Ruifang Sui

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5582555/publications.pdf

Version: 2024-02-01

64 papers	1,263 citations	471509 17 h-index	434195 31 g-index
66 all docs	66 docs citations	66 times ranked	4053 citing authors

#	Article	IF	Citations
1	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Human Genetics, 2014, 133, 331-345.	3.8	204
2	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.	13.7	91
3	Comprehensive Molecular Diagnosis of a Large Chinese Leber Congenital Amaurosis Cohort. , 2015, 56, 3642.		82
4	Mutations in human IFT140 cause non-syndromic retinal degeneration. Human Genetics, 2015, 134, 1069-1078.	3.8	62
5	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
6	Comprehensive molecular diagnosis of 67 Chinese Usher syndrome probands: high rate of ethnicity specific mutations in Chinese USH patients. Orphanet Journal of Rare Diseases, 2015, 10, 110.	2.7	47
7	Mutations inPOMGNT1cause non-syndromic retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 1479-1488.	2.9	42
8	CEP78is mutated in a distinct type of Usher syndrome. Journal of Medical Genetics, 2017, 54, 190-195.	3.2	42
9	Comprehensive analysis of patients with Stargardt macular dystrophy reveals new genotypeâ€“phenotype correlations and unexpected diagnostic revisions. Genetics in Medicine, 2015, 17, 262-270.	2.4	41
10	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. American Journal of Human Genetics, 2016, 99, 470-480.	6.2	39
11	Bacteriaâ€Targeting Photodynamic Nanoassemblies for Efficient Treatment of Multidrugâ€Resistant Biofilm Infected Keratitis. Advanced Functional Materials, 2022, 32, .	14.9	36
12	Clinical and Genetic Characteristics of East Asian Patients with Occult Macular Dystrophy (Miyake) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	5.2	28
13	Leveraging spliceâ€affecting variant predictors and a minigene validation system to identify Mendelian diseaseâ€causing variants among exonâ€captured variants of uncertain significance. Human Mutation, 2017, 38, 1521-1533.	2.5	27
14	A Homozygous Missense Mutation in NEUROD1 Is Associated With Nonsyndromic Autosomal Recessive Retinitis Pigmentosa. Investigative Ophthalmology and Visual Science, 2015, 56, 150-155.	3.3	25
15	Disrupted intraflagellar transport due to IFT74 variants causes Joubert syndrome. Genetics in Medicine, 2021, 23, 1041-1049.	2.4	25
16	USH2A variants in Chinese patients with Usher syndrome type II and non-syndromic retinitis pigmentosa. British Journal of Ophthalmology, 2021, 105, 694-703.	3.9	22
17	Improved Diagnosis of Inherited Retinal Dystrophies by High-Fidelity PCR of ORF15 followed by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 817-824.	2.8	21
18	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21

#	Article	IF	Citations
19	Novel CNGA3 mutations in Chinese patients with achromatopsia. British Journal of Ophthalmology, 2015, 99, 571-576.	3.9	18
20	PHENOTYPIC VARIABILITY OF RECESSIVE RDH12-ASSOCIATED RETINAL DYSTROPHY. Retina, 2019, 39, 2040-2052.	1.7	18
21	TNFRSF21 mutations cause high myopia. Journal of Medical Genetics, 2019, 56, 671-677.	3.2	18
22	Next-generation sequencing-based molecular diagnosis of 12 inherited retinal disease probands of Uyghur ethnicity. Scientific Reports, 2016, 6, 21384.	3.3	17
23	Mutations in crystallin genes result in congenital cataract associated with other ocular abnormalities. Molecular Vision, 2017, 23, 977-986.	1.1	15
24	Transthyretin Ala36Pro mutation in a Chinese pedigree of familial transthyretin amyloidosis with elevated vitreous and serum vascular endothelial growth factor. Experimental Eye Research, 2013, 110, 44-49.	2.6	14
25	Whole-exome sequencing revealed HKDC1 as a candidate gene associated with autosomal-recessive retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 4157-4168.	2.9	14
26	Treating Bietti crystalline dystrophy in a high-fat diet-exacerbated murine model using gene therapy. Gene Therapy, 2020, 27, 370-382.	4.5	14
27	Novel variants in PNPLA6 causing syndromic retinal dystrophy. Experimental Eye Research, 2021, 202, 108327.	2.6	13
28	De novoMutations in the Cone-rod Homeobox Gene Associated with Leber Congenital Amaurosis in Chinese Patients. Ophthalmic Genetics, 2015, 36, 21-26.	1.2	12
29	Whole exome sequencing identified a novel truncation mutation in the NHS gene associated with Nance-Horan syndrome. BMC Medical Genetics, 2019, 20, 14.	2.1	12
30	Molecular genetic and clinical evaluation of three Chinese families with X-linked ocular albinism. Scientific Reports, 2017, 7, 33713.	3.3	11
31	Genetic and phenotypic characteristics of three Mainland Chinese families with choroideremia. Molecular Vision, 2012, 18, 309-16.	1.1	11
32	A novel small deletion in the NHS gene associated with Nance-Horan syndrome. Scientific Reports, 2018, 8, 2398.	3.3	10
33	Ocular Features in Chinese Patients with Blau Syndrome. Ocular Immunology and Inflammation, 2020, 28, 79-85.	1.8	10
34	Juvenile Onset Splenomegaly and Oculopathy Due to Germline Mutation in ALPK1. Journal of Clinical Immunology, 2020, 40, 350-358.	3.8	10
35	Spatial Functional Characteristics of East Asian Patients With Occult Macular Dystrophy (Miyake) Tj ETQq1 1 0.784314 rgBT /Overloc	3.3	10
36	A heterozygous mutation in RPGR associated with Xâ€linked retinitis pigmentosa in a patient with Turner syndrome mosaicism (45,X/46,XX). American Journal of Medical Genetics, Part A, 2018, 176, 214-218.	1.2	9

#	Article	IF	Citations
37	IFT81 as a Candidate Gene for Nonsyndromic Retinal Degeneration. , 2017, 58, 2483-2490.		9
38	Phenotypic characterization of a Chinese family with autosomal dominant coneâ€“rod dystrophy related to GUCY2D. Documenta Ophthalmologica, 2013, 126, 233-240.	2.2	8
39	Rep1 copy number variation is an important genetic cause of choroideremia in Chinese patients. Experimental Eye Research, 2017, 164, 64-73.	2.6	8
40	A novel porcine model reproduces human oculocutaneous albinism type II. Cell Discovery, 2019, 5, 48.	6.7	7
41	Structural modeling, mutation analysis, and in vitro expression of usherin, a major protein in inherited retinal degeneration and hearing loss. Computational and Structural Biotechnology Journal, 2020, 18, 1363-1382.	4.1	7
42	Clinical and genetic analysis of the ABCA4 gene associated retinal dystrophy in a large Chinese cohort. Experimental Eye Research, 2021, 202, 108389.	2.6	7
43	Clinical and genetic features of eight Chinese autosomal-dominant optic atrophy pedigrees with six novelOPA1pathogenic variants. Ophthalmic Genetics, 2018, 39, 569-576.	1.2	6
44	Genetic and clinical characterization of mainland Chinese patients with sialidosis type 1. Molecular Genetics & Genomic Medicine, 2020, 8, e1316.	1.2	6
45	Unilateral retinocytoma associated with a variant in the RB1 gene. Molecular Genetics & Genomic Medicine, 2020, 8, e1156.	1.2	6
46	A novel tandem duplication of PRDM13 in a Chinese family with North Carolina macular dystrophy. Graefe's Archive for Clinical and Experimental Ophthalmology, 2022, 260, 645-653.	1.9	6
47	Variants at codon 838 in the GUCY2D gene result in different phenotypes of cone rod dystrophy. Ophthalmic Genetics, 2020, 41, 548-555.	1.2	5
48	Generation of a human induced pluripotent stem cell line from a Bietti crystalline corneoretinal dystrophy patient with CYP4V2 mutations. Stem Cell Research, 2021, 53, 102330.	0.7	4
49	Clinical and Genetic Characterization of a Chinese Family with CSNB1. Advances in Experimental Medicine and Biology, 2008, 613, 245-252.	1.6	4
50	Genetic architecture of inherited retinal disease. , 2020, , 71-93.		3
51	Detailed comparison of phenotype between male patients carrying variants in exons 1â€“14 and ORF15 of RPGR. Experimental Eye Research, 2020, 198, 108147.	2.6	3
52	CLINICAL CHARACTERISTICS AND MOLECULAR GENETIC ANALYSIS OF A COHORT OF CHINESE PATIENTS WITH CHOROIDEREMIA. Retina, 2020, 40, 2240-2253.	1.7	3
53	Clinical and genetic study on two Chinese families with Wagner vitreoretinopathy. Ophthalmic Genetics, 2020, 41, 432-439.	1.2	3
54	Targeted lipidomics reveals plasmalogen phosphatidylethanolamines and storage triacylglycerols as the major systemic lipid aberrations in Bietti crystalline corneoretinal dystrophy. Journal of Genetics and Genomics, 2022, 49, 380-383.	3.9	3

#	Article	IF	Citations
55	Novel mutation in FBN1 causes ectopia lentis and varicose great saphenous vein in one Chinese autosomal dominant family. Molecular Vision, 2014, 20, 812-21.	1.1	3
56	Visual Field Characteristics in East Asian Patients With Occult Macular Dystrophy (Miyake Disease): EAOMD Report No. 3. , 2022, 63, 12.		3
57	Generation of a human induced pluripotent stem cell line (PUMCHi018-A) from an early-onset severe retinal dystrophy patient with RDH12 mutations. Stem Cell Research, 2022, 59, 102655.	0.7	2
58	Generation of two human induced pluripotent stem cell lines from patients with biallelic USH2A variants. Stem Cell Research, 2021, 55, 102502.	0.7	1
59	Generation of a human induced pluripotent stem cell line PUMCHi019-A from a dominant optic atrophy patient with an OPA1 mutation. Stem Cell Research, 2022, 60, 102705.	0.7	1
60	Retinitis Pigmentosa in China. Essentials in Ophthalmology, 2017, , 105-109.	0.1	0
61	Clinical characterization and the improved molecular diagnosis of autosomal dominant cone-rod dystrophy in patients with SCA7. Molecular Vision, 2021, 27, 221-232.	1.1	0
62	Leber congenital amaurosis as an initial manifestation in a Chinese patient with thiamineâ€responsive megaloblastic anemia syndrome. American Journal of Medical Genetics, Part A, 2021, , .	1.2	0
63	Generation of a human induced pluripotent stem cell line PUMCHi017-A from a Choroideremia patient with CHM mutation. Stem Cell Research, 2022, 59, 102661.	0.7	0
64	Clinical and genetic study of a pseudo-dominant retinoschisis pedigree: the first female patient reported in Chinese population. Ophthalmic Genetics, 2022, , 1-5.	1.2	0