Qingjiong Zhang

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Bietti Crystalline Corneoretinal Dystrophy Is Caused by Mutations in the Novel Gene CYP4V2. American Journal of Human Genetics, 2004, 74, 817-826.	6.2	237
2	Mutations of 60 known causative genes in 157 families with retinitis pigmentosa based on exome sequencing. Human Genetics, 2014, 133, 1255-1271.	3.8	148
3	Functions of the intermediate filament cytoskeleton in the eye lens. Journal of Clinical Investigation, 2009, 119, 1837-1848.	8.2	142
4	Mitochondrial DNA Haplogroups M7b1′2 and M8a Affect Clinical Expression of Leber Hereditary Optic Neuropathy in Chinese Families with the m.11778G→A Mutation. American Journal of Human Genetics, 2008, 83, 760-768.	6.2	124
5	Genetic Variants at 13q12.12 Are Associated with High Myopia in the Han Chinese Population. American Journal of Human Genetics, 2011, 88, 805-813.	6.2	106
6	Detection of Variants in 15 Genes in 87 Unrelated Chinese Patients with Leber Congenital Amaurosis. PLoS ONE, 2011, 6, e19458.	2.5	101
7	Mutations in \hat{I}^2 B3-Crystallin Associated with Autosomal Recessive Cataract in Two Pakistani Families. , 2005, 46, 2100.		97
8	Severe retinitis pigmentosa mapped to 4p15 and associated with a novel mutation in the PROM1 gene. Human Genetics, 2007, 122, 293-299.	3.8	97
9	Detection of Mutations in LRPAP1, CTSH, LEPREL1, ZNF644, SLC39A5, and SCO2 in 298 Families With Early-Onset High Myopia by Exome Sequencing. Investigative Ophthalmology and Visual Science, 2015, 56, 339-345.	3.3	96
10	Comprehensive Mutation Analysis by Whole-Exome Sequencing in 41 Chinese Families With Leber Congenital Amaurosis. , 2013, 54, 4351.		93
11	Novel locus for X linked recessive high myopia maps to Xq23-q25 but outside MYP1. Journal of Medical Genetics, 2005, 43, e20-e20.	3.2	88
12	Molecular epidemiology of mtDNA mutations in 903 Chinese families suspected with Leber hereditary optic neuropathy. Journal of Human Genetics, 2006, 51, 851-856.	2.3	87
13	Retinitis Pigmentosa. Asia-Pacific Journal of Ophthalmology, 2016, 5, 265-271.	2.5	79
14	An overview of myopia genetics. Experimental Eye Research, 2019, 188, 107778.	2.6	79
15	A new locus for autosomal dominant high myopia maps to 4q22-q27 between D4S1578 and D4S1612. Molecular Vision, 2005, 11, 554-60.	1.1	79
16	Exome Sequencing on 298 Probands With Early-Onset High Myopia: Approximately One-Fourth Show Potential Pathogenic Mutations in RetNet Genes. , 2015, 56, 8365.		77
17	Trio-based exome sequencing arrests de novo mutations in early-onset high myopia. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 4219-4224.	7.1	77
18	Identification of CYP4V2 mutation in 21 families and overview of mutation spectrum in Bietti crystalline corneoretinal dystrophy. Biochemical and Biophysical Research Communications, 2011, 409, 181-186.	2.1	64

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19	Evaluation of 12 Myopia-Associated Genes in Chinese Patients With High Myopia. Investigative Ophthalmology and Visual Science, 2015, 56, 722-729.	3.3	60
20	Molecular genetics of cone-rod dystrophy in Chinese patients: New data from 61 probands and mutation overview of 163 probands. Experimental Eye Research, 2016, 146, 252-258.	2.6	60
21	<i>KIF11</i> mutations are a common cause of autosomal dominant familial exudative vitreoretinopathy. British Journal of Ophthalmology, 2016, 100, 278-283.	3.9	59
22	Negatively regulating TLR4/NF-κB signaling via PPARα in endotoxin-induced uveitis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1109-1120.	3.8	54
23	An Ophthalmic Targeted Exome Sequencing Panel as a Powerful Tool to Identify Causative Mutations in Patients Suspected of Hereditary Eye Diseases. Translational Vision Science and Technology, 2019, 8, 21.	2.2	54
24	Protective effect of lutein on ARPE-19 cells upon H2O2-induced G2/M arrest. Molecular Medicine Reports, 2017, 16, 2069-2074.	2.4	53
25	Strikingly different penetrance of LHON in two Chinese families with primary mutation G11778A is independent of mtDNA haplogroup background and secondary mutation G13708A. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2008, 643, 48-53.	1.0	52
26	Exome Sequencing of 47 Chinese Families with Cone-Rod Dystrophy: Mutations in 25 Known Causative Genes. PLoS ONE, 2013, 8, e65546.	2.5	52
27	The De-Ubiquitinylating Enzyme, USP2, Is Associated with the Circadian Clockwork and Regulates Its Sensitivity to Light. PLoS ONE, 2011, 6, e25382.	2.5	51
28	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
29	Mutation Analysis of Seven Known Glaucoma-Associated Genes in Chinese Patients With Glaucoma. , 2014, 55, 3594.		50
30	Insights into the beaded filament of the eye lens. Experimental Cell Research, 2007, 313, 2180-2188.	2.6	49
31	Protective effects of tetramethylpyrazine on rat retinal cell cultures. Neurochemistry International, 2008, 52, 1176-1187.	3.8	47
32	Exome Sequencing of 18 Chinese Families with Congenital Cataracts: A New Sight of the NHS Gene. PLoS ONE, 2014, 9, e100455.	2.5	47
33	Unique Variants in <i>OPN1LW</i> Cause Both Syndromic and Nonsyndromic X-Linked High Myopia Mapped to MYP1. , 2015, 56, 4150.		46
34	Mutation analysis of 12 genes in Chinese families with congenital cataracts. Molecular Vision, 2011, 17, 2197-206.	1.1	45
35	Replication Study SupportsCTNND2as a Susceptibility Gene for High Myopia. , 2011, 52, 8258.		44
36	CRX variants in cone–rod dystrophy and mutation overview. Biochemical and Biophysical Research Communications, 2012, 426, 498-503.	2.1	44

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37	High Myopia Is Not Associated with the SNPs in theTGIF, Lumican,TGFB1, andHGFGenes. , 2009, 50, 1546.		43
38	m ⁶ A methyltransferase METTL3 promotes retinoblastoma progression via PI3K/AKT/mTOR pathway. Journal of Cellular and Molecular Medicine, 2020, 24, 12368-12378.	3.6	42
39	Upregulation of hepatic VLDLR via PPARα is required for the triglyceride-lowering effect of fenofibrate. Journal of Lipid Research, 2014, 55, 1622-1633.	4.2	40
40	Clinical and linkage study on a consanguineous Chinese family with autosomal recessive high myopia. Molecular Vision, 2009, 15, 312-8.	1.1	40
41	Mitochondrial DNA Haplogroup Background Affects LHON, but Not Suspected LHON, in Chinese Patients. PLoS ONE, 2011, 6, e27750.	2.5	39
42	Association of the single nucleotide polymorphisms in the extracellular matrix metalloprotease-9 gene with PACG in southern China. Molecular Vision, 2009, 15, 1412-7.	1.1	39
43	Mitochondrial DNA haplogroup distribution in Chaoshanese with and without myopia. Molecular Vision, 2010, 16, 303-9.	1.1	39
44	The 208delG Mutation inFSCN2Does Not Associate with Retinal Degeneration in Chinese Individuals. , 2007, 48, 530.		38
45	Novel SOX2 Mutation Associated With Ocular Coloboma in a Chinese Family. JAMA Ophthalmology, 2008, 126, 709.	2.4	37
46	Insight into the molecular genetics of myopia. Molecular Vision, 2017, 23, 1048-1080.	1.1	37
47	Frequent mutations of RetNet genes in eoHM: Further confirmation in 325 probands and comparison with late-onset high myopia based on exome sequencing. Experimental Eye Research, 2018, 171, 76-91.	2.6	36
48	Genetics of Refraction and Myopia. Progress in Molecular Biology and Translational Science, 2015, 134, 269-279.	1.7	34
49	Mutations in NYX of individuals with high myopia, but without night blindness. Molecular Vision, 2007, 13, 330-6.	1.1	34
50	A New Locus for Autosomal Recessive Nuclear Cataract Mapped to Chromosome 19q13 in a Pakistani Family. , 2005, 46, 623.		33
51	Nonsyndromic High Myopia in a Chinese Family Mapped to MYP1. JAMA Ophthalmology, 2010, 128, 1473.	2.4	33
52	Evaluation of the X-linked modifier loci for Leber hereditary optic neuropathy with the G11778A mutation in Chinese. Molecular Vision, 2010, 16, 416-24.	1.1	33
53	Exome sequencing identified null mutations in LOXL3 associated with early-onset high myopia. Molecular Vision, 2016, 22, 161-7.	1.1	31
54	Confirmation of a genetic locus for X-linked recessive high myopia outside MYP1. Journal of Human Genetics, 2007, 52, 469-472.	2.3	30

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55	Molecular genetics of Leber congenital amaurosis in Chinese: New data from 66 probands and mutation overview of 159 probands. Experimental Eye Research, 2016, 149, 93-99.	2.6	30
56	Generation and Characterization of Induced Pluripotent Stem Cells and Retinal Organoids From a Leber's Congenital Amaurosis Patient With Novel RPE65 Mutations. Frontiers in Molecular Neuroscience, 2019, 12, 212.	2.9	30
57	Identification of <i>CNGA3</i> Mutations in 46 Families. JAMA Ophthalmology, 2014, 132, 1076.	2.5	29
58	X-linked heterozygous mutations in cause female-limited early onset high myopia. Molecular Vision, 2016, 22, 1257-1266.	1.1	29
59	Clinical description and genome wide linkage study of Y-sutural cataract and myopia in a Chinese family. Molecular Vision, 2004, 10, 890-900.	1.1	29
60	Severe autosomal recessive retinitis pigmentosa maps to chromosome 1p13.3–p21.2 between D1S2896 and D1S457 but outside ABCA4. Human Genetics, 2005, 118, 356-365.	3.8	28
61	Exome sequencing reveals CHM mutations in six families with atypical choroideremia initially diagnosed as retinitis pigmentosa. International Journal of Molecular Medicine, 2014, 34, 573-577.	4.0	28
62	Mutational screening of six genes in Chinese patients with congenital cataract and microcornea. Molecular Vision, 2011, 17, 1508-13.	1.1	28
63	Identification of FZD4 and LRP5 mutations in 11 of 49 families with familial exudative vitreoretinopathy. Molecular Vision, 2012, 18, 2438-46.	1.1	28
64	The MT-ND1 and MT-ND5 genes are mutational hotspots for Chinese families with clinical features of LHON but lacking the three primary mutations. Biochemical and Biophysical Research Communications, 2010, 399, 179-185.	2.1	27
65	A novel locus for autosomal dominant congenital motor nystagmus mapped to 1q31-q32.2 between D1S2816 and D1S2692. Human Genetics, 2012, 131, 697-702.	3.8	27
66	CPSF1 mutations are associated with early-onset high myopia and involved in retinal ganglion cell axon projection. Human Molecular Genetics, 2019, 28, 1959-1970.	2.9	27
67	Novel TSPAN12 mutations in patients with familial exudative vitreoretinopathy and their associated phenotypes. Molecular Vision, 2011, 17, 1128-35.	1.1	27
68	Germline Mutations in CTNNB1 Associated With Syndromic FEVR or Norrie Disease. , 2019, 60, 93.		26
69	FRMD7 mutations in Chinese families with X-linked congenital motor nystagmus. Molecular Vision, 2007, 13, 1375-8.	1.1	26
70	Novel truncation mutations in MYRF cause autosomal dominant high hyperopia mapped to 11p12–q13.3. Human Genetics, 2019, 138, 1077-1090.	3.8	25
71	Mutational screening of 10 genes in Chinese patients with microphthalmia and/or coloboma. Molecular Vision, 2009, 15, 2911-8.	1.1	25
72	Screening for CRX gene mutations in Chinese patients with Leber congenital amaurosis and mutational phenotype. Ophthalmic Genetics, 2001, 22, 89-96.	1.2	24

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73	Mutation spectrum and frequency of the RHO gene in 248 Chinese families with retinitis pigmentosa. Biochemical and Biophysical Research Communications, 2010, 401, 42-47.	2.1	24
74	<i><scp>ALMS1</scp></i> null mutations: a common cause of Leber congenital amaurosis and earlyâ€onset severe cone–rod dystrophy. Clinical Genetics, 2016, 89, 442-447.	2.0	24
75	A recurrent mutation in GUCY2D associated with autosomal dominant cone dystrophy in a Chinese family. Molecular Vision, 2011, 17, 3271-8.	1.1	24
76	Microphthalmia, late onset keratitis, and iris coloboma/aniridia in a family with a novel <i>PAX6</i> mutation. Ophthalmic Genetics, 2012, 33, 119-121.	1.2	23
77	Mitochondrial DNA Sequence Variation and Haplogroup Distribution in Chinese Patients with LHON and m.14484T>C. PLoS ONE, 2010, 5, e13426.	2.5	23
78	A variant form of Oguchi disease mapped to 13q34 associated with partial deletion of GRK1 gene. Molecular Vision, 2005, 11, 977-85.	1.1	23
79	Co-occurrence of A1555G and G11778A in a Chinese family with high penetrance of Leber's hereditary optic neuropathy. Biochemical and Biophysical Research Communications, 2008, 376, 221-224.	2.1	22
80	Molecular characterization of six Chinese families with m.3460G>A and Leber hereditary optic neuropathy. Neurogenetics, 2010, 11, 349-356.	1.4	22
81	Novel <i><scp>BEST</scp>1</i> mutations and special clinical characteristics of autosomal recessive bestrophinopathy in Chinese patients. Acta Ophthalmologica, 2019, 97, 247-259.	1.1	22
82	<i><scp>RPE</scp>65</i> mutation frequency and phenotypic variation according to exome sequencing in a tertiary centre for genetic eye diseases in China. Acta Ophthalmologica, 2020, 98, e181-e190.	1.1	21
83	Novel GPR143 mutations and clinical characteristics in six Chinese families with X-linked ocular albinism. Molecular Vision, 2008, 14, 1974-82.	1.1	21
84	Mutation analysis in 129 genes associated with other forms of retinal dystrophy in 157 families with retinitis pigmentosa based on exome sequencing. Molecular Vision, 2015, 21, 477-86.	1.1	21
85	Linkage analysis of two families with X-linked recessive congenital motor nystagmus. Journal of Human Genetics, 2006, 51, 76-80.	2.3	20
86	No association between the SNPs (rs3749446 and rs1402000) in the PARL gene and LHON in Chinese patients with m.11778G>A. Human Genetics, 2010, 128, 465-468.	3.8	20
87	Evaluation of the ELOVL4, PRPH2 and ABCA4 genes in patients with Stargardt macular degeneration. Molecular Medicine Reports, 2012, 6, 1045-1049.	2.4	20
88	Blockage of Notch Signaling Inhibits the Migration and Proliferation of Retinal Pigment Epithelial Cells. Scientific World Journal, The, 2013, 2013, 1-6.	2.1	20
89	Mutation analysis of the genes associated with anterior segment dysgenesis, microcornea and microphthalmia in 257 patients with glaucoma. International Journal of Molecular Medicine, 2015, 36, 1111-1117.	4.0	20
90	Evaluation of PRSS56 in Chinese subjects with high hyperopia or primary angle-closure glaucoma. Molecular Vision, 2013, 19, 2217-26.	1.1	20

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91	Mutations in <i>RPGR</i> and <i>RP2</i> of Chinese Patients with X-Linked Retinitis Pigmentosa. Current Eye Research, 2010, 35, 73-79.	1.5	19
92	ID2 protects retinal pigment epithelium cells from oxidative damage through p-ERK1/2/ID2/NRF2. Archives of Biochemistry and Biophysics, 2018, 650, 1-13.	3.0	19
93	Identification of Genetic Defects in 33 Probands with Stargardt Disease by WES-Based Bioinformatics Gene Panel Analysis. PLoS ONE, 2015, 10, e0132635.	2.5	19
94	Cerulean cataract mapped to 12q13 and associated with a novel initiation codon mutation in MIP. Molecular Vision, 2011, 17, 2049-55.	1.1	19
95	Association of markers at chromosome 15q14 in Chinese patients with moderate to high myopia. Molecular Vision, 2012, 18, 2633-46.	1.1	19
96	Complete mitochondrial DNA genome sequence variation of Chinese families with mutation m.3635G>A and Leber hereditary optic neuropathy. Molecular Vision, 2012, 18, 3087-94.	1.1	19
97	Pathogenicity discrimination and genetic test reference for CRX variants based on genotype-phenotype analysis. Experimental Eye Research, 2019, 189, 107846.	2.6	18
98	Screening for variants in 20 genes in 130 unrelated patients with cone-rod dystrophy. Molecular Medicine Reports, 2013, 7, 1779-1785.	2.4	17
99	Flat Anterior Chamber after Trabeculectomy in Secondary Angle-Closure Glaucoma with BEST1 Gene Mutation: Case Series. PLoS ONE, 2017, 12, e0169395.	2.5	17
100	Notch signaling pathway mediates Doxorubicin-driven apoptosis in cancers. Cancer Management and Research, 2018, Volume 10, 1439-1448.	1.9	17
101	Structural variations in a non-coding region at 1q32.1 are responsible for the NYS7 locus in two large families. Human Genetics, 2020, 139, 1057-1064.	3.8	17
102	Sequence variations of GRM6 in patients with high myopia. Molecular Vision, 2009, 15, 2094-100.	1.1	17
103	Autosomal recessive retinitis pigmentosa in a Pakistani family mapped to CNGA1 with identification of a novel mutation. Molecular Vision, 2004, 10, 884-9.	1.1	17
104	CSNB1 in Chinese families associated with novel mutations in NYX. Journal of Human Genetics, 2006, 51, 634-640.	2.3	16
105	Mitochondrial DNA mutation m.3635G>A may be associated with Leber hereditary optic neuropathy in Chinese. Biochemical and Biophysical Research Communications, 2009, 386, 392-395.	2.1	16
106	Novel GUCA1A mutation identified in a Chinese family with cone-rod dystrophy. Neuroscience Letters, 2013, 541, 179-183.	2.1	16
107	Biallelic mutations in <i>USP45,</i> encoding a deubiquitinating enzyme, are associated with Leber congenital amaurosis. Journal of Medical Genetics, 2019, 56, 325-331.	3.2	16
108	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.	2.5	16

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109	Mutation survey of the optic atrophy 1 gene in 193 Chinese families with suspected hereditary optic neuropathy. Molecular Vision, 2013, 19, 292-302.	1.1	16
110	Iris hyperpigmentation in a Chinese family with ocular albinism and the <i>GPR143</i> mutation. American Journal of Medical Genetics, Part A, 2009, 149A, 1786-1788.	1.2	15
111	Screening for <i>NDP</i> Mutations in 44 Unrelated Patients with Familial Exudative Vitreoretinopathy or Norrie Disease. Current Eye Research, 2012, 37, 726-729.	1.5	15
112	Mutation spectrum of PAX6 in Chinese patients with aniridia. Molecular Vision, 2011, 17, 2139-47.	1.1	15
113	Phenotypic characterization of patients with early-onset high myopia due to mutations in or : Why not Stickler syndrome?. Molecular Vision, 2018, 24, 560-573.	1.1	15
114	<i>PAX6</i> Mutations Identified in 4 of 35 Families with Microcornea. , 2012, 53, 6338.		14
115	Mitochondrial DNA mutation m.10680G > A is associated with Leber hereditary optic neuropathy in Chinese patients. Journal of Translational Medicine, 2012, 10, 43.	4.4	14
116	Common variants in chromosome 4q25 are associated with myopia in Chinese adults. Ophthalmic and Physiological Optics, 2012, 32, 68-73.	2.0	14
117	PTEN Reduced UVB-Mediated Apoptosis in Retinal Pigment Epithelium Cells. BioMed Research International, 2017, 2017, 1-11.	1.9	14
118	Spectrum-frequency and genotype–phenotype analysis of rhodopsin variants. Experimental Eye Research, 2021, 203, 108405.	2.6	14
119	mtDNA m.3635G>A may be classified as a common primary mutation for Leber hereditary optic neuropathy in the Chinese population. Biochemical and Biophysical Research Communications, 2010, 403, 237-241.	2.1	13
120	Is Mitochondrial tRNAphe Variant m.593T>C a Synergistically Pathogenic Mutation in Chinese LHON Families with m.11778G>A?. PLoS ONE, 2011, 6, e26511.	2.5	13
121	Cone-Rod Dysfunction Is a Sign of Early-Onset High Myopia. Optometry and Vision Science, 2013, 90, 1327-1330.	1.2	13
122	Mutation analysis of Leber congenital amaurosis-associated genes in patients with retinitis pigmentosa. Molecular Medicine Reports, 2015, 11, 1827-1832.	2.4	13
123	Evaluation of EGR1 as a candidate gene for high myopia. Molecular Vision, 2008, 14, 1309-12.	1.1	13
124	Further evidence of autosomal-dominant Leber congenital amaurosis caused by heterozygous CRX mutation. Graefe's Archive for Clinical and Experimental Ophthalmology, 2007, 245, 1401-1402.	1.9	12
125	KIF21A novel deletion and recurrent mutation in patients with congenital fibrosis of the extraocular muscles-1. International Journal of Molecular Medicine, 2011, 28, 973-5.	4.0	12
126	PRIMPOL Mutation: Functional Study Does Not Always Reveal the Truth. Investigative Ophthalmology and Visual Science, 2015, 56, 1181-1182.	3.3	12

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127	The OPA1 Gene Mutations Are Frequent in Han Chinese Patients with Suspected Optic Neuropathy. Molecular Neurobiology, 2017, 54, 1622-1630.	4.0	12
128	A novel truncation mutation in GJA1 associated with open angle glaucoma and microcornea in a large Chinese family. Eye, 2015, 29, 972-977.	2.1	11
129	Anti-tumor Drug THZ1 Suppresses TGFβ2-mediated EMT in Lens Epithelial Cells via Notch and TGFβ/Smad Signaling Pathway. Journal of Cancer, 2019, 10, 3778-3788.	2.5	11
130	Evaluation of MFRP as a candidate gene for high hyperopia. Molecular Vision, 2009, 15, 181-6.	1.1	11
131	mtDNA haplogroup distribution in Chinese patients with Leber's hereditary optic neuropathy and G11778A mutation. Biochemical and Biophysical Research Communications, 2007, 364, 238-242.	2.1	10
132	Does the Association Between <i>TMEM98</i> and Nanophthalmos Require Further Confirmation?. JAMA Ophthalmology, 2015, 133, 358.	2.5	10
133	Comparative exome sequencing reveals novel candidate genes for retinitis pigmentosa. EBioMedicine, 2020, 56, 102792.	6.1	10
134	Clinical and Genetic Analysis of 63 Families Demonstrating Early and Advanced Characteristic Fundus as the Signature of CRB1 Mutations. American Journal of Ophthalmology, 2021, 223, 160-168.	3.3	10
135	DNAH17 is essential for rat spermatogenesis and fertility. Journal of Genetics, 2021, 100, 1.	0.7	10
136	Genotype–Phenotype Analysis of RPGR Variations: Reporting of 62 Chinese Families and a Literature Review. Frontiers in Genetics, 2021, 12, 600210.	2.3	10
137	Replication study of significant single nucleotide polymorphisms associated with myopia from two genome-wide association studies. Molecular Vision, 2011, 17, 3290-9.	1.1	10
138	Identification and characterization of novel alternative splice variants of human SAMD11. Gene, 2013, 530, 215-221.	2.2	9
139	Detection of CRB1 mutations in families with retinal dystrophy through phenotype-oriented mutational screening. International Journal of Molecular Medicine, 2014, 33, 913-918.	4.0	9
140	Identification of MFRP Mutations in Chinese Families with High Hyperopia. Optometry and Vision Science, 2016, 93, 19-26.	1.2	9
141	The inhibition of NOTCH2 reduces UVB-induced damage in retinal pigment epithelium cells. Molecular Medicine Reports, 2017, 16, 730-736.	2.4	9
142	Rare variants in novel and known genes associated with primary angle closure glaucoma based on whole exome sequencing of 549 probands. Journal of Genetics and Genomics, 2019, 46, 353-357.	3.9	9
143	Diseases associated with mutations in CNGA3: Genotype–phenotype correlation and diagnostic guideline. Progress in Molecular Biology and Translational Science, 2019, 161, 1-27.	1.7	9
144	Mutation screening of TRPM1, GRM6, NYX and CACNA1F genes in patients with congenital stationary night blindness. International Journal of Molecular Medicine, 2012, 30, 521-526.	4.0	8

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145	DNA methyltransferase inhibitor CDA-II inhibits myogenic differentiation. Biochemical and Biophysical Research Communications, 2012, 422, 522-526.	2.1	8
146	Covalent CDK7 Inhibitor THZ1 Inhibits Myogenic Differentiation. Journal of Cancer, 2018, 9, 3149-3155.	2.5	8
147	Evaluation of the genetic association between early-onset primary angle-closure glaucoma and retinitis pigmentosa. Experimental Eye Research, 2020, 197, 108118.	2.6	8
148	An evaluation of OPTC and EPYC as candidate genes for high myopia. Molecular Vision, 2009, 15, 2045-9.	1.1	8
149	Late-onset CORD in a patient with <i>RDH12</i> mutations identified by whole exome sequencing. Ophthalmic Genetics, 2016, 37, 345-348.	1.2	7
150	A novel missense mutation of <i>GJA8</i> causes congenital cataract in a large Mauritanian family. European Journal of Ophthalmology, 2019, 29, 621-628.	1.3	7
151	A novel deep intronic COL2A1 mutation in a family with earlyâ€onset high myopia/ocularâ€only Stickler syndrome. Ophthalmic and Physiological Optics, 2020, 40, 281-288.	2.0	7
152	Pathogenic variants and associated phenotypic spectrum of TSPAN12 based on data from a large cohort. Graefe's Archive for Clinical and Experimental Ophthalmology, 2021, 259, 2929-2939.	1.9	7
153	Novel variants in GUCY2D causing retinopathy and the genotype-phenotype correlation. Experimental Eye Research, 2021, 208, 108637.	2.6	7
154	Novel RS1 mutations associated with X-linked juvenile retinoschisis. International Journal of Molecular Medicine, 2012, 29, 644-648.	4.0	6
155	Abnormal expression of seven myogenesis-related genes in extraocular muscles of patients with concomitant strabismus. Molecular Medicine Reports, 2013, 7, 217-222.	2.4	6
156	A novel variant in <i>IDH3A</i> identified in a case with Leber congenital amaurosis accompanied by macular pseudocoloboma. Ophthalmic Genetics, 2018, 39, 662-663.	1.2	6
157	Variants in <i>RCBTB1</i> are Associated with Autosomal Recessive Retinitis Pigmentosa but Not Autosomal Dominant FEVR. Current Eye Research, 2021, 46, 839-844.	1.5	6
158	Dominant RP in the Middle While Recessive in Both the N- and C-Terminals Due to RP1 Truncations: Confirmation, Refinement, and Questions. Frontiers in Cell and Developmental Biology, 2021, 9, 634478.	3.7	6
159	Pathogenicity evaluation and the genotype–phenotype analysis of OPA1 variants. Molecular Genetics and Genomics, 2021, 296, 845-862.	2.1	6
160	Characterization of <i>PROM1</i> p.Arg373Cys Variant in a Cohort of Chinese Patients: Macular Dystrophy Plus Peripheral Bone-Spicule Degeneration. , 2021, 62, 19.		6
161	Biallelic variants in <i>CPAMD8</i> are associated with primary open-angle glaucoma and primary angle-closure glaucoma. British Journal of Ophthalmology, 2022, 106, 1710-1715.	3.9	6

162 Genetics of Pathologic Myopia. , 2021, , 43-58.

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163	Clinical and genetic features of retinoschisis in 120 families with <i>RS1</i> mutations. British Journal of Ophthalmology, 2023, 107, 367-372.	3.9	6
164	Genotypes and phenotypes of genes associated with achromatopsia: A reference for clinical genetic testing. Molecular Vision, 2020, 26, 588-602.	1.1	6
165	Different Phenotypes Represent Advancing Stages of <i>ABCA4</i> -Associated Retinopathy: A Longitudinal Study of 212 Chinese Families From a Tertiary Center. , 2022, 63, 28.		6
166	RGR variants in different forms of retinal diseases: The undetermined role of truncation mutations. Molecular Medicine Reports, 2016, 14, 4811-4815.	2.4	5
167	Genotype-Phenotype of Isolated Foveal Hypoplasia in a Large Cohort: Minor Iris Changes as an Indicator of PAX6 Involvement. , 2021, 62, 23.		5
168	Lack of phenotypic effect of triallelic variation in SPATA7 in a family with Leber congenital amaurosis resulting from CRB1 mutations. Molecular Vision, 2011, 17, 3326-32.	1.1	5
169	Cataracts, ataxia, short stature, and mental retardation in a Chinese family mapped to Xpter-q13.1. Journal of Human Genetics, 2006, 51, 695-700.	2.3	4
170	High myopia is not associated with single nucleotide polymorphisms in the COL2A1 gene in the Chinese population. Molecular Medicine Reports, 2011, 5, 133-7.	2.4	4
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