

Shujin Li

List of Publications by Year in descending order

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Version: 2024-02-01

24
papers

352
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759233

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#	ARTICLE	IF	CITATIONS
1	Catenin β 1 mutations cause familial exudative vitreoretinopathy by overactivating Norrin/ β 2-catenin signaling. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	37
2	Exome sequencing revealed Notch ligand JAG1 as a novel candidate gene for familial exudative vitreoretinopathy. <i>Genetics in Medicine</i> , 2020, 22, 77-84.	2.4	34
3	A splicing mutation in aryl hydrocarbon receptor associated with retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2018, 27, 2563-2572.	2.9	29
4	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population. <i>Scientific Reports</i> , 2016, 6, 19432.	3.3	27
5	Association of SNPs in miR-146a, miR-196a2, and miR-499 with the risk of endometrial/ovarian cancer. <i>Acta Biochimica Et Biophysica Sinica</i> , 2015, 47, 564-566.	2.0	23
6	Disruption of Tmem30a results in cerebellar ataxia and degeneration of Purkinje cells. <i>Cell Death and Disease</i> , 2018, 9, 899.	6.3	23
7	Loss of Tmem30a leads to photoreceptor degeneration. <i>Scientific Reports</i> , 2017, 7, 9296.	3.3	22
8	Variants in the Wnt co-receptor LRP6 are associated with familial exudative vitreoretinopathy. <i>Journal of Genetics and Genomics</i> , 2022, 49, 590-594.	3.9	18
9	Whole Exome Sequencing Analysis Identifies Mutations in <i>LRP5</i> in Indian Families with Familial Exudative Vitreoretinopathy. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 346-351.	0.7	17
10	Whole-Exome Sequencing Identified <i>DLG1</i> as a Candidate Gene for Familial Exudative Vitreoretinopathy. <i>Genetic Testing and Molecular Biomarkers</i> , 2021, 25, 309-316.	0.7	17
11	CTNND1 variants cause familial exudative vitreoretinopathy through the Wnt/cadherin axis. <i>JCI Insight</i> , 2022, 7, .	5.0	17
12	The ER membrane protein complex subunit Emc3 controls angiogenesis via the FZD4/WNT signaling axis. <i>Science China Life Sciences</i> , 2021, 64, 1868-1883.	4.9	16
13	Tmem30a Deficiency in endothelial cells impairs cell proliferation and angiogenesis. <i>Journal of Cell Science</i> , 2019, 132, .	2.0	12
14	Novel truncating variants in <i>CTNNB1</i> cause familial exudative vitreoretinopathy. <i>Journal of Medical Genetics</i> , 2023, 60, 174-182.	3.2	10
15	Targeted Next-Generation Sequencing Reveals Novel <i>RP1</i> Mutations in Autosomal Recessive Retinitis Pigmentosa. <i>Genetic Testing and Molecular Biomarkers</i> , 2018, 22, 109-114.	0.7	9
16	Candidate Gene Analysis Identifies Mutations in <i>CYP1B1</i> and <i>LTBP2</i> in Indian Families with Primary Congenital Glaucoma. <i>Genetic Testing and Molecular Biomarkers</i> , 2017, 21, 252-258.	0.7	8
17	Targeted next-generation sequencing reveals that a compound heterozygous mutation in phosphodiesterase 6a gene leads to retinitis pigmentosa in a Chinese family. <i>Ophthalmic Genetics</i> , 2018, 39, 487-491.	1.2	5
18	Targeted Next-Generation Sequencing Reveals a Novel Frameshift Mutation in the <i>MERTK</i> Gene in a Chinese Family with Retinitis Pigmentosa. <i>Genetic Testing and Molecular Biomarkers</i> , 2018, 22, 165-169.	0.7	5

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19	Whole-Exome Sequencing Reveals Novel NDP Variants in X-Linked Familial Exudative Vitreoretinopathy. <i>European Journal of Ophthalmology</i> , 2022, 32, 3220-3226.	1.3	5
20	Heterozygote loss-of-function variants in the <i>LRP5</i> gene cause familial exudative vitreoretinopathy. <i>Clinical and Experimental Ophthalmology</i> , 2022, 50, 441-448.	2.6	5
21	LMBR1L regulates the proliferation and migration of endothelial cells through Norrin/ β 2-catenin signaling. <i>Journal of Cell Science</i> , 2022, 135, .	2.0	5
22	Whole-Exome Sequencing Reveals Novel <i>TSPAN12</i> Variants in Autosomal Dominant Familial Exudative Vitreoretinopathy. <i>Genetic Testing and Molecular Biomarkers</i> , 2021, 25, 399-404.	0.7	4
23	A novel frameshift variant in the <i>TSPAN12</i> gene causes autosomal dominant <i>FEVR</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1949.	1.2	2
24	Identification of Two Novel Variants in the <i>LRP5</i> Gene that Cause Familial Exudative Vitreoretinopathy. <i>Genetic Testing and Molecular Biomarkers</i> , 2022, 26, 146-151.	0.7	1