# Shujin Li

## List of Publications by Year in descending order



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|---|---|---|---|
| 24 papers | 352 citations | 759233 12 h-index | 839539 18 g-index |
| 25 all docs | 25 docs citations | 25 times ranked | 326 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Novel truncating variants inCTNNB1cause familial exudative vitreoretinopathy. Journal of Medical Genetics, 2023, 60, 174-182. | 3.2 | 10 |
| 2 | Whole-Exome Sequencing Reveals Novel NDP Variants in X-Linked Familial Exudative Vitreoretinopathy. European Journal of Ophthalmology, 2022, 32, 3220-3226. | 1.3 | 5 |
| 3 | Heterozygote lossâ€ofâ€function variants in the LRP5 gene cause familial exudative vitreoretinopathy. Clinical and Experimental Ophthalmology, 2022, 50, 441-448. | 2.6 | 5 |
| 4 | LMBR1L regulates the proliferation and migration of endothelial cells through Norrin/Î²-catenin signaling. Journal of Cell Science, 2022, 135, . | 2.0 | 5 |
| 5 | Variants in the Wnt co-receptor LRP6 are associated with familial exudative vitreoretinopathy. Journal of Genetics and Genomics, 2022, 49, 590-594. | 3.9 | 18 |
| 6 | Identification of Two Novel Variants in the LRP5 Gene that Cause Familial Exudative Vitreoretinopathy. Genetic Testing and Molecular Biomarkers, 2022, 26, 146-151. | 0.7 | 1 |
| 7 | A novel frameshift variant in the TSPAN12 gene causes autosomal dominant <scp>FEVR</scp>. Molecular Genetics & Genomic Medicine, 2022, 10, e1949. | 1.2 | 2 |
| 8 | CTNND1 variants cause familial exudative vitreoretinopathy through the Wnt/cadherin axis. JCI Insight, 2022, 7, . | 5.0 | 17 |
| 9 | Catenin Î± 1 mutations cause familial exudative vitreoretinopathy by overactivating Norrin/Î²-catenin signaling. Journal of Clinical Investigation, 2021, 131, . | 8.2 | 37 |
| 10 | Whole-Exome Sequencing Identified DLG1 as a Candidate Gene for Familial Exudative Vitreoretinopathy. Genetic Testing and Molecular Biomarkers, 2021, 25, 309-316. | 0.7 | 17 |
| 11 | Whole-Exome Sequencing Reveals Novel TSPAN12 Variants in Autosomal Dominant Familial Exudative Vitreoretinopathy. Genetic Testing and Molecular Biomarkers, 2021, 25, 399-404. | 0.7 | 4 |
| 12 | The ER membrane protein complex subunit Emc3 controls angiogenesis via the FZD4/WNT signaling axis. Science China Life Sciences, 2021, 64, 1868-1883. | 4.9 | 16 |
| 13 | Exome sequencing revealed Notch ligand JAG1 as a novel candidate gene for familial exudative vitreoretinopathy. Genetics in Medicine, 2020, 22, 77-84. | 2.4 | 34 |
| 14 | Tmem30a Deficiency in endothelial cells impairs cell proliferation and angiogenesis. Journal of Cell Science, 2019, 132, . | 2.0 | 12 |
| 15 | Targeted next-generation sequencing reveals that a compound heterozygous mutation in phosphodiesterase 6a gene leads to retinitis pigmentosa in a Chinese family. Ophthalmic Genetics, 2018, 39, 487-491. | 1.2 | 5 |
| 16 | Targeted Next-Generation Sequencing Reveals a Novel Frameshift Mutation in the MERTK Gene in a Chinese Family with Retinitis Pigmentosa. Genetic Testing and Molecular Biomarkers, 2018, 22, 165-169. | 0.7 | 5 |
| 17 | Targeted Next-Generation Sequencing Reveals Novel RP1 Mutations in Autosomal Recessive Retinitis Pigmentosa. Genetic Testing and Molecular Biomarkers, 2018, 22, 109-114. | 0.7 | 9 |
| 18 | A splicing mutation in aryl hydrocarbon receptor associated with retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 2563-2572. | 2.9 | 29 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Disruption of Tmem30a results in cerebellar ataxia and degeneration of Purkinje cells. Cell Death and Disease, 2018, 9, 899. | 6.3 | 23 |
| 20 | Candidate Gene Analysis Identifies Mutations inCYP1B1andLTBP2in Indian Families with Primary Congenital Glaucoma. Genetic Testing and Molecular Biomarkers, 2017, 21, 252-258. | 0.7 | 8 |
| 21 | Loss of Tmem30a leads to photoreceptor degeneration. Scientific Reports, 2017, 7, 9296. | 3.3 | 22 |
| 22 | Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population. Scientific Reports, 2016, 6, 19432. | 3.3 | 27 |
| 23 | Whole Exome Sequencing Analysis Identifies Mutations in LRP5 in Indian Families with Familial Exudative Vitreoretinopathy. Genetic Testing and Molecular Biomarkers, 2016, 20, 346-351. | 0.7 | 17 |
| 24 | Association of SNPs in miR-146a, miR-196a2, and miR-499 with the risk of endometrial/ovarian cancer. Acta Biochimica Et Biophysica Sinica, 2015, 47, 564-566. | 2.0 | 23 |