

# Stephen H Tsang

## List of Publications by Year in descending order

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Version: 2024-02-01

356  
papers

9,477  
citations

57758

44  
h-index

88630

70  
g-index

361  
all docs

361  
docs citations

361  
times ranked

8667  
citing authors

| #  | ARTICLE  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. <i>Cell</i> , 2014, 159, 200-214.  | 28.9 | 322       |
| 2  | Unexpected mutations after CRISPR-Cas9 editing in vivo. <i>Nature Methods</i> , 2017, 14, 547-548.   | 19.0 | 294       |
| 3  | Allele-Specific Chromosome Removal after Cas9 Cleavage in Human Embryos. <i>Cell</i> , 2020, 183, 1650-1664.e15.   | 28.9 | 198       |
| 4  | Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. <i>Nature Genetics</i> , 2015, 47, 757-765.   | 21.4 | 183       |
| 5  | Viral Delivery Systems for CRISPR. <i>Viruses</i> , 2019, 11, 28.  | 3.3  | 174       |
| 6  | Long-term Safety and Efficacy of Human-Induced Pluripotent Stem Cell (iPS) Grafts in a Preclinical Model of Retinitis Pigmentosa. <i>Molecular Medicine</i> , 2012, 18, 1312-1319.                 | 4.4  | 162       |
| 7  | Quantitative Fundus Autofluorescence in Recessive Stargardt Disease. , 2014, 55, 2841.   |      | 160       |
| 8  | Frequent hypomorphic alleles account for a significant fraction of ABCA4 disease and distinguish it from age-related macular degeneration. <i>Journal of Medical Genetics</i> , 2017, 54, 404-412. | 3.2  | 140       |
| 9  | Precision Medicine: Genetic Repair of Retinitis Pigmentosa in Patient-Derived Stem Cells. <i>Scientific Reports</i> , 2016, 6, 19969.  | 3.3  | 135       |
| 10 | A Comparison of Fundus Autofluorescence and Retinal Structure in Patients with Stargardt Disease. , 2009, 50, 3953.  |      | 128       |
| 11 | Analysis of the ABCA4 genomic locus in Stargardt disease. <i>Human Molecular Genetics</i> , 2014, 23, 6797-6806.   | 2.9  | 117       |
| 12 | Differentiation of hypothalamic-like neurons from human pluripotent stem cells. <i>Journal of Clinical Investigation</i> , 2015, 125, 796-808.   | 8.2  | 112       |
| 13 | Gene therapy and genome surgery in the retina. <i>Journal of Clinical Investigation</i> , 2018, 128, 2177-2188.  | 8.2  | 111       |
| 14 | Clustered Regularly Interspaced Short Palindromic Repeats-Based Genome Surgery for the Treatment of Autosomal Dominant Retinitis Pigmentosa. <i>Ophthalmology</i> , 2018, 125, 1421-1430.          | 5.2  | 100       |
| 15 | STRUCTURAL ASSESSMENT OF HYPERAUTOFLUORESCENT RING IN PATIENTS WITH RETINITIS PIGMENTOSA. <i>Retina</i> , 2009, 29, 1025-1031.   | 1.7  | 98        |
| 16 | CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa. <i>Molecular Therapy</i> , 2016, 24, 1388-1394.   | 8.2  | 93        |
| 17 | G1961E mutant allele in the Stargardt disease gene ABCA4 causes bull's eye maculopathy. <i>Experimental Eye Research</i> , 2009, 89, 16-24.  | 2.6  | 90        |
| 18 | Quantitative Fundus Autofluorescence and Optical Coherence Tomography in Best Vitelliform Macular Dystrophy. , 2014, 55, 1471.   |      | 89        |

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|----|--|-----|-----------|
| 19 | Phototransduction Influences Metabolic Flux and Nucleotide Metabolism in Mouse Retina. <i>Journal of Biological Chemistry</i> , 2016, 291, 4698-4710.  | 3.4 | 87        |
| 20 | Validation of genome-wide association study (GWAS)-identified disease risk alleles with patient-specific stem cell lines. <i>Human Molecular Genetics</i> , 2014, 23, 3445-3455.   | 2.9 | 86        |
| 21 | Allelic and phenotypic heterogeneity in <i>ABCA4</i> mutations. <i>Ophthalmic Genetics</i> , 2011, 32, 165-174.  | 1.2 | 85        |
| 22 | Comparison of Near-Infrared and Short-Wavelength Autofluorescence in Retinitis Pigmentosa. , 2013, 54, 585.  |     | 83        |
| 23 | Reprogramming metabolism by targeting sirtuin 6 attenuates retinal degeneration. <i>Journal of Clinical Investigation</i> , 2016, 126, 4659-4673.  | 8.2 | 82        |
| 24 | Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in the <i>ABCA4</i> Gene. , 2012, 53, 4458.  |     | 81        |
| 25 | Multimodal Imaging of Central Retinal Disease Progression in a 2-Year Mean Follow-up of Retinitis Pigmentosa. <i>American Journal of Ophthalmology</i> , 2015, 160, 786-798.e4.  | 3.3 | 81        |
| 26 | Gene Therapy in Patient-specific Stem Cell Lines and a Preclinical Model of Retinitis Pigmentosa With Membrane Frizzled-related Protein Defects. <i>Molecular Therapy</i> , 2014, 22, 1688-1697.                                     | 8.2 | 80        |
| 27 | <i>HTRA1</i> , an age-related macular degeneration protease, processes extracellular matrix proteins <i>EFEMP1</i> and <i>TSP1</i> . <i>Aging Cell</i> , 2018, 17, e12710.   | 6.7 | 79        |
| 28 | Progressive Constriction of the Hyperautofluorescent Ring in Retinitis Pigmentosa. <i>American Journal of Ophthalmology</i> , 2012, 153, 718-727.e2.   | 3.3 | 75        |
| 29 | Quantitative Fundus Autofluorescence Distinguishes <i>ABCA4</i> -Associated and Non- <i>ABCA4</i> -Associated Bull's-Eye Maculopathy. <i>Ophthalmology</i> , 2015, 122, 345-355.   | 5.2 | 75        |
| 30 | General Pathophysiology in Retinal Degeneration. <i>Developments in Ophthalmology</i> , 2014, 53, 33-43.   | 0.1 | 74        |
| 31 | Transplantation of Reprogrammed Embryonic Stem Cells Improves Visual Function in a Mouse Model for Retinitis Pigmentosa. <i>Transplantation</i> , 2010, 89, 911-919.   | 1.0 | 71        |
| 32 | Correlations Among Near-Infrared and Short-Wavelength Autofluorescence and Spectral-Domain Optical Coherence Tomography in Recessive Stargardt Disease. <i>Investigative Ophthalmology and Visual Science</i> , 2014, 55, 8134-8143. | 3.3 | 69        |
| 33 | Halting progressive neurodegeneration in advanced retinitis pigmentosa. <i>Journal of Clinical Investigation</i> , 2015, 125, 3704-3713.   | 8.2 | 68        |
| 34 | Novel Phenotypic and Genotypic Findings in X-Linked Retinoschisis. <i>JAMA Ophthalmology</i> , 2007, 125, 259.   | 2.4 | 62        |
| 35 | Whole Exome Sequencing Identifies <i>CRB1</i> Defect in an Unusual Maculopathy Phenotype. <i>Ophthalmology</i> , 2014, 121, 1773-1782.   | 5.2 | 62        |
| 36 | PREFERRED RETINAL LOCUS IN MACULAR DISEASE. <i>Retina</i> , 2008, 28, 1234-1240.   | 1.7 | 61        |

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|----|--|-----|-----------|
| 37 | Extremely hypomorphic and severe deep intronic variants in the <i>ABCA4</i> locus result in varying Stargardt disease phenotypes. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002733. | 1.2 | 61        |
| 38 | Gene and cell-based therapies for inherited retinal disorders: An update. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 349-366.                                | 1.6 | 60        |
| 39 | Modulation of Phosphodiesterase6 Turnoff during Background Illumination in Mouse Rod Photoreceptors. <i>Journal of Neuroscience</i> , 2008, 28, 2064-2074.   | 3.6 | 59        |
| 40 | Spectral-Domain Optical Coherence Tomography Staging and Autofluorescence Imaging in Achromatopsia. <i>JAMA Ophthalmology</i> , 2014, 132, 437.  | 2.5 | 58        |
| 41 | Ciliopathy: Bardet-Biedl Syndrome. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 171-174.   | 1.6 | 58        |
| 42 | Functional Rescue of Degenerating Photoreceptors in Mice Homozygous for a Hypomorphic cGMP Phosphodiesterase 6 b Allele ( <i>Pde6b</i> <sup>H620Q</sup> ). , 2008, 49, 5067.                                     |     | 57        |
| 43 | STRUCTURAL AND FUNCTIONAL CHANGES ASSOCIATED WITH NORMAL AND ABNORMAL FUNDUS AUTOFLUORESCENCE IN PATIENTS WITH RETINITIS PIGMENTOSA. <i>Retina</i> , 2012, 32, 349-357.  | 1.7 | 57        |
| 44 | Quantitative Fundus Autofluorescence and Optical Coherence Tomography in <i>PRPH2/RDS</i> - and <i>ABCA4</i> -Associated Disease Exhibiting Phenotypic Overlap. , 2015, 56, 3159.                                |     | 56        |
| 45 | Quantifying Fundus Autofluorescence in Patients With Retinitis Pigmentosa. , 2017, 58, 1843.   |     | 56        |
| 46 | Gene therapy in inherited retinal degenerative diseases, a review. <i>Ophthalmic Genetics</i> , 2018, 39, 560-568.   | 1.2 | 55        |
| 47 | Gene therapy provides long-term visual function in a pre-clinical model of retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2013, 22, 558-567.  | 2.9 | 54        |
| 48 | The External Limiting Membrane in Early-Onset Stargardt Disease. , 2014, 55, 6139.   |     | 54        |
| 49 | Precision Medicine. <i>JAMA Ophthalmology</i> , 2016, 134, 444.  | 2.5 | 54        |
| 50 | The unfolded protein response regulator ATF6 promotes mesodermal differentiation. <i>Science Signaling</i> , 2018, 11, .   | 3.6 | 54        |
| 51 | A Novel Mutation and Phenotypes in Phosphodiesterase 6 Deficiency. <i>American Journal of Ophthalmology</i> , 2008, 146, 780-788.e1.   | 3.3 | 51        |
| 52 | Measurement and Reproducibility of Preserved Ellipsoid Zone Area and Preserved Retinal Pigment Epithelium Area in Eyes With Choroideremia. <i>American Journal of Ophthalmology</i> , 2017, 179, 110-117.        | 3.3 | 51        |
| 53 | Achromatopsia mutations target sequential steps of ATF6 activation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 400-405.                                 | 7.1 | 50        |
| 54 | Two pathways of rod photoreceptor cell death induced by elevated cGMP. <i>Human Molecular Genetics</i> , 2017, 26, 2299-2306.  | 2.9 | 49        |

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|----|--|-----|-----------|
| 55 | Leber Congenital Amaurosis. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 131-137.  | 1.6 | 49        |
| 56 | Patients and animal models of CNG $\beta$ -deficient retinitis pigmentosa support gene augmentation approach. <i>Journal of Clinical Investigation</i> , 2017, 128, 190-206.   | 8.2 | 48        |
| 57 | Retrospective Analysis of Structural Disease Progression in Retinitis Pigmentosa Utilizing Multimodal Imaging. <i>Scientific Reports</i> , 2017, 7, 10347.   | 3.3 | 46        |
| 58 | Quantification of Peripapillary Sparing and Macular Involvement in Stargardt Disease (STGD1). , 2011, 52, 8006.  |     | 45        |
| 59 | Evaluation of multimodal imaging in carriers of X-linked retinitis pigmentosa. <i>Experimental Eye Research</i> , 2013, 113, 41-48.  | 2.6 | 45        |
| 60 | Personalized therapeutic strategies for patients with retinitis pigmentosa. <i>Expert Opinion on Biological Therapy</i> , 2015, 15, 391-402.   | 3.1 | 43        |
| 61 | BESTROPHIN1 mutations cause defective chloride conductance in patient stem cell-derived RPE. <i>Human Molecular Genetics</i> , 2016, 25, ddw126.   | 2.9 | 43        |
| 62 | Patient-specific mutations impair BESTROPHIN1's essential role in mediating Ca <sup>2+</sup> -dependent Cl <sup>-</sup> currents in human RPE. <i>ELife</i> , 2017, 6, .   | 6.0 | 43        |
| 63 | Characterization of Retinal Structure in <i>ATF6</i> -Associated Achromatopsia. , 2019, 60, 2631.  |     | 43        |
| 64 | Adeno-Associated Viral Gene Therapy for Inherited Retinal Disease. <i>Pharmaceutical Research</i> , 2019, 36, 34.  | 3.5 | 43        |
| 65 | Gene Therapy Restores Mfrp and Corrects Axial Eye Length. <i>Scientific Reports</i> , 2017, 7, 16151.  | 3.3 | 41        |
| 66 | Retinitis Pigmentosa (Non-syndromic). <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 125-130.  | 1.6 | 41        |
| 67 | Transgenic mice carrying the H258N mutation in the gene encoding the $\beta$ -subunit of phosphodiesterase-6 (PDE6B) provide a model for human congenital stationary night blindness. <i>Human Mutation</i> , 2007, 28, 243-254. | 2.5 | 40        |
| 68 | Therapy in Rhodopsin-Mediated Autosomal Dominant Retinitis Pigmentosa. <i>Molecular Therapy</i> , 2020, 28, 2139-2149.   | 8.2 | 40        |
| 69 | <i>CAPN5</i> mutation in hereditary uveitis: the R243L mutation increases calpain catalytic activity and triggers intraocular inflammation in a mouse model. <i>Human Molecular Genetics</i> , 2015, 24, 4584-4598.              | 2.9 | 39        |
| 70 | Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. <i>Human Genetics</i> , 2016, 135, 9-19.  | 3.8 | 39        |
| 71 | The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. <i>Ophthalmology</i> , 2018, 125, 89-99.   | 5.2 | 39        |
| 72 | BEST1: the Best Target for Gene and Cell Therapies. <i>Molecular Therapy</i> , 2015, 23, 1805-1809.  | 8.2 | 38        |

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|----|---|------|-----------|
| 73 | Correction of Monogenic and Common Retinal Disorders with Gene Therapy. <i>Genes</i> , 2017, 8, 53.   | 2.4  | 37        |
| 74 | Quantitative progression of retinitis pigmentosa by optical coherence tomography angiography. <i>Scientific Reports</i> , 2018, 8, 13130.   | 3.3  | 37        |
| 75 | Gene therapy for inherited retinal diseases. <i>Annals of Translational Medicine</i> , 2021, 9, 1278-1278.  | 1.7  | 36        |
| 76 | Functional validation of a human CAPN5 exome variant by lentiviral transduction into mouse retina. <i>Human Molecular Genetics</i> , 2014, 23, 2665-2677.   | 2.9  | 35        |
| 77 | Choroidal and Retinal Thickening in Severe Preeclampsia. , 2014, 55, 5723.  |      | 35        |
| 78 | Mid-stage intervention achieves similar efficacy as conventional early-stage treatment using gene therapy in a pre-clinical model of retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2014, 23, 514-523.                           | 2.9  | 35        |
| 79 | Structural Modeling of a Novel CAPN5 Mutation that Causes Uveitis and Neovascular Retinal Detachment. <i>PLoS ONE</i> , 2015, 10, e0122352.   | 2.5  | 35        |
| 80 | Stem Cell Therapies in Retinal Disorders. <i>Cells</i> , 2017, 6, 4.  | 4.1  | 35        |
| 81 | Stargardt Disease. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 139-151.  | 1.6  | 35        |
| 82 | Proteomic analysis of the human retina reveals region-specific susceptibilities to metabolic- and oxidative stress-related diseases. <i>PLoS ONE</i> , 2018, 13, e0193250.  | 2.5  | 35        |
| 83 | Autofluorescence Imaging and Spectral-Domain Optical Coherence Tomography in Incomplete Congenital Stationary Night Blindness and Comparison With Retinitis Pigmentosa. <i>American Journal of Ophthalmology</i> , 2012, 153, 143-154.e2. | 3.3  | 34        |
| 84 | Disease progression in autosomal dominant cone-rod dystrophy caused by a novel mutation (D100G) in the GUCA1A gene. <i>Documenta Ophthalmologica</i> , 2014, 128, 59-67.  | 2.2  | 34        |
| 85 | Personalized Proteomics in Proliferative Vitreoretinopathy Implicate Hematopoietic Cell Recruitment and mTOR as a Therapeutic Target. <i>American Journal of Ophthalmology</i> , 2018, 186, 152-163.                                      | 3.3  | 34        |
| 86 | Therapeutic Margins in a Novel Preclinical Model of Retinitis Pigmentosa. <i>Journal of Neuroscience</i> , 2013, 33, 13475-13483.   | 3.6  | 33        |
| 87 | Personalized Proteomics for Precision Health: Identifying Biomarkers of Vitreoretinal Disease. <i>Translational Vision Science and Technology</i> , 2018, 7, 12.  | 2.2  | 33        |
| 88 | Extracellular superoxide dismutase (SOD3) regulates oxidative stress at the vitreoretinal interface. <i>Free Radical Biology and Medicine</i> , 2018, 124, 408-419.   | 2.9  | 32        |
| 89 | Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. <i>European Journal of Human Genetics</i> , 2017, 25, 735-743.  | 2.8  | 31        |
| 90 | Distinct Imprinting Signatures and Biased Differentiation of Human Androgenetic and Parthenogenetic Embryonic Stem Cells. <i>Cell Stem Cell</i> , 2019, 25, 419-432.e9.   | 11.1 | 31        |

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|-----|--|-----|-----------|
| 91  | Structural and Genetic Assessment of the ABCA4-Associated Optical Gap Phenotype. , 2014, 55, 7217.   |     | 30        |
| 92  | New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.  | 2.9 | 30        |
| 93  | Laser-Induced Photic Injury Phenocopies Macular Dystrophy. Ophthalmic Genetics, 2016, 37, 59-67.   | 1.2 | 30        |
| 94  | Reprogramming towards anabolism impedes degeneration in a preclinical model of retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 4244-4255.  | 2.9 | 30        |
| 95  | Metabolite therapy guided by liquid biopsy proteomics delays retinal neurodegeneration. EBioMedicine, 2020, 52, 102636.  | 6.1 | 30        |
| 96  | Bardet-Biedl syndrome proteins regulate intracellular signaling and neuronal function in patient-specific iPSC-derived neurons. Journal of Clinical Investigation, 2021, 131, .                    | 8.2 | 30        |
| 97  | Calpain-5 Expression in the Retina Localizes to Photoreceptor Synapses. , 2016, 57, 2509.  |     | 29        |
| 98  | Quantitative Fundus Autofluorescence and Optical Coherence Tomography in ABCA4 Carriers. , 2015, 56, 7274.   |     | 28        |
| 99  | Unilateral Retinitis Pigmentosa: A Proposal of Genetic Pathogenic Mechanisms. European Journal of Ophthalmology, 2012, 22, 654-660.  | 1.3 | 27        |
| 100 | Abnormality in the external limiting membrane in early Stargardt Disease. Ophthalmic Genetics, 2013, 34, 75-77.  | 1.2 | 27        |
| 101 | CRISPR applications in ophthalmologic genome surgery. Current Opinion in Ophthalmology, 2017, 28, 252-259.   | 2.9 | 27        |
| 102 | Ciliopathy: Usher Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 167-170.  | 1.6 | 27        |
| 103 | A case-control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic ABCA4 variants. Genetics in Medicine, 2019, 21, 2336-2344. | 2.4 | 27        |
| 104 | Investigation and Restoration of BEST1 Activity in Patient-derived RPEs with Dominant Mutations. Scientific Reports, 2019, 9, 19026.   | 3.3 | 27        |
| 105 | Mitochondrial Disorder: Kearns-Sayre Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 161-162.   | 1.6 | 27        |
| 106 | Therapeutic drug repositioning using personalized proteomics of liquid biopsies. JCI Insight, 2017, 2, .   | 5.0 | 27        |
| 107 | Lentivirus-mediated expression of cDNA and shRNA slows degeneration in retinitis pigmentosa. Experimental Biology and Medicine, 2011, 236, 1211-1217.  | 2.4 | 26        |
| 108 | Patient-Specific iPSC-Derived RPE for Modeling of Retinal Diseases. Journal of Clinical Medicine, 2015, 4, 567-578.  | 2.4 | 26        |

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|-----|--|------|-----------|
| 109 | MULTIMODAL IMAGING OF DISEASE-ASSOCIATED PIGMENTARY CHANGES IN RETINITIS PIGMENTOSA. <i>Retina</i> , 2016, 36, S147-S158.  | 1.7  | 26        |
| 110 | Personalized Medicine: Cell and Gene Therapy Based on Patient-Specific iPSC-Derived Retinal Pigment Epithelium Cells. <i>Advances in Experimental Medicine and Biology</i> , 2016, 854, 549-555.   | 1.6  | 26        |
| 111 | Genetic rescue models refute nonautonomous rod cell death in retinitis pigmentosa. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 5259-5264.  | 7.1  | 26        |
| 112 | Electroretinography Reveals Difference in Cone Function between Syndromic and Nonsyndromic USH2A Patients. <i>Scientific Reports</i> , 2017, 7, 11170.   | 3.3  | 26        |
| 113 | Multimodal Imaging in Best Vitelliform Macular Dystrophy. , 2019, 60, 2012.  |      | 26        |
| 114 | shRNA knockdown of guanylate cyclase 2a, cyclic nucleotide gated channel alpha 1, increases photoreceptor survival in a cGMP, phosphodiesterase, mouse model of retinitis pigmentosa. <i>Journal of Cellular and Molecular Medicine</i> , 2011, 15, 1778-1787. | 3.6  | 25        |
| 115 | A Comparison of En Face Optical Coherence Tomography and Fundus Autofluorescence in Stargardt Disease. , 2017, 58, 5227.   |      | 25        |
| 116 | Revolution in Gene Medicine Therapy and Genome Surgery. <i>Genes</i> , 2018, 9, 575.   | 2.4  | 25        |
| 117 | Mutations in GPR143/OA1 and ABCA4 Inform Interpretations of Short-Wavelength and Near-Infrared Fundus Autofluorescence. , 2018, 59, 2459.  |      | 25        |
| 118 | Cis-acting modifiers in the ABCA4 locus contribute to the penetrance of the major disease-causing variant in Stargardt disease. <i>Human Molecular Genetics</i> , 2021, 30, 1293-1304.   | 2.9  | 25        |
| 119 | Subretinal Injection of Gene Therapy Vectors and Stem Cells in the Perinatal Mouse Eye. <i>Journal of Visualized Experiments</i> , 2012, , .   | 0.3  | 24        |
| 120 | Quantitative Autofluorescence Intensities in Acute Zonal Occult Outer Retinopathy vs Healthy Eyes. <i>JAMA Ophthalmology</i> , 2017, 135, 1330.  | 2.5  | 24        |
| 121 | Autosomal Dominant Retinitis Pigmentosa. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 69-77.   | 1.6  | 24        |
| 122 | Two-year progression analysis of RPE65 autosomal dominant retinitis pigmentosa. <i>Ophthalmic Genetics</i> , 2018, 39, 544-549.  | 1.2  | 24        |
| 123 | Effects of deficiency in the RLBP1-encoded visual cycle protein CRALBP on visual dysfunction in humans and mice. <i>Journal of Biological Chemistry</i> , 2020, 295, 6767-6780.  | 3.4  | 24        |
| 124 | Fundus autofluorescence in cone dystrophy. <i>Documenta Ophthalmologica</i> , 2009, 119, 141-144.  | 2.2  | 23        |
| 125 | Loss of peripapillary sparing in non-group I Stargardt disease. <i>Experimental Eye Research</i> , 2010, 91, 592-600.  | 2.6  | 23        |
| 126 | Neuroretinal hypoxic signaling in a new preclinical murine model for proliferative diabetic retinopathy. <i>Signal Transduction and Targeted Therapy</i> , 2016, 1, .  | 17.1 | 23        |



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|-----|---|-----|-----------|
| 127 | Peripapillary sparing in <i>RDH12</i> -associated Leber congenital amaurosis. <i>Ophthalmic Genetics</i> , 2017, 38, 575-579.   | 1.2 | 23        |
| 128 | Structural modeling of a novel <i>SLC38A8</i> mutation that causes foveal hypoplasia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2017, 5, 202-209.  | 1.2 | 23        |
| 129 | A novel de novo <i>CAPN5</i> mutation in a patient with inflammatory vitreoretinopathy, hearing loss, and developmental delay. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002519.                                 | 1.2 | 23        |
| 130 | A Distinct Phenotype of Eyes Shut Homolog ( <i>EYS</i> )-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. <i>American Journal of Ophthalmology</i> , 2018, 190, 99-112.  | 3.3 | 23        |
| 131 | Disruption of the human cone photoreceptor mosaic from a defect in NR2E3 transcription factor function in young adults. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2013, 251, 2299-2309.                           | 1.9 | 22        |
| 132 | Cellular imaging demonstrates genetic mosaicism in heterozygous carriers of an X-linked ciliopathy gene. <i>European Journal of Human Genetics</i> , 2013, 21, 1240-1248.   | 2.8 | 22        |
| 133 | Bilateral Concordance of the Fundus Hyperautofluorescent Ring in Typical Retinitis Pigmentosa Patients. <i>Ophthalmic Genetics</i> , 2015, 36, 113-122.   | 1.2 | 22        |
| 134 | Vigabatrin-Induced Retinal Toxicity Is Partially Mediated by Signaling in Rod and Cone Photoreceptors. <i>PLoS ONE</i> , 2012, 7, e43889.   | 2.5 | 22        |
| 135 | Mice with a D190N Mutation in the Gene Encoding Rhodopsin: A Model for Human Autosomal-Dominant Retinitis Pigmentosa. <i>Molecular Medicine</i> , 2012, 18, 549-555.  | 4.4 | 21        |
| 136 | A novel RPGR mutation masquerading as Stargardt disease. <i>British Journal of Ophthalmology</i> , 2014, 98, 709-711.   | 3.9 | 21        |
| 137 | Spectrum of Disease Severity and Phenotype in Choroideremia Carriers. <i>American Journal of Ophthalmology</i> , 2019, 207, 77-86.  | 3.3 | 21        |
| 138 | Early Structural Anomalies Observed by High-Resolution Imaging in Two Related Cases of Autosomal-Dominant Retinitis Pigmentosa. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2014, 45, 469-473.                                      | 0.7 | 21        |
| 139 | Phenotype-Genotype Correlations in Autosomal Dominant Retinitis Pigmentosa Caused by RHO, D190N. <i>Current Eye Research</i> , 2008, 33, 1014-1022.   | 1.5 | 20        |
| 140 | Ciliopathy: Alström Syndrome. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 179-180.   | 1.6 | 20        |
| 141 | Fundus autofluorescence and ellipsoid zone (EZ) line width can be an outcome measurement in RHO-associated autosomal dominant retinitis pigmentosa. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2019, 257, 725-731. | 1.9 | 20        |
| 142 | Treatment-Emergent Adverse Events in Gene Therapy Trials for Inherited Retinal Diseases: A Narrative Review. <i>Ophthalmology and Therapy</i> , 2020, 9, 709-724.   | 2.3 | 20        |
| 143 | Progressive Cone Dystrophy and Cone-Rod Dystrophy (XL, AD, and AR). <i>Advances in Experimental Medicine and Biology</i> , 2018, 1085, 53-60.   | 1.6 | 20        |
| 144 | Case Report: Autofluorescence Imaging and Phenotypic Variance in a Sibling Pair with Early-Onset Retinal Dystrophy Due to Defective <i>CRB1</i> Function. <i>Current Eye Research</i> , 2009, 34, 395-400.                                    | 1.5 | 19        |

| #   | ARTICLE   | IF   | CITATIONS |
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