

# Qingjiong Zhang

## List of Publications by Year in descending order

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193  
papers

5,275  
citations

101384

36  
h-index

149479

56  
g-index

196  
all docs

196  
docs citations

196  
times ranked

5058  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical and genetic features of retinoschisis in 120 families with <i>RS1</i> mutations. <i>British Journal of Ophthalmology</i> , 2023, 107, 367-372.	2.1	6
2	Biallelic variants in <i>CPAMD8</i> are associated with primary open-angle glaucoma and primary angle-closure glaucoma. <i>British Journal of Ophthalmology</i> , 2022, 106, 1710-1715.	2.1	6
3	Missense mutation in the <i>PAX6</i> gene can cause a complex mild variable phenotype predominated by concomitant strabismus. <i>Ophthalmic Genetics</i> , 2022, 43, 88-96.	0.5	1
4	Clinical features and genetic spectrum of <i>NMNAT1</i> -associated retinal degeneration. <i>Eye</i> , 2022, 36, 2279-2285.	1.1	3
5	Landscape of pathogenic variants in six pre-mRNA processing factor genes for retinitis pigmentosa based on large in-house data sets and database comparisons. <i>Acta Ophthalmologica</i> , 2022, , .	0.6	4
6	Autosomal Dominant Retinitis Pigmentosa-Associated <i>TOPORS</i> Protein Truncating Variants Are Exclusively Located in the Region of Amino Acid Residues 807 to 867. , 2022, 63, 19.		1
7	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
8	Spectrum-frequency and genotype-phenotype analysis of rhodopsin variants. <i>Experimental Eye Research</i> , 2021, 203, 108405.	1.2	14
9	Clinical and Genetic Analysis of 63 Families Demonstrating Early and Advanced Characteristic Fundus as the Signature of <i>CRB1</i> Mutations. <i>American Journal of Ophthalmology</i> , 2021, 223, 160-168.	1.7	10
10	Variants in <i>RCBTB1</i> are Associated with Autosomal Recessive Retinitis Pigmentosa but Not Autosomal Dominant FEVR. <i>Current Eye Research</i> , 2021, 46, 839-844.	0.7	6
11	Genotype-Phenotype of <i>RPE65</i> Mutations: A Reference Guide for Gene Testing and Its Clinical Application. <i>Essentials in Ophthalmology</i> , 2021, , 181-196.	0.0	0
12	Dominant RP in the Middle While Recessive in Both the N- and C-Terminals Due to <i>RP1</i> Truncations: Confirmation, Refinement, and Questions. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 634478.	1.8	6
13	<i>DNAH17</i> is essential for rat spermatogenesis and fertility. <i>Journal of Genetics</i> , 2021, 100, 1.	0.4	10
14	Systemic Genotype-Phenotype Analysis of <i>MYOC</i> Variants Based on Exome Sequencing and Literature Review. <i>Asia-Pacific Journal of Ophthalmology</i> , 2021, 10, 173-182.	1.3	2
15	Pathogenicity evaluation and the genotype-phenotype analysis of <i>OPA1</i> variants. <i>Molecular Genetics and Genomics</i> , 2021, 296, 845-862.	1.0	6
16	Pathogenic variants and associated phenotypic spectrum of <i>TSPAN12</i> based on data from a large cohort. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2021, 259, 2929-2939.	1.0	7
17	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
18	Start and End with Genetics: <i>RCBTB1</i> and Beyond. <i>Current Eye Research</i> , 2021, 46, 1932-1933.	0.7	1

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19	Genotype-Phenotype Analysis of RPGR Variations: Reporting of 62 Chinese Families and a Literature Review. <i>Frontiers in Genetics</i> , 2021, 12, 600210.	1.1	10
20	An Early Diagnostic Clue for COL18A1- and LAMA1-Associated Diseases: High Myopia With Alopecia Areata in the Cranial Midline. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 644947.	1.8	1
21	Novel variants in GUCY2D causing retinopathy and the genotype-phenotype correlation. <i>Experimental Eye Research</i> , 2021, 208, 108637.	1.2	7
22	Genotype-Phenotype of Isolated Foveal Hypoplasia in a Large Cohort: Minor Iris Changes as an Indicator of PAX6 Involvement. , 2021, 62, 23.		5
23	Severe exudative vitreoretinopathy as a common feature for CTNNA1, KIF11 and NDP variants plus sector degeneration for KIF11. <i>American Journal of Ophthalmology</i> , 2021, , .	1.7	2
24	Genetics of Pathologic Myopia. , 2021, , 43-58.		6
25	Confirming and expanding the phenotypes of variants: Coloboma, inferior chorioretinal hypoplasia, and high myopia. <i>Molecular Vision</i> , 2021, 27, 50-60.	1.1	1
26	Heterozygous variants with ocular phenotype: Missense in domain but truncation out of domain. <i>Molecular Vision</i> , 2021, 27, 309-322.	1.1	0
27	Novel BMP4 Truncations Resulted in Opposite Ocular Anomalies: Pathologic Myopia Rather Than Microphthalmia. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 769636.	1.8	2
28	is essential for rat spermatogenesis and fertility. <i>Journal of Genetics</i> , 2021, 100, .	0.4	1
29	<i>rs112076656</i> mutation frequency and phenotypic variation according to exome sequencing in a tertiary centre for genetic eye diseases in China. <i>Acta Ophthalmologica</i> , 2020, 98, e181-e190.	0.6	21
30	<i>rs112076656</i> A methyltransferase METTL3 promotes retinoblastoma progression via PI3K/AKT/mTOR pathway. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 12368-12378.	1.6	42
31	Comparative exome sequencing reveals novel candidate genes for retinitis pigmentosa. <i>EBioMedicine</i> , 2020, 56, 102792.	2.7	10
32	Evaluation of the genetic association between early-onset primary angle-closure glaucoma and retinitis pigmentosa. <i>Experimental Eye Research</i> , 2020, 197, 108118.	1.2	8
33	A novel deep intronic COL2A1 mutation in a family with early-onset high myopia/ocular-only Stickler syndrome. <i>Ophthalmic and Physiological Optics</i> , 2020, 40, 281-288.	1.0	7
34	Structural variations in a non-coding region at 1q32.1 are responsible for the NYS7 locus in two large families. <i>Human Genetics</i> , 2020, 139, 1057-1064.	1.8	17
35	Heterozygous structural variation mimicking homozygous missense mutations in NEU1 associated with presenting clinical signs in eyes alone. <i>Ophthalmic Genetics</i> , 2020, 41, 279-283.	0.5	2
36	Curcumin Inhibits Proliferation and Epithelial-Mesenchymal Transition in Lens Epithelial Cells through Multiple Pathways. <i>BioMed Research International</i> , 2020, 2020, 1-11.	0.9	4

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37	Genotypes and phenotypes of genes associated with achromatopsia: A reference for clinical genetic testing. <i>Molecular Vision</i> , 2020, 26, 588-602.	1.1	6
38	Correspondence to Rossetti et al.'s review of the phenotypic spectrum associated with haploinsufficiency of <i>MYRF</i> . <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2315-2316.	0.7	2
39	Anti-tumor Drug THZ1 Suppresses TGF $\beta$ 2-mediated EMT in Lens Epithelial Cells via Notch and TGF $\beta$ 2/Smad Signaling Pathway. <i>Journal of Cancer</i> , 2019, 10, 3778-3788.	1.2	11
40	Rare variants in novel and known genes associated with primary angle closure glaucoma based on whole exome sequencing of 549 probands. <i>Journal of Genetics and Genomics</i> , 2019, 46, 353-357.	1.7	9
41	Pathogenicity discrimination and genetic test reference for CRX variants based on genotype-phenotype analysis. <i>Experimental Eye Research</i> , 2019, 189, 107846.	1.2	18
42	Generation and Characterization of Induced Pluripotent Stem Cells and Retinal Organoids From a Leber's Congenital Amaurosis Patient With Novel RPE65 Mutations. <i>Frontiers in Molecular Neuroscience</i> , 2019, 12, 212.	1.4	30
43	An overview of myopia genetics. <i>Experimental Eye Research</i> , 2019, 188, 107778.	1.2	79
44	CPSF1 mutations are associated with early-onset high myopia and involved in retinal ganglion cell axon projection. <i>Human Molecular Genetics</i> , 2019, 28, 1959-1970.	1.4	27
45	Diseases associated with mutations in CNGA3: Genotype-phenotype correlation and diagnostic guideline. <i>Progress in Molecular Biology and Translational Science</i> , 2019, 161, 1-27.	0.9	9
46	An Ophthalmic Targeted Exome Sequencing Panel as a Powerful Tool to Identify Causative Mutations in Patients Suspected of Hereditary Eye Diseases. <i>Translational Vision Science and Technology</i> , 2019, 8, 21.	1.1	54
47	Novel truncation mutations in MYRF cause autosomal dominant high hyperopia mapped to 11p12-q13.3. <i>Human Genetics</i> , 2019, 138, 1077-1090.	1.8	25
48	Biallelic mutations in <i>USP45</i> , encoding a deubiquitinating enzyme, are associated with Leber congenital amaurosis. <i>Journal of Medical Genetics</i> , 2019, 56, 325-331.	1.5	16
49	Novel <i>BEST1</i> mutations and special clinical characteristics of autosomal recessive bestrophinopathy in Chinese patients. <i>Acta Ophthalmologica</i> , 2019, 97, 247-259.	0.6	22
50	Germline Mutations in CTNNB1 Associated With Syndromic FEVR or Norrie Disease. , 2019, 60, 93.		26
51	A novel missense mutation of <i>GJA8</i> causes congenital cataract in a large Mauritanian family. <i>European Journal of Ophthalmology</i> , 2019, 29, 621-628.	0.7	7
52	Mutation profile of glaucoma candidate genes in Mauritanian families with primary congenital glaucoma. <i>Molecular Vision</i> , 2019, 25, 373-381.	1.1	3
53	Spectrum, frequency, and genotype-phenotype of mutations in. <i>Molecular Vision</i> , 2019, 25, 821-833.	1.1	2
54	Frequent mutations of RetNet genes in eoHM: Further confirmation in 325 probands and comparison with late-onset high myopia based on exome sequencing. <i>Experimental Eye Research</i> , 2018, 171, 76-91.	1.2	36

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55	Covalent CDK7 Inhibitor THZ1 Inhibits Myogenic Differentiation. <i>Journal of Cancer</i> , 2018, 9, 3149-3155.	1.2	8
56	ID2 protects retinal pigment epithelium cells from oxidative damage through p-ERK1/2/ID2/NRF2. <i>Archives of Biochemistry and Biophysics</i> , 2018, 650, 1-13.	1.4	19
57	A novel variant in <i>IDH3A</i> identified in a case with Leber congenital amaurosis accompanied by macular pseudocoloboma. <i>Ophthalmic Genetics</i> , 2018, 39, 662-663.	0.5	6
58	Notch signaling pathway mediates Doxorubicin-driven apoptosis in cancers. <i>Cancer Management and Research</i> , 2018, Volume 10, 1439-1448.	0.9	17
59	Phenotypic characterization of patients with early-onset high myopia due to mutations in <i>COL1A1</i> : Why not Stickler syndrome?. <i>Molecular Vision</i> , 2018, 24, 560-573.	1.1	15
60	The OPA1 Gene Mutations Are Frequent in Han Chinese Patients with Suspected Optic Neuropathy. <i>Molecular Neurobiology</i> , 2017, 54, 1622-1630.	1.9	12
61	The inhibition of NOTCH2 reduces UVB-induced damage in retinal pigment epithelium cells. <i>Molecular Medicine Reports</i> , 2017, 16, 730-736.	1.1	9
62	Trio-based exome sequencing arrests de novo mutations in early-onset high myopia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 4219-4224.	3.3	77
63	PTEN Reduced UVB-Mediated Apoptosis in Retinal Pigment Epithelium Cells. <i>BioMed Research International</i> , 2017, 2017, 1-11.	0.9	14
64	Protective effect of lutein on ARPE-19 cells upon H <sub>2</sub> O <sub>2</sub> -induced G <sub>2</sub> /M arrest. <i>Molecular Medicine Reports</i> , 2017, 16, 2069-2074.	1.1	53
65	Flat Anterior Chamber after Trabeculectomy in Secondary Angle-Closure Glaucoma with BEST1 Gene Mutation: Case Series. <i>PLoS ONE</i> , 2017, 12, e0169395.	1.1	17
66	Genetic Analysis of Families with Retinal Dystrophies. <i>Essentials in Ophthalmology</i> , 2017, , 71-82.	0.0	0
67	Association and interaction of myopia with SNP markers rs13382811 and rs6469937 at and in Han Chinese and European populations. <i>Molecular Vision</i> , 2017, 23, 588-604.	1.1	2
68	Insight into the molecular genetics of myopia. <i>Molecular Vision</i> , 2017, 23, 1048-1080.	1.1	37
69	Identification of MFRP Mutations in Chinese Families with High Hyperopia. <i>Optometry and Vision Science</i> , 2016, 93, 19-26.	0.6	9
70	<i>ALMS1</i> null mutations: a common cause of Leber congenital amaurosis and early-onset severe cone-rod dystrophy. <i>Clinical Genetics</i> , 2016, 89, 442-447.	1.0	24
71	Molecular genetics of cone-rod dystrophy in Chinese patients: New data from 61 probands and mutation overview of 163 probands. <i>Experimental Eye Research</i> , 2016, 146, 252-258.	1.2	60
72	Molecular genetics of Leber congenital amaurosis in Chinese: New data from 66 probands and mutation overview of 159 probands. <i>Experimental Eye Research</i> , 2016, 149, 93-99.	1.2	30

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73	RGR variants in different forms of retinal diseases: The undetermined role of truncation mutations. <i>Molecular Medicine Reports</i> , 2016, 14, 4811-4815.	1.1	5
74	Retinitis Pigmentosa. <i>Asia-Pacific Journal of Ophthalmology</i> , 2016, 5, 265-271.	1.3	79
75	Late-onset CORD in a patient with <i>RDH12</i> mutations identified by whole exome sequencing. <i>Ophthalmic Genetics</i> , 2016, 37, 345-348.	0.5	7
76	<i>KIF11</i> mutations are a common cause of autosomal dominant familial exudative vitreoretinopathy. <i>British Journal of Ophthalmology</i> , 2016, 100, 278-283.	2.1	59
77	Exome sequencing identified null mutations in <i>LOXL3</i> associated with early-onset high myopia. <i>Molecular Vision</i> , 2016, 22, 161-7.	1.1	31
78	X-linked heterozygous mutations in cause female-limited early onset high myopia. <i>Molecular Vision</i> , 2016, 22, 1257-1266.	1.1	29
79	Mutation analysis of Leber congenital amaurosis-associated genes in patients with retinitis pigmentosa. <i>Molecular Medicine Reports</i> , 2015, 11, 1827-1832.	1.1	13
80	PRIMPOL Mutation: Functional Study Does Not Always Reveal the Truth. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 1181-1182.	3.3	12
81	Exome Sequencing on 298 Proband With Early-Onset High Myopia: Approximately One-Fourth Show Potential Pathogenic Mutations in RetNet Genes. , 2015, 56, 8365.		77
82	Unique Variants in <i>OPN1LW</i> Cause Both Syndromic and Nonsyndromic X-Linked High Myopia Mapped to MYP1. , 2015, 56, 4150.		46
83	A novel truncation mutation in <i>GJA1</i> associated with open angle glaucoma and microcornea in a large Chinese family. <i>Eye</i> , 2015, 29, 972-977.	1.1	11
84	Does the Association Between <i>TMEM98</i> and Nanophthalmos Require Further Confirmation?. <i>JAMA Ophthalmology</i> , 2015, 133, 358.	1.4	10
85	Mutation analysis of the genes associated with anterior segment dysgenesis, microcornea and microphthalmia in 257 patients with glaucoma. <i>International Journal of Molecular Medicine</i> , 2015, 36, 1111-1117.	1.8	20
86	Detection of Mutations in <i>LRPAP1</i> , <i>CTSH</i> , <i>LEPREL1</i> , <i>ZNF644</i> , <i>SLC39A5</i> , and <i>SCO2</i> in 298 Families With Early-Onset High Myopia by Exome Sequencing. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 339-345.	3.3	96
87	Evaluation of 12 Myopia-Associated Genes in Chinese Patients With High Myopia. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 722-729.	3.3	60
88	Genetics of Refraction and Myopia. <i>Progress in Molecular Biology and Translational Science</i> , 2015, 134, 269-279.	0.9	34
89	Identification of Genetic Defects in 33 Proband with Stargardt Disease by WES-Based Bioinformatics Gene Panel Analysis. <i>PLoS ONE</i> , 2015, 10, e0132635.	1.1	19
90	Mutation analysis in 129 genes associated with other forms of retinal dystrophy in 157 families with retinitis pigmentosa based on exome sequencing. <i>Molecular Vision</i> , 2015, 21, 477-86.	1.1	21

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91	Exome Sequencing of 18 Chinese Families with Congenital Cataracts: A New Sight of the NHS Gene. PLoS ONE, 2014, 9, e100455.	1.1	47
92	Identification of CNGA3 Mutations in 46 Families. JAMA Ophthalmology, 2014, 132, 1076.	1.4	29
93	Mutation Analysis of Seven Known Glaucoma-Associated Genes in Chinese Patients With Glaucoma. , 2014, 55, 3594.		50
94	Upregulation of hepatic VLDLR via PPAR $\alpha$ is required for the triglyceride-lowering effect of fenofibrate. Journal of Lipid Research, 2014, 55, 1622-1633.	2.0	40
95	Detection of CRB1 mutations in families with retinal dystrophy through phenotype-oriented mutational screening. International Journal of Molecular Medicine, 2014, 33, 913-918.	1.8	9
96	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.	1.8	28
97	Negatively regulating TLR4/NF- $\kappa$ B signaling via PPAR $\alpha$ in endotoxin-induced uveitis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1109-1120.	1.8	54
98	Mutations of 60 known causative genes in 157 families with retinitis pigmentosa based on exome sequencing. Human Genetics, 2014, 133, 1255-1271.	1.8	148
99	Overexpression of Six1 leads to retardation of myogenic differentiation in C2C12 myoblasts. Molecular Biology Reports, 2013, 40, 217-223.	1.0	4
100	Identification and characterization of novel alternative splice variants of human SAMD11. Gene, 2013, 530, 215-221.	1.0	9
101	Novel GUCA1A mutation identified in a Chinese family with cone-rod dystrophy. Neuroscience Letters, 2013, 541, 179-183.	1.0	16
102	Screening for variants in 20 genes in 130 unrelated patients with cone-rod dystrophy. Molecular Medicine Reports, 2013, 7, 1779-1785.	1.1	17
103	Comprehensive Mutation Analysis by Whole-Exome Sequencing in 41 Chinese Families With Leber Congenital Amaurosis. , 2013, 54, 4351.		93
104	Abnormal expression of seven myogenesis-related genes in extraocular muscles of patients with concomitant strabismus. Molecular Medicine Reports, 2013, 7, 217-222.	1.1	6
105	Cone-Rod Dysfunction Is a Sign of Early-Onset High Myopia. Optometry and Vision Science, 2013, 90, 1327-1330.	0.6	13
106	Exome Sequencing of 47 Chinese Families with Cone-Rod Dystrophy: Mutations in 25 Known Causative Genes. PLoS ONE, 2013, 8, e65546.	1.1	52
107	Blockage of Notch Signaling Inhibits the Migration and Proliferation of Retinal Pigment Epithelial Cells. Scientific World Journal, The, 2013, 2013, 1-6.	0.8	20
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.	1.1	16

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109	Mutation survey of the optic atrophy 1 gene in 193 Chinese families with suspected hereditary optic neuropathy. <i>Molecular Vision</i> , 2013, 19, 292-302.	1.1	16
110	Evaluation of PRSS56 in Chinese subjects with high hyperopia or primary angle-closure glaucoma. <i>Molecular Vision</i> , 2013, 19, 2217-26.	1.1	20
111	<i>PAX6</i> Mutations Identified in 4 of 35 Families with Microcornea. , 2012, 53, 6338.		14
112	Evaluation of the ELOVL4, PRPH2 and ABCA4 genes in patients with Stargardt macular degeneration. <i>Molecular Medicine Reports</i> , 2012, 6, 1045-1049.	1.1	20
113	Confirmation and refinement of an autosomal dominant congenital motor nystagmus locus in chromosome 1q31.3â€“q32.1. <i>Journal of Human Genetics</i> , 2012, 57, 756-759.	1.1	4
114	Novel RS1 mutations associated with X-linked juvenile retinoschisis. <i>International Journal of Molecular Medicine</i> , 2012, 29, 644-648.	1.8	6
115	Mutation screening of TRPM1, GRM6, NYX and CACNA1F genes in patients with congenital stationary night blindness. <i>International Journal of Molecular Medicine</i> , 2012, 30, 521-526.	1.8	8
116	Microphthalmia, late onset keratitis, and iris coloboma/aniridia in a family with a novel <i>PAX6</i> mutation. <i>Ophthalmic Genetics</i> , 2012, 33, 119-121.	0.5	23
117	DNA methyltransferase inhibitor CDA-II inhibits myogenic differentiation. <i>Biochemical and Biophysical Research Communications</i> , 2012, 422, 522-526.	1.0	8
118	Mitochondrial DNA mutation m.10680G > A is associated with Leber hereditary optic neuropathy in Chinese patients. <i>Journal of Translational Medicine</i> , 2012, 10, 43.	1.8	14
119	Screening for <i>NDP</i> Mutations in 44 Unrelated Patients with Familial Exudative Vitreoretinopathy or Norrie Disease. <i>Current Eye Research</i> , 2012, 37, 726-729.	0.7	15
120	CRX variants in coneâ€“rod dystrophy and mutation overview. <i>Biochemical and Biophysical Research Communications</i> , 2012, 426, 498-503.	1.0	44
121	A novel locus for autosomal dominant congenital motor nystagmus mapped to 1q31-q32.2 between D1S2816 and D1S2692. <i>Human Genetics</i> , 2012, 131, 697-702.	1.8	27
122	Common variants in chromosome 4q25 are associated with myopia in Chinese adults. <i>Ophthalmic and Physiological Optics</i> , 2012, 32, 68-73.	1.0	14
123	Identification of FZD4 and LRP5 mutations in 11 of 49 families with familial exudative vitreoretinopathy. <i>Molecular Vision</i> , 2012, 18, 2438-46.	1.1	28
124	Association of markers at chromosome 15q14 in Chinese patients with moderate to high myopia. <i>Molecular Vision</i> , 2012, 18, 2633-46.	1.1	19
125	Complete mitochondrial DNA genome sequence variation of Chinese families with mutation m.3635G>A and Leber hereditary optic neuropathy. <i>Molecular Vision</i> , 2012, 18, 3087-94.	1.1	19
126	Identification of CYP4V2 mutation in 21 families and overview of mutation spectrum in Bietti crystalline corneoretinal dystrophy. <i>Biochemical and Biophysical Research Communications</i> , 2011, 409, 181-186.	1.0	64

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127	The De-Ubiquitinating Enzyme, USP2, Is Associated with the Circadian Clockwork and Regulates Its Sensitivity to Light. <i>PLoS ONE</i> , 2011, 6, e25382.	1.1	51
128	Is Mitochondrial tRNA <sup>phe</sup> Variant m.593T>C a Synergistically Pathogenic Mutation in Chinese LHON Families with m.11778G>A?. <i>PLoS ONE</i> , 2011, 6, e26511.	1.1	13
129	KIF21A novel deletion and recurrent mutation in patients with congenital fibrosis of the extraocular muscles-1. <i>International Journal of Molecular Medicine</i> , 2011, 28, 973-5.	1.8	12
130	High myopia is not associated with single nucleotide polymorphisms in the COL2A1 gene in the Chinese population. <i>Molecular Medicine Reports</i> , 2011, 5, 133-7.	1.1	4
131	Genetic Variants at 13q12.12 Are Associated with High Myopia in the Han Chinese Population. <i>American Journal of Human Genetics</i> , 2011, 88, 805-813.	2.6	106
132	Replication Study Supports CTNND2 as a Susceptibility Gene for High Myopia. , 2011, 52, 8258.		44
133	Detection of Variants in 15 Genes in 87 Unrelated Chinese Patients with Leber Congenital Amaurosis. <i>PLoS ONE</i> , 2011, 6, e19458.	1.1	101
134	Mitochondrial DNA Haplogroup Background Affects LHON, but Not Suspected LHON, in Chinese Patients. <i>PLoS ONE</i> , 2011, 6, e27750.	1.1	39
135	Novel TSPAN12 mutations in patients with familial exudative vitreoretinopathy and their associated phenotypes. <i>Molecular Vision</i> , 2011, 17, 1128-35.	1.1	27
136	Mutational screening of six genes in Chinese patients with congenital cataract and microcornea. <i>Molecular Vision</i> , 2011, 17, 1508-13.	1.1	28
137	Cerulean cataract mapped to 12q13 and associated with a novel initiation codon mutation in MIP. <i>Molecular Vision</i> , 2011, 17, 2049-55.	1.1	19
138	Mutation spectrum of PAX6 in Chinese patients with aniridia. <i>Molecular Vision</i> , 2011, 17, 2139-47.	1.1	15
139	Mutation analysis of 12 genes in Chinese families with congenital cataracts. <i>Molecular Vision</i> , 2011, 17, 2197-206.	1.1	45
140	A recurrent mutation in GUCY2D associated with autosomal dominant cone dystrophy in a Chinese family. <i>Molecular Vision</i> , 2011, 17, 3271-8.	1.1	24
141	Replication study of significant single nucleotide polymorphisms associated with myopia from two genome-wide association studies. <i>Molecular Vision</i> , 2011, 17, 3290-9.	1.1	10
142	Lack of phenotypic effect of triallelic variation in SPATA7 in a family with Leber congenital amaurosis resulting from CRB1 mutations. <i>Molecular Vision</i> , 2011, 17, 3326-32.	1.1	5
143	Molecular characterization of six Chinese families with m.3460G>A and Leber hereditary optic neuropathy. <i>Neurogenetics</i> , 2010, 11, 349-356.	0.7	22
144	No association between the SNPs (rs3749446 and rs1402000) in the PARL gene and LHON in Chinese patients with m.11778G>A. <i>Human Genetics</i> , 2010, 128, 465-468.	1.8	20

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145	Mutations in <i>RPGR</i> and <i>RP2</i> of Chinese Patients with X-Linked Retinitis Pigmentosa. <i>Current Eye Research</i> , 2010, 35, 73-79.	0.7	19
146	Nonsyndromic High Myopia in a Chinese Family Mapped to MYP1. <i>JAMA Ophthalmology</i> , 2010, 128, 1473.	2.6	33
147	The MT-ND1 and MT-ND5 genes are mutational hotspots for Chinese families with clinical features of LHON but lacking the three primary mutations. <i>Biochemical and Biophysical Research Communications</i> , 2010, 399, 179-185.	1.0	27
148	Mutation spectrum and frequency of the RHO gene in 248 Chinese families with retinitis pigmentosa. <i>Biochemical and Biophysical Research Communications</i> , 2010, 401, 42-47.	1.0	24
149	mtDNA m.3635G>A may be classified as a common primary mutation for Leber hereditary optic neuropathy in the Chinese population. <i>Biochemical and Biophysical Research Communications</i> , 2010, 403, 237-241.	1.0	13
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