, 121, 456-456.	3.9	1
158	Molecular genetic analysis of retinitis pigmentosa in Indonesia using genome-wide homozygosity mapping. Molecular Vision, 2011, 17, 3013-24.	1.1	28
159	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	2.6	202
160	Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. American Journal of Human Genetics, 2010, 86, 138-147.	2.6	58
161	Mutations in C2ORF71 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 783-788.	2.6	88
162	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	2.6	98

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163	CDK19 is disrupted in a female patient with bilateral congenital retinal folds, microcephaly and mild mental retardation. Human Genetics, 2010, 128, 281-291.	1.8	50
164	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. Human Mutation, 2010, 31, 656-666.	1.1	126
165	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	9.4	171
166	Novel Null Mutations in theEYSGene Are a Frequent Cause of Autosomal Recessive Retinitis Pigmentosa in the Israeli Population. , 2010, 51, 4387.		57
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