Frans P M Cremers

List of Publications by Year
in descending order

301 papers	22,650 citations	5261 83 h-index	13365 130 g-index
313 all docs	313 docs citations	313 times ranked	14224 citing authors

#	Article	IF	Citations
1	Leber congenital amaurosis: Genes, proteins and disease mechanisms. Progress in Retinal and Eye Research, 2008, 27, 391-419.	7.3	708
2	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
3	Mutations in the CEP290 (NPHP6) Gene Are a Frequent Cause of Leber Congenital Amaurosis. American Journal of Human Genetics, 2006, 79, 556-561.	2.6	608
4	Mutations in a human homologue of Drosophila crumbs cause retinitis pigmentosa (RP12). Nature Genetics, 1999, 23, 217-221.	9.4	427
5	Positional cloning of the gene for X-linked retinitis pigmentosa 2. Nature Genetics, 1998, 19, 327-332.	9.4	371
6	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	9.4	329
7	Cloning of a gene that is rearranged in patients with choroideraemia. Nature, 1990, 347, 674-677.	13.7	325
8	cDNA cloning of component A of Rab geranylgeranyl transferase and demonstration of its role as a Rab escort protein. Cell, 1993, 73, 1091-1099.	13.5	325
9	Leber Congenital Amaurosis and Retinitis Pigmentosa with Coats-like Exudative Vasculopathy Are Associated with Mutations in the Crumbs Homologue 1 (CRB1) Gene. American Journal of Human Genetics, 2001, 69, 198-203.	2.6	322
10	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
11	Mutations in the ABCA4 (ABCR) Gene Are the Major Cause of Autosomal Recessive Cone-Rod Dystrophy. American Journal of Human Genetics, 2000, 67, 960-966.	2.6	294
12	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. American Journal of Human Genetics, 2001, 69, 722-737.	2.6	294
13	Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. Nature Genetics, 2007, 39, 882-888.	9.4	285
14	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
15	The 2588Gâ†'C Mutation in the ABCR Gene Is a Mild Frequent Founder Mutation in the Western European Population and Allows the Classification of ABCR Mutations in Patients with Stargardt Disease. American Journal of Human Genetics, 1999, 64, 1024-1035.	2.6	242
16	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. European Journal of Human Genetics, 2005, 13, 302-308.	1.4	216
17	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
18	The spectrum of retinal dystrophies caused by mutations in the peripherin/RDS gene. Progress in Retinal and Eye Research, 2008, 27, 213-235.	7.3	200

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19	Homozygosity Mapping Reveals PDE6C Mutations in Patients with Early-Onset Cone Photoreceptor Disorders. American Journal of Human Genetics, 2009, 85, 240-247.	2.6	194
20	Positional cloning of the gene for X-linked retinitis pigmentosa 3: homology with the guanine-nucleotide-exchange factor RCC1. Human Molecular Genetics, 1996, 5, 1035-1041.	1.4	186
21	Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. Nature Genetics, 2007, 39, 889-895.	9.4	186
22	Lighting a candle in the dark: advances in genetics and gene therapy of recessive retinal dystrophies. Journal of Clinical Investigation, 2010, 120, 3042-3053.	3.9	183
23	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765.	9.4	183
24	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. American Journal of Human Genetics, 2009, 85, 465-481.	2.6	180
25	The retinitis pigmentosa GTPase regulator (RPGR) interacts with novel transport-like proteins in the outer segments of rod photoreceptors. Human Molecular Genetics, 2000, 9, 2095-2105.	1.4	179
26	Identification of 51 Novel Exons of the Usher Syndrome Type 2A (USH2A) Gene That Encode Multiple Conserved Functional Domains and That Are Mutated in Patients with Usher Syndrome Type II. American Journal of Human Genetics, 2004, 74, 738-744.	2.6	176
27	Molecular genetics of Leber congenital amaurosis. Human Molecular Genetics, 2002, 11, 1169-1176.	1.4	175
28	L1 retrotransposition can occur early in human embryonic development. Human Molecular Genetics, 2007, 16, 1587-1592.	1.4	174
29	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
30	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
31	The DFNB31 gene product whirlin connects to the Usher protein network in the cochlea and retina by direct association with USH2A and VLGR1. Human Molecular Genetics, 2006, 15, 751-765.	1.4	162
32	Clinical and Genetic Characteristics of Late-onset Stargardt's Disease. Ophthalmology, 2012, 119, 1199-1210.	2.5	162
33	CRB1 mutation spectrum in inherited retinal dystrophies. Human Mutation, 2004, 24, 355-369.	1.1	159
34	Genotyping Microarray (Disease Chip) for Leber Congenital Amaurosis: Detection of Modifier Alleles. , 2005, 46, 3052.		153
35	Progressive Loss of Cones in Achromatopsia: An Imaging Study Using Spectral-Domain Optical Coherence Tomography. , 2010, 51, 5952.		149
36	A Nonsense Mutation in PDE6H Causes Autosomal-Recessive Incomplete Achromatopsia. American Journal of Human Genetics, 2012, 91, 527-532.	2.6	148

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37	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
38	Norrie disease is caused by mutations in an extracellular protein resembling Câ€“terminal globular domain of mucins. Nature Genetics, 1992, 2, 139-143.	9.4	145
39	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
40	Identification of a 2 Mb Human Ortholog of Drosophila eyes shut/spacemaker that Is Mutated in Patients with Retinitis Pigmentosa. American Journal of Human Genetics, 2008, 83, 594-603.	2.6	141
41	Identification of a hot spot for microdeletions in patients with X- linked deafness type 3 (DFN3) 900 kb proximal to the DFN3 gene POU3F4. Human Molecular Genetics, 1996, 5, 1229-1235.	1.4	138
42	Mutations inGRM6Cause Autosomal Recessive Congenital Stationary Night Blindness with a Distinctive Scotopic 15-Hz Flicker Electroretinogram. , 2005, 46, 4328.		136
43	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	1.4	136
44	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
45	Cloning and characterization of the human choroideremia gene. Human Molecular Genetics, 1994, 3, 1041-1046.	1.4	131
46	Mutations of theCEP290gene encoding a centrosomal protein cause Meckel-Gruber syndrome. Human Mutation, 2008, 29, 45-52.	1.1	131
47	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	2.6	127
48	Causes and consequences of inherited cone disorders. Progress in Retinal and Eye Research, 2014, 42, 1-26.	7.3	127
49	Early-Onset Stargardt Disease. Ophthalmology, 2015, 122, 335-344.	2.5	127
50	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
51	Mutations in the candidate gene for Norrie disease. Human Molecular Genetics, 1992, 1, 461-465.	1.4	126
52	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
53	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
54	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	1.1	118

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55	Non-syndromic retinal ciliopathies: translating gene discovery into therapy. Human Molecular Genetics, 2012, 21, R111-R124.	1.4	117
56	Interaction of nephrocystin-4 and RPGRIP1 is disrupted by nephronophthisis or Leber congenital amaurosis-associated mutations. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 18520-18525.	3.3	116
57	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
58	ABCR unites what ophthalmologists divide(s). Ophthalmic Genetics, 1998, 19, 117-122.	0.5	115
59	Clinical Course, Genetic Etiology, and Visual Outcome in Cone and Coneâ€“Rod Dystrophy. Ophthalmology, 2012, 119, 819-826.	2.5	115
60	A Novel Ribosomal S6-Kinase (RSK4; RPS6KA6) Is Commonly Deleted in Patients with Complex X-Linked Mental Retardation. Genomics, 1999, 62, 332-343.	1.3	113
61	Towards understanding CRUMBS function in retinal dystrophies. Human Molecular Genetics, 2006, 15, R235-R243.	1.4	112
62	Genetic Etiology and Clinical Consequences of Complete and Incomplete Achromatopsia. Ophthalmology, 2009, 116, 1984-1989.e1.	2.5	112
63	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
64	Mutations in the peripherin/RDS gene are an important cause of multifocal pattern dystrophy simulating STGD1/fundus flavimaculatus. British Journal of Ophthalmology, 2007, 91, 1504-1511.	2.1	110
65	Intein-mediated protein trans-splicing expands adeno-associated virus transfer capacity in the retina. Science Translational Medicine, 2019, 11, .	5.8	109
66	IQCB1Mutations in Patients with Leber Congenital Amaurosis. , 2011, 52, 834.		107
67	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
68	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
69	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	1.4	104
70	The spectrum of retinal phenotypes caused by mutations in the ABCA4 gene. Graefe's Archive for Clinical and Experimental Ophthalmology, 2005, 243, 90-100.	1.0	103
71	Genetic testing for retinal dystrophies and dysfunctions: benefits, dilemmas and solutions. Clinical and Experimental Ophthalmology, 2007, 35, 473-485.	1.3	103
72	Physical fine mapping of the choroideremia locus using Xq21 deletions associated with complex syndromes. Genomics, 1989, 4, 41-46.	1.3	101

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73	Basal Laminar Drusen Caused by Compound Heterozygous Variants in the CFH Gene. American Journal of Human Genetics, 2008, 82, 516-523.	2.6	99
74	Isolation of Crb1, a mouse homologue of Drosophila crumbs, and analysis of its expression pattern in eye and brain. Mechanisms of Development, 2002, 110, 203-207.	1.7	98
75	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
76	Novel types of mutation in the choroideremia (CHM) gene: a full-length L1 insertion and an intronic mutation activating a cryptic exon. Human Genetics, 2003, 113, 268-275.	1.8	96
77	Microarray-based mutation analysis of the ABCA4 (ABCR) gene in autosomal recessive coneâ€“rod dystrophy and retinitis pigmentosa. European Journal of Human Genetics, 2004, 12, 1024-1032.	1.4	96
78	Photoreceptor Progenitor mRNA Analysis Reveals Exon Skipping Resulting from the ABCA4 c.5461-10Tâ†'C Mutation in Stargardt Disease. Ophthalmology, 2016, 123, 1375-1385.	2.5	96
79	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
80	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	1.5	94
81	Central Areolar Choroidal Dystrophy. Ophthalmology, 2009, 116, 771-782.e1.	2.5	94
82	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
83	Composition and function of the Crumbs protein complex in the mammalian retina. Experimental Eye Research, 2008, 86, 713-726.	1.2	93
84	Homozygosity Mapping in Patients with Coneâ€“Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
85	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
86	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
87	Mutation analysis of 272 Spanish families affected by autosomal recessive retinitis pigmentosa using a genotyping microarray. Molecular Vision, 2010, 16, 2550-8.	1.1	91
88	Identification of Novel Mutations in Patients with Leber Congenital Amaurosis and Juvenile RP by Genome-wide Homozygosity Mapping with SNP Microarrays. , 2007, 48, 5690.		90
89	Clinical and Genetic Heterogeneity in Multifocal Vitelliform Dystrophy. JAMA Ophthalmology, 2007, 125, 1100.	2.6	88
90	Mutations in C2ORF71 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 783-788.	2.6	88

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91	Microarray-Based Mutation Detection and Phenotypic Characterization of Patients with Leber Congenital Amaurosis. , 2006, 47, 1167.		86
92	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
93	Aberrant splicing of the CHM gene is a significant cause of choroideremia. Nature Genetics, 1992, 1, 109-113.	9.4	82
94	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
95	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2012, 53, 4458.		81
96	A Novel Mutation in theELOVL4Gene Causes Autosomal Dominant Stargardt-like Macular Dystrophy. , 2004, 45, 4263.		79
97	Erosive Vitreoretinopathy and Wagner Disease Are Caused by Intronic Mutations inCSPG2/VersicanThat Result in an Imbalance of Splice Variants. , 2006, 47, 3565.		77
98	A Novel Homozygous Nonsense Mutation inCABP4Causes Congenital Coneâ€“Rod Synaptic Disorder. , 2009, 50, 2344.		76
99	The spectrum of phenotypes caused by variants in the CFH gene. Molecular Immunology, 2009, 46, 1573-1594.	1.0	76
100	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
101	Genotypic and Phenotypic Characteristics of CRB1 -Associated Retinal Dystrophies. Ophthalmology, 2017, 124, 884-895.	2.5	75
102	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
103	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
104	A Novel Mutation Identified in the DFNA5 Gene in a Dutch Family: A Clinical and Genetic Evaluation. Audiology and Neuro-Otology, 2004, 9, 34-46.	0.6	74
105	An autosomal homologue of the choroideremia gene colocalizes with the usher syndrome type II locus on the distal part of chromosome 1q. Human Molecular Genetics, 1992, 1, 71-75.	1.4	73
106	Mutations in the EYS Gene Account for Approximately 5% of Autosomal Recessive Retinitis Pigmentosa and Cause a Fairly Homogeneous Phenotype. Ophthalmology, 2010, 117, 2026-2033.e7.	2.5	72
107	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	2.6	71
108	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

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109	Mutation spectrum in the CHM gene of Danish and Swedish choroideremia patients. Human Molecular Genetics, 1994, 3, 1047-1051.	1.4	69
110	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
111	Characterization of the Crumbs homolog 2 (CRB2) gene and analysis of its role in retinitis pigmentosa and Leber congenital amaurosis. Molecular Vision, 2005, 11, 263-73.	1.1	69
112	Clinical and Molecular Evaluation of Probands and Family Members with Familial Exudative Vitreoretinopathy. , 2009, 50, 4379.		68
113	Heterozygous Deep-Intronic Variants and Deletions inABCA4in Persons with Retinal Dystrophies and One ExonicABCA4Variant. Human Mutation, 2015, 36, 43-47.	1.1	68
114	Further mutations in Brain 4 (POU3F4) clarify the phenotype in the X-linked deafness, DFN3. Human Molecular Genetics, 1995, 4, 1467-1469.	1.4	67
115	Mouse Choroideremia Gene Mutation Causes Photoreceptor Cell Degeneration and is not Transmitted through the Female Germline. Human Molecular Genetics, 1997, 6, 851-858.	1.4	67
116	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
117	The CommonABCA4Variant p.Asn1868Ile Shows Nonpenetrance and Variable Expression of Stargardt Disease When Present intransWith Severe Variants. , 2018, 59, 3220.		67
118	A Novel Nonsense Mutation inCEP290Induces Exon Skipping and Leads to a Relatively Mild Retinal Phenotype. , 2010, 51, 3646.		65
119	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
120	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
121	Choroideremia: Variability of Clinical and Electrophysiological Characteristics and First Report of a Negative Electroretinogram. Ophthalmology, 2006, 113, 2066-2073.e2.	2.5	63
122	MPP5 Recruits MPP4 to the CRB1 Complex in Photoreceptors. , 2005, 46, 2192.		62
123	Missense mutations inPOU4F3cause autosomal dominant hearing impairment DFNA15 and affect subcellular localization and DNA binding. Human Mutation, 2008, 29, 545-554.	1.1	62
124	CLINICAL AND MOLECULAR GENETIC ANALYSIS OF BEST VITELLIFORM MACULAR DYSTROPHY. Retina, 2009, 29, 835-847.	1.0	62
125	Comprehensive Analysis of the Achromatopsia Genes CNGA3 and CNGB3 in Progressive Cone Dystrophy. Ophthalmology, 2010, 117, 825-830.e1.	2.5	61
126	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2011, 118, 1444-1448.	2.5	61

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127	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
128	Foveal Sparing in Stargardt Disease. , 2014, 55, 7467.		60
129	Mutations in MFSD8, Encoding a Lysosomal Membrane Protein, Are Associated with Nonsyndromic Autosomal Recessive Macular Dystrophy. Ophthalmology, 2015, 122, 170-179.	2.5	60
130	Benchmarking deep learning splice prediction tools using functional splice assays. Human Mutation, 2021, 42, 799-810.	1.1	59
131	Mutations in thelipoma HMGIC fusion partner-like 5 (LHFPL5)gene cause autosomal recessive nonsyndromic hearing loss. Human Mutation, 2006, 27, 633-639.	1.1	58
132	Involvement ofDFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. Human Mutation, 2007, 28, 718-723.	1.1	58
133	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
134	Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotypeâ€“Phenotype Correlations, and Inheritance Models. Genes, 2018, 9, 215.	1.0	58
135	Novel Null Mutations in theEYSGene Are a Frequent Cause of Autosomal Recessive Retinitis Pigmentosa in the Israeli Population. , 2010, 51, 4387.		57
136	Mutations in the Mevalonate Kinase (MVK) Gene Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2013, 120, 2697-2705.	2.5	56
137	CLINICAL AND GENETIC CHARACTERISTICS OF MALE PATIENTS WITH RPGR-ASSOCIATED RETINAL DYSTROPHIES. Retina, 2019, 39, 1186-1199.	1.0	56
138	FERM protein EPB41L5 is a novel member of the mammalian CRBâ€“MPP5 polarity complex. Experimental Cell Research, 2007, 313, 3959-3970.	1.2	55
139	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	2.3	55
140	Unexpected CEP290 mRNA Splicing in a Humanized Knock-In Mouse Model for Leber Congenital Amaurosis. PLoS ONE, 2013, 8, e79369.	1.1	55
141	Two human Î³-crystallin genes are linked and riddled with Alu-repeats. Gene, 1985, 38, 197-204.	1.0	52
142	Progressive Cochleovestibular Impairment Caused by a Point Mutation in theCOCHGene at DFNA9. Laryngoscope, 1999, 109, 1525-1530.	1.1	51
143	Phenotypic spectrum of autosomal recessive cone-rod dystrophies caused by mutations in the ABCA4 (ABCR) gene. Investigative Ophthalmology and Visual Science, 2002, 43, 1980-5.	3.3	51
144	USH2A Mutation analysis in 70 Dutch families with Usher syndrome type II. Human Mutation, 2004, 24, 185-185.	1.1	50

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145	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
146	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	9.4	50
147	Three families displaying the combination of Stargardt's disease with coneâ€“rod dystrophy or retinitis pigmentosa. Ophthalmology, 2004, 111, 546-553.	2.5	49
148	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
149	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
150	Choroideremia gene product affects trophoblast development and vascularization in mouse extra-embryonic tissues. Developmental Biology, 2004, 272, 53-65.	0.9	46
151	Identification of splice defects due to noncanonical splice site or deepâ€intronic variants in ABCA4. Human Mutation, 2019, 40, 2365-2376.	1.1	46
152	Identification of novel mutations in X-linked retinitis pigmentosa families and implications for diagnostic testing. Molecular Vision, 2008, 14, 1081-93.	1.1	46
153	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. European Journal of Human Genetics, 2002, 10, 197-203.	1.4	45
154	Identification and molecular modelling of a mutation in the motor head domain of myosin VIIA in a family with autosomal dominant hearing impairment (DFNA11). Human Genetics, 2004, 115, 149-156.	1.8	45
155	Isolation and Mapping of Novel Candidate Genes for Retinal Disorders Using Suppression Subtractive Hybridization. Genomics, 1999, 58, 240-249.	1.3	44
156	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. Ophthalmology, 2014, 121, 1620-1627.	2.5	44
157	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
158	Exclusion mapping of the X-linked dominant chondrodysplasia punctata/ichthyosis/cataract/short stature (Happle) syndrome: possible involvement of an unstable pre-mutation. Human Genetics, 1992, 89, 659-665.	1.8	43
159	Mutations in the calcium-binding motifs of CDH23 and the 35delG mutation in GJB2 cause hearing loss in one family. Human Genetics, 2003, 112, 156-163.	1.8	43
160	Usher syndrome and Leber congenital amaurosis are molecularly linked via a novel isoform of the centrosomal ninein-like protein. Human Molecular Genetics, 2009, 18, 51-64.	1.4	43
161	A common ancestor for COCH related cochleovestibular (DFNA9) patients in Belgium and The Netherlands bearing the P51S mutation. Journal of Medical Genetics, 2001, 38, 61-65.	1.5	43
162	Speech Recognition Scores Related to Age and Degree of Hearing Impairment in DFNA2/KCNQ4 and DFNA9/COCH. JAMA Otolaryngology, 2001, 127, 1045.	1.5	42

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