# Stephen H Tsang

List of Publications by Year
in descending order



| 356 | 9,477 | 66250 | 100535 |
|---|---|---|---|
| papers | citations | 44 h-index | 70 g-index |

| 361 | 361 | 361 | 9374 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Vitamin A deficiency and the retinal â€œdouble carrotâ€•sign with optical coherence tomography. Eye, 2023, 37, 1489-1495. | 1.1 | 2 |
| 2 | VITAMIN A DEFICIENCY MONITORED BY QUANTITATIVE SHORT WAVELENGTH FUNDUS AUTOFLUORESCENCE IN A CASE OF BARIATRIC SURGERY. Retinal Cases and Brief Reports, 2022, 16, 218-221. | 0.3 | 9 |
| 3 | A mutation in CRX causing pigmented paravenous retinochoroidal atrophy. European Journal of Ophthalmology, 2022, 32, NP235-NP239. | 0.7 | 4 |
| 4 | Renormalization of metabolic coupling treats age-related degenerative disorders: an oxidative RPE niche fuels the more glycolytic photoreceptors. Eye, 2022, 36, 278-283. | 1.1 | 6 |
| 5 | A genotype-phenotype correlation matrix for ABCA4 disease based on long-term prognostic outcomes. JCI Insight, 2022, 7, . | 2.3 | 16 |
| 6 | Comparisons Among Optical Coherence Tomography and Fundus Autofluorescence Modalities as Measurements of Atrophy in ABCA4-Associated Disease. Translational Vision Science and Technology, 2022, 11, 36. | 1.1 | 1 |
| 7 | CRISPR genome surgery in a novel humanized model for autosomal dominant retinitis pigmentosa. Molecular Therapy, 2022, 30, 1407-1420. | 3.7 | 16 |
| 8 | Long-term vitamin A supplementation in a preclinical mouse model forRhoD190N-associated retinitis pigmentosa. Human Molecular Genetics, 2022, 31, 2438-2451. | 1.4 | 5 |
| 9 | Late-stage rescue of visually guided behavior in the context of a significantly remodeled retinitis pigmentosa mouse model. Cellular and Molecular Life Sciences, 2022, 79, 148. | 2.4 | 3 |
| 10 | Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease. PLoS Genetics, 2022, 18, e1010129. | 1.5 | 8 |
| 11 | Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858. | 1.1 | 8 |
| 12 | Expanding the phenotype of TTLL5-associated retinal dystrophy: a case series. Orphanet Journal of Rare Diseases, 2022, 17, 146. | 1.2 | 3 |
| 13 | Proteomic Analysis of Autoimmune Retinopathy Implicates Neuronal Cell Adhesion Molecule as a Potential Biomarker. Ophthalmology Science, 2022, 2, 100131. | 1.0 | 4 |
| 14 | Multimodal imaging reveals retinoschisis masquerading as retinal detachment in patients with choroideremia. American Journal of Ophthalmology Case Reports, 2022, 26, 101543. | 0.4 | 2 |
| 15 | Longitudinal Analysis of a Resolving Foveomacular Vitelliform Lesion in ABCA4 Disease. Ophthalmology Retina, 2022, 6, 847-860. | 1.2 | 1 |
| 16 | CRISPR/Cas therapeutic strategies for autosomal dominant disorders. Journal of Clinical Investigation, 2022, 132, . | 3.9 | 8 |
| 17 | Choroideremia Carriers: Dark-Adapted Perimetry and Retinal Structures. , 2022, 63, 4. | | 3 |
| 18 | PROGRESSION OF SCOTOPIC SINGLE-FLASH ELECTRORETINOGRAPHY IN THE STAGES OF CAPN5 VITREORETINOPATHY. Retinal Cases and Brief Reports, 2021, 15, 473-478. | 0.3 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | CHORIORETINAL CHANGES IN A GENETICALLY CONFIRMED CASE OF BOUCHERâ€“NEUHÃ„USER SYNDROME. Retinal Cases and Brief Reports, 2021, 15, 179-184. | 0.3 | 9 |
| 20 | Whole-Exome Sequencing of Patients With Posterior Segment Uveitis. American Journal of Ophthalmology, 2021, 221, 246-259. | 1.7 | 10 |
| 21 | Retinal pigment epithelium lipid metabolic demands and therapeutic restoration. Taiwan Journal of Ophthalmology, 2021, 11, 216. | 0.3 | 6 |
| 22 | Retinal Pigment Epithelium Atrophy in Recessive Stargardt Disease as Measured by Short-Wavelength and Near-Infrared Autofluorescence. Translational Vision Science and Technology, 2021, 10, 3. | 1.1 | 9 |
| 23 | Nutrigenetic reprogramming of oxidative stress. Taiwan Journal of Ophthalmology, 2021, 11, 207. | 0.3 | 2 |
| 24 | Central serous chorioretinopathy treatment with a systemic PDE5 and PDE6 inhibitor (sildenafil). American Journal of Ophthalmology Case Reports, 2021, 21, 100998. | 0.4 | 2 |
| 25 | Commentary on â€œExome sequencing and electro-clinical features in pediatric patients with very early-onset retinal dystrophies: A cohort studyâ€. European Journal of Paediatric Neurology, 2021, 31, 106-107. | 0.7 | 0 |
| 26 | Telegenetics for inherited retinal diseases in the COVID-19 environment. International Journal of Retina and Vitreous, 2021, 7, 25. | 0.9 | 4 |
| 27 | Bardet-Biedl syndrome proteins regulate intracellular signaling and neuronal function in patient-specific iPSC-derived neurons. Journal of Clinical Investigation, 2021, 131, . | 3.9 | 30 |
| 28 | Cis-acting modifiers in the ABCA4 locus contribute to the penetrance of the major disease-causing variant in Stargardt disease. Human Molecular Genetics, 2021, 30, 1293-1304. | 1.4 | 25 |
| 29 | A novel KCNV2 mutation in a patient taking hydroxychloroquine associated with cone dystrophy with supernormal rod response. Ophthalmic Genetics, 2021, 42, 458-463. | 0.5 | 2 |
| 30 | CNGB1 â€related rodâ€cone dystrophy: A mutation review and update. Human Mutation, 2021, 42, 641-666. | 1.1 | 16 |
| 31 | Impaired cholesterol efflux in retinal pigment epithelium of individuals with juvenile macular degeneration. American Journal of Human Genetics, 2021, 108, 903-918. | 2.6 | 10 |
| 32 | Overcoming translational barriers in modeling macular degenerations. Cell Stem Cell, 2021, 28, 781-783. | 5.2 | 1 |
| 33 | Distinct expression requirements and rescue strategies for BEST1 loss- and gain-of-function mutations. ELife, 2021, 10, . | 2.8 | 11 |
| 34 | Correspondence. Ophthalmology Retina, 2021, 5, e7-e8. | 1.2 | 0 |
| 35 | Shared Features in Retinal Disorders With Involvement of Retinal Pigment Epithelium. , 2021, 62, 15. | | 5 |
| 36 | Stage-dependent choriocapillaris impairment in Best vitelliform macular dystrophy characterized by optical coherence tomography angiography. Scientific Reports, 2021, 11, 14300. | 1.6 | 4 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. Ophthalmic Genetics, 2021, 42, 664-673. | 0.5 | 14 |
| 38 | Mouse Models of Achromatopsia in Addressing Temporal â€œPoint of No Returnâ€•in Gene-Therapy. International Journal of Molecular Sciences, 2021, 22, 8069. | 1.8 | 2 |
| 39 | Gene therapy for inherited retinal diseases. Annals of Translational Medicine, 2021, 9, 1278-1278. | 0.7 | 36 |
| 40 | Precision Medicine Trials in Retinal Degenerations. Annual Review of Vision Science, 2021, 7, 851-865. | 2.3 | 6 |
| 41 | Clinical and genetic findings in Italian patients with sector retinitis pigmentosa. Molecular Vision, 2021, 27, 78-94. | 1.1 | 2 |
| 42 | Management and treatment of inherited retinal dystrophies. Taiwan Journal of Ophthalmology, 2021, 11, 205-206. | 0.3 | 0 |
| 43 | Phase transition specified by a binary code patterns the vertebrate eye cup. Science Advances, 2021, 7, eabj9846. | 4.7 | 19 |
| 44 | Novel REEP6 gene mutation associated with autosomal recessive retinitis pigmentosa. Documenta Ophthalmologica, 2020, 140, 67-75. | 1.0 | 6 |
| 45 | Therapy in Rhodopsin-Mediated Autosomal Dominant Retinitis Pigmentosa. Molecular Therapy, 2020, 28, 2139-2149. | 3.7 | 40 |
| 46 | Retinal Manifestations of Mitochondrial Oxidative Phosphorylation Disorders. , 2020, 61, 12. | | 9 |
| 47 | Compound heterozygous inheritance of two novel COQ2 variants results in familial coenzyme Q deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 320. | 1.2 | 8 |
| 48 | Treatment-Emergent Adverse Events in Gene Therapy Trials for Inherited Retinal Diseases: A Narrative Review. Ophthalmology and Therapy, 2020, 9, 709-724. | 1.0 | 20 |
| 49 | Quantitative Autofluorescence Following Gene Therapy With Voretigene Neparvovec. JAMA Ophthalmology, 2020, 138, 919. | 1.4 | 9 |
| 50 | Allele-Specific Chromosome Removal after Cas9 Cleavage in Human Embryos. Cell, 2020, 183, 1650-1664.e15. | 13.5 | 198 |
| 51 | Dark noise and retinal degeneration from D190N-rhodopsin. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 23033-23043. | 3.3 | 6 |
| 52 | Quantitative Fundus Autofluorescence in HCQ Retinopathy. , 2020, 61, 41. | | 14 |
| 53 | Presumed Chloroquine Retinopathy With Short-term Therapy for Glioblastoma Multiforme. JAMA Ophthalmology, 2020, 138, 1215. | 1.4 | 3 |
| 54 | Spectral-Domain Optical Coherence Tomography Is More Sensitive for Hydroxychloroquine-Related Structural Abnormalities Than Short-Wavelength and Near-Infrared Autofluorescence. Translational Vision Science and Technology, 2020, 9, 8. | 1.1 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Stickler Syndrome Genotype (COL2A1 mutation) with Retinitis Pigmentosa Phenotype. Ophthalmology Retina, 2020, 4, 522. | 1.2 | 2 |
| 56 | Sequential multiple retinal vein occlusions and transient ischemic attack in MTHFR polymorphism and protein S deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1273. | 0.6 | 4 |
| 57 | PKM2 ablation enhanced retinal function and survival in a preclinical model of retinitis pigmentosa. Mammalian Genome, 2020, 31, 77-85. | 1.0 | 9 |
| 58 | Phenotypic variance in Calpain-5 retinal degeneration. American Journal of Ophthalmology Case Reports, 2020, 18, 100627. | 0.4 | 7 |
| 59 | Optical coherence tomography in the evaluation of retinitis pigmentosa. Ophthalmic Genetics, 2020, 41, 413-419. | 0.5 | 9 |
| 60 | Stargardt Juvenile Macular Degeneration. New England Journal of Medicine, 2020, 382, 2353-2353. | 13.9 | 0 |
| 61 | Quasidominance in autosomal recessive RDH12-Leber congenital amaurosis. Ophthalmic Genetics, 2020, 41, 198-200. | 0.5 | 2 |
| 62 | Phenotypic expansion of autosomal dominant retinitis pigmentosa associated with the D477G mutation in RPE65. Journal of Physical Education and Sports Management, 2020, 6, a004952. | 0.5 | 11 |
| 63 | Prospective Impact of Sildenafil on Chronic cEntral Serous Chorioretinopathy. Ophthalmology Retina, 2020, 4, 1119-1123. | 1.2 | 11 |
| 64 | Short-Wavelength and Near-Infrared Autofluorescence in Patients with Deficiencies of the Visual Cycle and Phototransduction. Scientific Reports, 2020, 10, 8998. | 1.6 | 9 |
| 65 | Metabolite therapy guided by liquid biopsy proteomics delays retinal neurodegeneration. EBioMedicine, 2020, 52, 102636. | 2.7 | 30 |
| 66 | Disease asymmetry and hyperautofluorescent ring shape in retinitis pigmentosa patients. Scientific Reports, 2020, 10, 3364. | 1.6 | 9 |
| 67 | Progressive RPE atrophy and photoreceptor death in KIZ-associated autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2020, 41, 26-30. | 0.5 | 3 |
| 68 | Inhibition of Ca <sup>2+</sup> channel surface expression by mutant bestrophinâ€1 in RPE cells. FASEB Journal, 2020, 34, 4055-4071. | 0.2 | 8 |
| 69 | Comparative Analysis of Functional and Structural Decline in Retinitis Pigmentosas. International Journal of Molecular Sciences, 2020, 21, 2730. | 1.8 | 5 |
| 70 | Progressive Choriocapillaris Impairment in ABCA4 Maculopathy Is Secondary to Retinal Pigment Epithelium Atrophy. , 2020, 61, 13. | | 5 |
| 71 | Perspectives on Gene Therapy: Choroideremia Represents a Challenging Model for the Treatment of Other Inherited Retinal Degenerations. Translational Vision Science and Technology, 2020, 9, 17. | 1.1 | 11 |
| 72 | Differences in Intraretinal Pigment Migration Across Inherited Retinal Dystrophies. American Journal of Ophthalmology, 2020, 217, 252-260. | 1.7 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Effects of deficiency in the RLBP1-encoded visual cycle protein CRALBP on visual dysfunction in humans and mice. Journal of Biological Chemistry, 2020, 295, 6767-6780. | 1.6 | 24 |
| 74 | Multiexon deletion alleles of ATF6 linked to achromatopsia. JCI Insight, 2020, 5, . | 2.3 | 13 |
| 75 | Precision metabolome reprogramming for imprecision therapeutics in retinitis pigmentosa. Journal of Clinical Investigation, 2020, 130, 3971-3973. | 3.9 | 11 |
| 76 | Fundoscopy-directed genetic testing to re-evaluate negative whole exome sequencing results. Orphanet Journal of Rare Diseases, 2020, 15, 32. | 1.2 | 8 |
| 77 | Optical Gap Biomarker in Cone-Dominant Retinal Dystrophy. American Journal of Ophthalmology, 2020, 218, 40-53. | 1.7 | 4 |
| 78 | Non-paraneoplastic related retinopathy: clinical challenges and review. Ophthalmic Genetics, 2019, 40, 293-297. | 0.5 | 3 |
| 79 | Choroidal neovascularization in an adolescent with RDH12-associated retinal degeneration. Ophthalmic Genetics, 2019, 40, 362-364. | 0.5 | 4 |
| 80 | CAPN5 genetic inactivation phenotype supports therapeutic inhibition trials. Human Mutation, 2019, 40, 2377-2392. | 1.1 | 9 |
| 81 | Significant Vision Recovery after Early Treatment of Diffuse Unilateral Subacute Neuroretinitis. Ophthalmology Retina, 2019, 3, 709. | 1.2 | 0 |
| 82 | Comparison of structural progression between ciliopathy and non-ciliopathy associated with autosomal recessive retinitis pigmentosa. Orphanet Journal of Rare Diseases, 2019, 14, 187. | 1.2 | 15 |
| 83 | Multimodal structural disease progression of retinitis pigmentosa according to mode of inheritance. Scientific Reports, 2019, 9, 10712. | 1.6 | 19 |
| 84 | Characterization of Retinal Structure in ATF6-Associated Achromatopsia. , 2019, 60, 2631. | | 43 |
| 85 | CRISPR Base Editing in Induced Pluripotent Stem Cells. Methods in Molecular Biology, 2019, 2045, 337-346. | 0.4 | 11 |
| 86 | HMGB1 and Caveolin-1 related to RPE cell senescence in age-related macular degeneration. Aging, 2019, 11, 4323-4337. | 1.4 | 17 |
| 87 | CLIC4 regulates late endosomal trafficking and matrix degradation activity of MMP14 at focal adhesions in RPE cells. Scientific Reports, 2019, 9, 12247. | 1.6 | 16 |
| 88 | Hypoxic drive caused type 3 neovascularization in a preclinical model of exudative age-related macular degeneration. Human Molecular Genetics, 2019, 28, 3475-3485. | 1.4 | 9 |
| 89 | Distinct Imprinting Signatures and Biased Differentiation of Human Androgenetic and Parthenogenetic Embryonic Stem Cells. Cell Stem Cell, 2019, 25, 419-432.e9. | 5.2 | 31 |
| 90 | Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375. | 0.5 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Fundus autofluorescence and ellipsoid zone (EZ) line width can be an outcome measurement in RHO-associated autosomal dominant retinitis pigmentosa. Graefe's Archive for Clinical and Experimental Ophthalmology, 2019, 257, 725-731. | 1.0 | 20 |
| 92 | VCANCanonical Splice Site Mutation is Associated With Vitreoretinal Degeneration and Disrupts an MMP Proteolytic Site. , 2019, 60, 282. | | 19 |
| 93 | Proteomic insight into the pathogenesis of CAPN5-vitreoretinopathy. Scientific Reports, 2019, 9, 7608. | 1.6 | 9 |
| 94 | Spectrum of Disease Severity and Phenotype in Choroideremia Carriers. American Journal of Ophthalmology, 2019, 207, 77-86. | 1.7 | 21 |
| 95 | Multimodal Imaging in Best Vitelliform Macular Dystrophy. , 2019, 60, 2012. | | 26 |
| 96 | Correlation between B-scan optical coherence tomography, en face thickness map ring and hyperautofluorescent ring in retinitis pigmentosa patients. Graefe's Archive for Clinical and Experimental Ophthalmology, 2019, 257, 1601-1609. | 1.0 | 4 |
| 97 | Multi-platform imaging in ABCA4-Associated Disease. Scientific Reports, 2019, 9, 6436. | 1.6 | 17 |
| 98 | Therapeutic Window for Phosphodiesterase 6â€“Related Retinitis Pigmentosa. JAMA Ophthalmology, 2019, 137, 679. | 1.4 | 3 |
| 99 | Mechanisms of neurodegeneration in a preclinical autosomal dominant retinitis pigmentosa knock-in model with a RhoD190N mutation. Cellular and Molecular Life Sciences, 2019, 76, 3657-3665. | 2.4 | 7 |
| 100 | A caseâ€“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. | 1.1 | 27 |
| 101 | Investigation and Restoration of BEST1 Activity in Patient-derived RPEs with Dominant Mutations. Scientific Reports, 2019, 9, 19026. | 1.6 | 27 |
| 102 | Inside Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, ii. | 1.1 | 0 |
| 103 | Compound heterozygous novel frameshift variants in the PROM1 gene result in Leber congenital amaurosis. Journal of Physical Education and Sports Management, 2019, 5, a004481. | 0.5 | 9 |
| 104 | Novel mutations in the 3-box motif of the BACK domain of KLHL7 associated with nonsyndromic autosomal dominant retinitis pigmentosa. Orphanet Journal of Rare Diseases, 2019, 14, 295. | 1.2 | 4 |
| 105 | SCAPERâ€associated nonsyndromic autosomal recessive retinitis pigmentosa. American Journal of Medical Genetics, Part A, 2019, 179, 312-316. | 0.7 | 10 |
| 106 | Attenuation of Inherited and Acquired Retinal Degeneration Progression with Gene-based Techniques. Molecular Diagnosis and Therapy, 2019, 23, 113-120. | 1.6 | 6 |
| 107 | Viral Delivery Systems for CRISPR. Viruses, 2019, 11, 28. | 1.5 | 174 |
| 108 | Adeno-Associated Viral Gene Therapy for Inherited Retinal Disease. Pharmaceutical Research, 2019, 36, 34. | 1.7 | 43 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Macular spatial distribution of preserved autofluorescence in patients with choroideremia. British Journal of Ophthalmology, 2019, 103, 933-937. | 2.1 | 12 |
| 110 | Hyperautofluorescent Dots are Characteristic in Ceramide Kinase Like-associated Retinal Degeneration. Scientific Reports, 2019, 9, 876. | 1.6 | 8 |
| 111 | Phenotypic expansion and progression of SPATA7-associated retinitis pigmentosa. Documenta Ophthalmologica, 2018, 136, 125-133. | 1.0 | 5 |
| 112 | A novel de novo CAPN5 mutation in a patient with inflammatory vitreoretinopathy, hearing loss, and developmental delay. Journal of Physical Education and Sports Management, 2018, 4, a002519. | 0.5 | 23 |
| 113 | The unfolded protein response regulator ATF6 promotes mesodermal differentiation. Science Signaling, 2018, 11, . | 1.6 | 54 |
| 114 | Autologous stem cell therapy for inherited and acquired retinal disease. Regenerative Medicine, 2018, 13, 89-96. | 0.8 | 10 |
| 115 | Personalized Proteomics in Proliferative Vitreoretinopathy Implicate Hematopoietic Cell Recruitment and mTOR as a Therapeutic Target. American Journal of Ophthalmology, 2018, 186, 152-163. | 1.7 | 34 |
| 116 | Reprogramming the metabolome rescues retinal degeneration. Cellular and Molecular Life Sciences, 2018, 75, 1559-1566. | 2.4 | 18 |
| 117 | Success of Gene Therapy in Late-Stage Treatment. Advances in Experimental Medicine and Biology, 2018, 1074, 101-107. | 0.8 | 4 |
| 118 | Treatment of Macular Degeneration with Sildenafil: Results of a Two-Year Trial. Ophthalmologica, 2018, 240, 45-54. | 1.0 | 19 |
| 119 | A Distinct Phenotype of Eyes Shut Homolog ( EYS )-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. American Journal of Ophthalmology, 2018, 190, 99-112. | 1.7 | 23 |
| 120 | CHOROIDEREMIA ASSOCIATED WITH A NOVEL SYNONYMOUS MUTATION IN GENE ENCODING REP-1. Retinal Cases and Brief Reports, 2018, 12, S67-S71. | 0.3 | 6 |
| 121 | HYPERREFLECTIVE DEPOSITION IN THE BACKGROUND OF ADVANCED STARGARDT DISEASE. Retina, 2018, 38, 2214-2219. | 1.0 | 7 |
| 122 | The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. Ophthalmology, 2018, 125, 89-99. | 2.5 | 39 |
| 123 | CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa: A Brief Methodology. Methods in Molecular Biology, 2018, 1715, 191-205. | 0.4 | 4 |
| 124 | Stem cell therapy and regenerative medicine in RPE degenerative disease: advances and challenges. Expert Review of Ophthalmology, 2018, 13, 321-327. | 0.3 | 0 |
| 125 | Caring for Hereditary Childhood Retinal Blindness. Asia-Pacific Journal of Ophthalmology, 2018, 7, 183-191. | 1.3 | 14 |
| 126 | Personalized Proteomics for Precision Health: Identifying Biomarkers of Vitreoretinal Disease. Translational Vision Science and Technology, 2018, 7, 12. | 1.1 | 33 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Extracellular Matrix: Alport Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 197-198. | 0.8 | 1 |
| 128 | Glossary of Relevant Genetic and Molecular/Cell Biology. Advances in Experimental Medicine and Biology, 2018, 1085, 23-28. | 0.8 | 0 |
| 129 | Autosomal Dominant Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2018, 1085, 69-77. | 0.8 | 24 |
| 130 | Best Vitelliform Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 79-90. | 0.8 | 8 |
| 131 | Occult Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 103-104. | 0.8 | 4 |
| 132 | Sorsby Pseudoinflammatory Fundus Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 105-108. | 0.8 | 2 |
| 133 | Retinitis Pigmentosa (Non-syndromic). Advances in Experimental Medicine and Biology, 2018, 1085, 125-130. | 0.8 | 41 |
| 134 | Stargardt Disease. Advances in Experimental Medicine and Biology, 2018, 1085, 139-151. | 0.8 | 35 |
| 135 | Optical Coherence Tomography. Advances in Experimental Medicine and Biology, 2018, 1085, 11-13. | 0.8 | 11 |
| 136 | Mitochondrial Disorder: Maternally Inherited Diabetes and Deafness. Advances in Experimental Medicine and Biology, 2018, 1085, 163-165. | 0.8 | 15 |
| 137 | Ciliopathy: AlstrÃ¶m Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 179-180. | 0.8 | 20 |
| 138 | Ciliopathy: SjÃ¶gren-Larsson Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 181-182. | 0.8 | 1 |
| 139 | Inborn Errors of Metabolism: Gyrate Atrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 183-185. | 0.8 | 9 |
| 140 | Inborn Errors of Metabolism: Refsum Disease. Advances in Experimental Medicine and Biology, 2018, 1085, 191-192. | 0.8 | 3 |
| 141 | Inborn Errors of Metabolism: Bietti Crystalline Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 193-195. | 0.8 | 2 |
| 142 | Neurofibromatosis. Advances in Experimental Medicine and Biology, 2018, 1085, 209-211. | 0.8 | 0 |
| 143 | Syphilis. Advances in Experimental Medicine and Biology, 2018, 1085, 219-221. | 0.8 | 2 |
| 144 | Drug-Induced Retinal Toxicity. Advances in Experimental Medicine and Biology, 2018, 1085, 227-232. | 0.8 | 8 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Genetic Testing for Inherited Retinal Dystrophy: Basic Understanding. Advances in Experimental Medicine and Biology, 2018, 1085, 261-268. | 0.8 | 5 |
| 146 | X-linked Choroideremia. Advances in Experimental Medicine and Biology, 2018, 1085, 37-42. | 0.8 | 5 |
| 147 | Rod Monochromatism (Achromatopsia). Advances in Experimental Medicine and Biology, 2018, 1085, 119-123. | 0.8 | 11 |
| 148 | Fundus Autofluorescence. Advances in Experimental Medicine and Biology, 2018, 1085, 15-16. | 0.8 | 7 |
| 149 | Tuberous Sclerosis. Advances in Experimental Medicine and Biology, 2018, 1085, 205-207. | 0.8 | 5 |
| 150 | X-linked Ocular Albinism. Advances in Experimental Medicine and Biology, 2018, 1085, 49-52. | 0.8 | 5 |
| 151 | Fluorescein Angiography. Advances in Experimental Medicine and Biology, 2018, 1085, 7-10. | 0.8 | 6 |
| 152 | Leber Congenital Amaurosis. Advances in Experimental Medicine and Biology, 2018, 1085, 131-137. | 0.8 | 49 |
| 153 | Ciliopathy: Usher Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 167-170. | 0.8 | 27 |
| 154 | Ciliopathy: Bardet-Biedl Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 171-174. | 0.8 | 58 |
| 155 | Autoimmune Retinopathy. Advances in Experimental Medicine and Biology, 2018, 1085, 223-226. | 0.8 | 2 |
| 156 | X-linked Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2018, 1085, 31-35. | 0.8 | 19 |
| 157 | Revolution in Gene Medicine Therapy and Genome Surgery. Genes, 2018, 9, 575. | 1.0 | 25 |
| 158 | Quantitative progression of retinitis pigmentosa by optical coherence tomography angiography. Scientific Reports, 2018, 8, 13130. | 1.6 | 37 |
| 159 | Structural disease progression in PDE6-associated autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2018, 39, 610-614. | 0.5 | 19 |
| 160 | Missense mutation in SLIT2 associated with congenital myopia, anisometropia, connective tissue abnormalities, and obesity. Orphanet Journal of Rare Diseases, 2018, 13, 138. | 1.2 | 7 |
| 161 | Deferoxamine-induced electronegative ERG responses. Documenta Ophthalmologica, 2018, 137, 15-23. | 1.0 | 2 |
| 162 | Congenital grouped albinotic spots of the retinal pigment epithelium in a patient with hemihypertrophy and cafÃ© au lait spots. Documenta Ophthalmologica, 2018, 137, 9-14. | 1.0 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Multimodal characterization of a novel mutation causing vitamin B6-responsive gyrate atrophy. Ophthalmic Genetics, 2018, 39, 512-516. | 0.5 | 9 |
| 164 | Extremely hypomorphic and severe deep intronic variants in the ABCA4 locus result in varying Stargardt disease phenotypes. Journal of Physical Education and Sports Management, 2018, 4, a002733. | 0.5 | 61 |
| 165 | Two-year progression analysis of RPE65 autosomal dominant retinitis pigmentosa. Ophthalmic Genetics, 2018, 39, 544-549. | 0.5 | 24 |
| 166 | Extracellular superoxide dismutase (SOD3) regulates oxidative stress at the vitreoretinal interface. Free Radical Biology and Medicine, 2018, 124, 408-419. | 1.3 | 32 |
| 167 | CRISPR/Cas9 genome surgery for retinal diseases. Drug Discovery Today: Technologies, 2018, 28, 23-32. | 4.0 | 10 |
| 168 | Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. American Journal of Ophthalmology, 2018, 195, 16-25. | 1.7 | 10 |
| 169 | Gene therapy in inherited retinal degenerative diseases, a review. Ophthalmic Genetics, 2018, 39, 560-568. | 0.5 | 55 |
| 170 | Genetic Rescue Reverses Microglial Activation in Preclinical Models of Retinitis Pigmentosa. Molecular Therapy, 2018, 26, 1953-1964. | 3.7 | 16 |
| 171 | Translation of CRISPR Genome Surgery to the Bedside for Retinal Diseases. Frontiers in Cell and Developmental Biology, 2018, 6, 46. | 1.8 | 18 |
| 172 | CRISPR GENOME SURGERY IN THE RETINA IN LIGHT OF OFF-TARGETING. Retina, 2018, 38, 1443-1455. | 1.0 | 11 |
| 173 | Clustered Regularly Interspaced Short Palindromic Repeats-Based Genome Surgery for the Treatment of Autosomal Dominant Retinitis Pigmentosa. Ophthalmology, 2018, 125, 1421-1430. | 2.5 | 100 |
| 174 | <scp>HTRA</scp>1, an ageâ€related macular degeneration protease, processes extracellular matrix proteins <scp>EFEMP</scp>1 and <scp>TSP</scp>1. Aging Cell, 2018, 17, e12710. | 3.0 | 79 |
| 175 | Mutations inGPR143/OA1andABCA4Inform Interpretations of Short-Wavelength and Near-Infrared Fundus Autofluorescence. , 2018, 59, 2459. | | 25 |
| 176 | Quantitative Comparison of Near-infrared Versus Short-wave Autofluorescence Imaging in Monitoring Progression of Retinitis Pigmentosa. American Journal of Ophthalmology, 2018, 194, 120-125. | 1.7 | 19 |
| 177 | Rates of Bone Spicule Pigment Appearance in Patients With Retinitis Pigmentosa Sine Pigmento. American Journal of Ophthalmology, 2018, 195, 176-180. | 1.7 | 12 |
| 178 | Proteomic analysis of the human retina reveals region-specific susceptibilities to metabolic- and oxidative stress-related diseases. PLoS ONE, 2018, 13, e0193250. | 1.1 | 35 |
| 179 | X-linked Juvenile Retinoschisis. Advances in Experimental Medicine and Biology, 2018, 1085, 43-48. | 0.8 | 6 |
| 180 | Progressive Cone Dystrophy and Cone-Rod Dystrophy (XL, AD, and AR). Advances in Experimental Medicine and Biology, 2018, 1085, 53-60. | 0.8 | 20 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | Congenital Stationary Night Blindness. Advances in Experimental Medicine and Biology, 2018, 1085, 61-64. | 0.8 | 10 |
| 182 | Pattern Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 91-96. | 0.8 | 6 |
| 183 | North Carolina Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 109-110. | 0.8 | 1 |
| 184 | Enhanced S-Cone Syndrome (Goldmann-Favre Syndrome). Advances in Experimental Medicine and Biology, 2018, 1085, 153-156. | 0.8 | 12 |
| 185 | Best Vitelliform Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 157-158. | 0.8 | 9 |
| 186 | Mitochondrial Disorder: Kearns-Sayre Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 161-162. | 0.8 | 27 |
| 187 | Ciliopathy: Senior-LÃ¸ken Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 175-178. | 0.8 | 17 |
| 188 | Von Hippel-Lindau Disease. Advances in Experimental Medicine and Biology, 2018, 1085, 201-203. | 0.8 | 5 |
| 189 | Electroretinography. Advances in Experimental Medicine and Biology, 2018, 1085, 17-20. | 0.8 | 14 |
| 190 | A Practical Approach to Retinal Dystrophies. Advances in Experimental Medicine and Biology, 2018, 1085, 245-259. | 0.8 | 6 |
| 191 | Electrooculography. Advances in Experimental Medicine and Biology, 2018, 1085, 21-22. | 0.8 | 2 |
| 192 | Gene therapy and genome surgery in the retina. Journal of Clinical Investigation, 2018, 128, 2177-2188. | 3.9 | 111 |
| 193 | Blue Cone Monochromatism. Advances in Experimental Medicine and Biology, 2018, 1085, 65-66. | 0.8 | 3 |
| 194 | Late-Onset Retinal Degeneration. Advances in Experimental Medicine and Biology, 2018, 1085, 115-116. | 0.8 | 0 |
| 195 | Retinal Histology and Anatomical Landmarks. Advances in Experimental Medicine and Biology, 2018, 1085, 3-5. | 0.8 | 3 |
| 196 | Pigmented Paravenous Chorioretinal Atrophy (PPCRA). Advances in Experimental Medicine and Biology, 2018, 1085, 111-113. | 0.8 | 6 |
| 197 | Rubella Retinopathy. Advances in Experimental Medicine and Biology, 2018, 1085, 215-217. | 0.8 | 1 |
| 198 | Diffuse Unilateral Subacute Neuroretinitis (DUSN). Advances in Experimental Medicine and Biology, 2018, 1085, 239-241. | 0.8 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | Inborn Errors of Metabolism: Pseudoxanthoma Elasticum. Advances in Experimental Medicine and Biology, 2018, 1085, 187-189. | 0.8 | 1 |
| 200 | Acute Zonal Occult Outer Retinopathy (AZOOR) and Related Diseases. Advances in Experimental Medicine and Biology, 2018, 1085, 233-237. | 0.8 | 9 |
| 201 | Doyne Honeycomb Retinal Dystrophy (Malattia Leventinese, Autosomal Dominant Drusen). Advances in Experimental Medicine and Biology, 2018, 1085, 97-102. | 0.8 | 5 |
| 202 | Multimodal analysis of the Preferred Retinal Location and the Transition Zone in patients with Stargardt Disease. Graefe's Archive for Clinical and Experimental Ophthalmology, 2017, 255, 1307-1317. | 1.0 | 14 |
| 203 | Measurement and Reproducibility of Preserved Ellipsoid Zone Area and Preserved Retinal Pigment Epithelium Area in Eyes With Choroideremia. American Journal of Ophthalmology, 2017, 179, 110-117. | 1.7 | 51 |
| 204 | Genetic rescue models refute nonautonomous rod cell death in retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 5259-5264. | 3.3 | 26 |
| 205 | Frequent hypomorphic alleles account for a significant fraction of ABCA4 disease and distinguish it from age-related macular degeneration. Journal of Medical Genetics, 2017, 54, 404-412. | 1.5 | 140 |
| 206 | Peripapillary sparing in RDH12-associated Leber congenital amaurosis. Ophthalmic Genetics, 2017, 38, 575-579. | 0.5 | 23 |
| 207 | â€œDisease in a Dishâ€•Modeling of Retinal Diseases. , 2017, , 107-115. | | 0 |
| 208 | Unexpected mutations after CRISPRâ€“Cas9 editing in vivo. Nature Methods, 2017, 14, 547-548. | 9.0 | 294 |
| 209 | Evaluating Structural Progression of Retinitis Pigmentosa After Cataract Surgery. American Journal of Ophthalmology, 2017, 180, 117-123. | 1.7 | 18 |
| 210 | Proteomic analysis of elevated intraocular pressure with retinal detachment. American Journal of Ophthalmology Case Reports, 2017, 5, 107-110. | 0.4 | 10 |
| 211 | Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. European Journal of Human Genetics, 2017, 25, 735-743. | 1.4 | 31 |
| 212 | CRISPR applications in ophthalmologic genome surgery. Current Opinion in Ophthalmology, 2017, 28, 252-259. | 1.3 | 27 |
| 213 | Two pathways of rod photoreceptor cell death induced by elevated cGMP. Human Molecular Genetics, 2017, 26, 2299-2306. | 1.4 | 49 |
| 214 | Structural modeling of a novel <scp>SLC</scp>38A8 mutation that causes foveal hypoplasia. Molecular Genetics & Genomic Medicine, 2017, 5, 202-209. | 0.6 | 23 |
| 215 | Achromatopsia mutations target sequential steps of ATF6 activation. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 400-405. | 3.3 | 50 |
| 216 | Quantitative Autofluorescence Intensities in Acute Zonal Occult Outer Retinopathy vs Healthy Eyes. JAMA Ophthalmology, 2017, 135, 1330. | 1.4 | 24 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | CRISPR-Mediated Ophthalmic Genome Surgery. Current Ophthalmology Reports, 2017, 5, 199-206. | 0.5 | 12 |
| 218 | Electroretinography Reveals Difference in Cone Function between Syndromic and Nonsyndromic USH2A Patients. Scientific Reports, 2017, 7, 11170. | 1.6 | 26 |
| 219 | Retrospective Analysis of Structural Disease Progression in Retinitis Pigmentosa Utilizing Multimodal Imaging. Scientific Reports, 2017, 7, 10347. | 1.6 | 46 |
| 220 | Gene Therapy Restores Mfrp and Corrects Axial Eye Length. Scientific Reports, 2017, 7, 16151. | 1.6 | 41 |
| 221 | Viral Vectors, Engineered Cells and the CRISPR Revolution. Advances in Experimental Medicine and Biology, 2017, 1016, 3-27. | 0.8 | 15 |
| 222 | CRISPR in the Retina: Evaluation of Future Potential. Advances in Experimental Medicine and Biology, 2017, 1016, 147-155. | 0.8 | 3 |
| 223 | Dissection of Human Retina and RPE-Choroid for Proteomic Analysis. Journal of Visualized Experiments, 2017, , . | 0.2 | 5 |
| 224 | PHENOTYPING CHOROIDEREMIA AND ITS CARRIER STATE WITH MULTIMODAL IMAGING TECHNIQUES. Retinal Cases and Brief Reports, 2017, 11, S178-S181. | 0.3 | 13 |
| 225 | Correction of Monogenic and Common Retinal Disorders with Gene Therapy. Genes, 2017, 8, 53. | 1.0 | 37 |
| 226 | Stem Cell Therapies in Retinal Disorders. Cells, 2017, 6, 4. | 1.8 | 35 |
| 227 | Patient-specific mutations impair BESTROPHIN1â€™s essential role in mediating Ca2+-dependent Cl- currents in human RPE. ELife, 2017, 6, . | 2.8 | 43 |
| 228 | Efficacy of rituximab in non-paraneoplastic autoimmune retinopathy. Orphanet Journal of Rare Diseases, 2017, 12, 129. | 1.2 | 13 |
| 229 | Calpain-5 gene expression in the mouse eye and brain. BMC Research Notes, 2017, 10, 602. | 0.6 | 5 |
| 230 | A Comparison of En Face Optical Coherence Tomography and Fundus Autofluorescence in Stargardt Disease. , 2017, 58, 5227. | | 25 |
| 231 | CRISPR-Cas Genome Surgery in Ophthalmology. Translational Vision Science and Technology, 2017, 6, 13. | 1.1 | 15 |
| 232 | Quantifying Fundus Autofluorescence in Patients With Retinitis Pigmentosa. , 2017, 58, 1843. | | 56 |
| 233 | Therapeutic drug repositioning using personalized proteomics of liquid biopsies. JCI Insight, 2017, 2, . | 2.3 | 27 |
| 234 | Patients and animal models of CNGÎ²1-deficient retinitis pigmentosa support gene augmentation approach. Journal of Clinical Investigation, 2017, 128, 190-206. | 3.9 | 48 |

| # | Article | IF | Citations |
|---|---|---|---|
| 235 | ERG and OCT findings of a patient with a clinical diagnosis of occult macular dystrophy in a patient of Ashkenazi Jewish descent associated with a novel mutation in the gene encoding RP1L1. BMJ Case Reports, 2017, 2017, bcr-2016-218364. | 0.2 | 3 |
| 236 | Genome Surgery and Gene Therapy in Retinal Disorders. Yale Journal of Biology and Medicine, 2017, 90, 523-532. | 0.2 | 9 |
| 237 | Calpain-5 Expression in the Retina Localizes to Photoreceptor Synapses. , 2016, 57, 2509. | | 29 |
| 238 | Quantitative Autofluorescence andABCA4Disease. , 2016, 57, 3297. | | 0 |
| 239 | Simultaneous Expression of ABCA4 and GPR143 Mutations: A Complex Phenotypic Manifestation. , 2016, 57, 3409. | | 6 |
| 240 | Secondary glaucoma in <em>CAPN5</em>-associated neovascular inflammatory vitreoretinopathy. Clinical Ophthalmology, 2016, Volume 10, 1187-1197. | 0.9 | 10 |
| 241 | Complication of Autologous Stem Cell Transplantation in Retinitis Pigmentosa. JAMA Ophthalmology, 2016, 134, 711. | 1.4 | 18 |
| 242 | CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa. Molecular Therapy, 2016, 24, 1388-1394. | 3.7 | 93 |
| 243 | BESTROPHIN1mutations cause defective chloride conductance in patient stem cell-derived RPE. Human Molecular Genetics, 2016, 25, ddw126. | 1.4 | 43 |
| 244 | Laser-Induced Photic Injury Phenocopies Macular Dystrophy. Ophthalmic Genetics, 2016, 37, 59-67. | 0.5 | 30 |
| 245 | Small-angle X-ray scattering of calpain-5 reveals a highly open conformation among calpains. Journal of Structural Biology, 2016, 196, 309-318. | 1.3 | 12 |
| 246 | Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. Human Molecular Genetics, 2016, 25, 4201-4210. | 1.4 | 10 |
| 247 | Reprogramming towards anabolism impedes degeneration in a preclinical model of retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 4244-4255. | 1.4 | 30 |
| 248 | Gene and cellâ€based therapies for inherited retinal disorders: An update. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 349-366. | 0.7 | 60 |
| 249 | Neuroretinal hypoxic signaling in a new preclinical murine model for proliferative diabetic retinopathy. Signal Transduction and Targeted Therapy, 2016, 1, . | 7.1 | 23 |
| 250 | Precision Medicine: Genetic Repair of Retinitis Pigmentosa in Patient-Derived Stem Cells. Scientific Reports, 2016, 6, 19969. | 1.6 | 135 |
| 251 | MULTIMODAL IMAGING OF DISEASE-ASSOCIATED PIGMENTARY CHANGES IN RETINITIS PIGMENTOSA. Retina, 2016, 36, S147-S158. | 1.0 | 26 |
| 252 | Precision Medicine. JAMA Ophthalmology, 2016, 134, 444. | 1.4 | 54 |

| # | Article | IF | Citations |
|---|---|---|---|
| 253 | Phototransduction Influences Metabolic Flux and Nucleotide Metabolism in Mouse Retina. Journal of Biological Chemistry, 2016, 291, 4698-4710. | 1.6 | 87 |
| 254 | Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19. | 1.8 | 39 |
| 255 | Photopsia and a temporal visual field defect. Survey of Ophthalmology, 2016, 61, 363-367. | 1.7 | 2 |
| 256 | Personalized Medicine: Cell and Gene Therapy Based on Patient-Specific iPSC-Derived Retinal Pigment Epithelium Cells. Advances in Experimental Medicine and Biology, 2016, 854, 549-555. | 0.8 | 26 |
| 257 | Reprogramming metabolism by targeting sirtuin 6 attenuates retinal degeneration. Journal of Clinical Investigation, 2016, 126, 4659-4673. | 3.9 | 82 |
| 258 | Quantitative Fundus Autofluorescence and Optical Coherence Tomography inABCA4Carriers. , 2015, 56, 7274. | | 28 |
| 259 | Quantitative Fundus Autofluorescence and Optical Coherence Tomography inPRPH2/RDS-andABCA4-Associated Disease Exhibiting Phenotypic Overlap. , 2015, 56, 3159. | | 56 |
| 260 | Patient-Specific iPSC-Derived RPE for Modeling of Retinal Diseases. Journal of Clinical Medicine, 2015, 4, 567-578. | 1.0 | 26 |
| 261 | Structural Modeling of a Novel CAPN5 Mutation that Causes Uveitis and Neovascular Retinal Detachment. PLoS ONE, 2015, 10, e0122352. | 1.1 | 35 |
| 262 | Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765. | 9.4 | 183 |
| 263 | Quantitative Autofluorescence as a Clinical Tool for Expedited Differential Diagnosis of Retinal Degeneration. JAMA Ophthalmology, 2015, 133, 219. | 1.4 | 8 |
| 264 | Personalized therapeutic strategies for patients with retinitis pigmentosa. Expert Opinion on Biological Therapy, 2015, 15, 391-402. | 1.4 | 43 |
| 265 | CAPN5mutation in hereditary uveitis: the R243L mutation increases calpain catalytic activity and triggers intraocular inflammation in a mouse model. Human Molecular Genetics, 2015, 24, 4584-4598. | 1.4 | 39 |
| 266 | Rapid resolution of retinoschisis with acetazolamide. Documenta Ophthalmologica, 2015, 131, 63-70. | 1.0 | 15 |
| 267 | BEST1: the Best Target for Gene and Cell Therapies. Molecular Therapy, 2015, 23, 1805-1809. | 3.7 | 38 |
| 268 | Multimodal Imaging of Central Retinal Disease Progression in a 2-Year Mean Follow-up of Retinitis Pigmentosa. American Journal of Ophthalmology, 2015, 160, 786-798.e4. | 1.7 | 81 |
| 269 | Bilateral Concordance of the Fundus Hyperautofluorescent Ring in Typical Retinitis Pigmentosa Patients. Ophthalmic Genetics, 2015, 36, 113-122. | 0.5 | 22 |
| 270 | Rod metabolic demand drives progression in retinopathies. Taiwan Journal of Ophthalmology, 2015, 5, 105-108. | 0.3 | 14 |

| # | Article | IF | Citations |
|---|---|---|---|
| 271 | Skin Biopsy and Patient-Specific Stem Cell Lines. Methods in Molecular Biology, 2015, 1353, 77-88. | 0.4 | 14 |
| 272 | Quantitative Fundus Autofluorescence Distinguishes ABCA4-Associated and Nonâ€“ABCA4-Associated Bull's-Eye Maculopathy. Ophthalmology, 2015, 122, 345-355. | 2.5 | 75 |
| 273 | Differentiation of hypothalamic-like neurons from human pluripotent stem cells. Journal of Clinical Investigation, 2015, 125, 796-808. | 3.9 | 112 |
| 274 | Halting progressive neurodegeneration in advanced retinitis pigmentosa. Journal of Clinical Investigation, 2015, 125, 3704-3713. | 3.9 | 68 |
| 275 | Emerging Treatments for Retinitis Pigmentosa: Genes and stem cells, as well as new electronic and medical therapies, are gaining ground. Retinal Physician, 2015, 12, 52-70. | 3.0 | 17 |
| 276 | Quantitative Fundus Autofluorescence and Optical Coherence Tomography in Best Vitelliform Macular Dystrophy. , 2014, 55, 1471. | | 89 |
| 277 | Quantitative Fundus Autofluorescence in Recessive Stargardt Disease. , 2014, 55, 2841. | | 160 |
| 278 | Vigabatrin Retinal Toxicity First Detected with Electroretinographic Changes: A Case Report. Journal of Clinical & Experimental Ophthalmology, 2014, 05, . | 0.1 | 4 |
| 279 | Retinal Damage in Chloroquine Maculopathy, Revealed by High Resolution Imaging: A Case Report Utilizing Adaptive Optics Scanning Laser Ophthalmoscopy. Korean Journal of Ophthalmology: KJO, 2014, 28, 100. | 0.5 | 16 |
| 280 | The External Limiting Membrane in Early-Onset Stargardt Disease. , 2014, 55, 6139. | | 54 |
| 281 | Functional validation of a human CAPN5 exome variant by lentiviral transduction into mouse retina. Human Molecular Genetics, 2014, 23, 2665-2677. | 1.4 | 35 |
| 282 | Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806. | 1.4 | 117 |
| 283 | Structural and Genetic Assessment of theABCA4-Associated Optical Gap Phenotype. , 2014, 55, 7217. | | 30 |
| 284 | Choroidal and Retinal Thickening in Severe Preeclampsia. , 2014, 55, 5723. | | 35 |
| 285 | General Pathophysiology in Retinal Degeneration. Developments in Ophthalmology, 2014, 53, 33-43. | 0.1 | 74 |
| 286 | MULTIMODAL IMAGING IN A CASE OF DEFEROXAMINE-INDUCED MACULOPATHY. Retinal Cases and Brief Reports, 2014, 8, 306-309. | 0.3 | 15 |
| 287 | Next-generation Sequencing Revealed a Novel Mutation in the Gene Encoding the Beta Subunit of Rod Phosphodiesterase. Ophthalmic Genetics, 2014, 35, 142-150. | 0.5 | 17 |
| 288 | Spectral-Domain Optical Coherence Tomography Staging and Autofluorescence Imaging in Achromatopsia. JAMA Ophthalmology, 2014, 132, 437. | 1.4 | 58 |

| # | Article | IF | Citations |
|---|---|---|---|
| 289 | Whole Exome Sequencing Identifies CRB1 Defect in an Unusual Maculopathy Phenotype. Ophthalmology, 2014, 121, 1773-1782. | 2.5 | 62 |
| 290 | Disease progression in autosomal dominant coneâ€“rod dystrophy caused by a novel mutation (D100G) in the GUCA1A gene. Documenta Ophthalmologica, 2014, 128, 59-67. | 1.0 | 34 |
| 291 | Validation of genome-wide association study (GWAS)-identified disease risk alleles with patient-specific stem cell lines. Human Molecular Genetics, 2014, 23, 3445-3455. | 1.4 | 86 |
| 292 | The Role of Fundus Autofluorescence in Late-Onset Retinitis Pigmentosa (LORP) Diagnosis. Ophthalmic Genetics, 2014, 35, 170-179. | 0.5 | 7 |
| 293 | Mid-stage intervention achieves similar efficacy as conventional early-stage treatment using gene therapy in a pre-clinical model of retinitis pigmentosa. Human Molecular Genetics, 2014, 23, 514-523. | 1.4 | 35 |
| 294 | Correlations Among Near-Infrared and Short-Wavelength Autofluorescence and Spectral-Domain Optical Coherence Tomography in Recessive Stargardt Disease. Investigative Ophthalmology and Visual Science, 2014, 55, 8134-8143. | 3.3 | 69 |
| 295 | A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214. | 13.5 | 322 |
| 296 | New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780. | 1.4 | 30 |
| 297 | A novel RPGR mutation masquerading as Stargardt disease. British Journal of Ophthalmology, 2014, 98, 709-711. | 2.1 | 21 |
| 298 | Gene Therapy in Patient-specific Stem Cell Lines and a Preclinical Model of Retinitis Pigmentosa With Membrane Frizzled-related Protein Defects. Molecular Therapy, 2014, 22, 1688-1697. | 3.7 | 80 |
| 299 | Cone Photoreceptor Abnormalities Correlate With Vision Loss in a Case of Acute Posterior Multifocal Placoid Pigment Epitheliopathy. Ophthalmic Surgery Lasers and Imaging Retina, 2014, 45, 74-78. | 0.4 | 5 |
| 300 | Early Structural Anomalies Observed by High-Resolution Imaging in Two Related Cases of Autosomal-Dominant Retinitis Pigmentosa. Ophthalmic Surgery Lasers and Imaging Retina, 2014, 45, 469-473. | 0.4 | 21 |
| 301 | Hyperautofluorescent Macular Ring in a Series of Patients With Enhanced S-Cone Syndrome. Ophthalmic Surgery Lasers and Imaging Retina, 2014, 45, 592-595. | 0.4 | 7 |
| 302 | Silencing of tuberin enhances photoreceptor survival and function in a preclinical model of retinitis pigmentosa (an american ophthalmological society thesis). Transactions of the American Ophthalmological Society, 2014, 112, 103-15. | 1.4 | 12 |
| 303 | Disruption of the human cone photoreceptor mosaic from a defect in NR2E3 transcription factor function in young adults. Graefe's Archive for Clinical and Experimental Ophthalmology, 2013, 251, 2299-2309. | 1.0 | 22 |
| 304 | Abnormality in the external limiting membrane in early Stargardt Disease. Ophthalmic Genetics, 2013, 34, 75-77. | 0.5 | 27 |
| 305 | Evaluation of multimodal imaging in carriers of X-linked retinitis pigmentosa. Experimental Eye Research, 2013, 113, 41-48. | 1.2 | 45 |
| 306 | Induced pluripotent stem cells and retinal degeneration treatment. Expert Review of Ophthalmology, 2013, 8, 5-8. | 0.3 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 307 | Cellular imaging demonstrates genetic mosaicism in heterozygous carriers of an X-linked ciliopathy gene. European Journal of Human Genetics, 2013, 21, 1240-1248. | 1.4 | 22 |
| 308 | Gene therapy provides long-term visual function in a pre-clinical model of retinitis pigmentosa. Human Molecular Genetics, 2013, 22, 558-567. | 1.4 | 54 |
| 309 | Comparison of Near-Infrared and Short-Wavelength Autofluorescence in Retinitis Pigmentosa. , 2013, 54, 585. | | 83 |
| 310 | Therapeutic Margins in a Novel Preclinical Model of Retinitis Pigmentosa. Journal of Neuroscience, 2013, 33, 13475-13483. | 1.7 | 33 |
| 311 | Disruption in Bruch membrane in patients with Stargardt disease. Ophthalmic Genetics, 2012, 33, 49-52. | 0.5 | 18 |
| 312 | Subretinal Injection of Gene Therapy Vectors and Stem Cells in the Perinatal Mouse Eye. Journal of Visualized Experiments, 2012, , . | 0.2 | 24 |
| 313 | Unilateral Retinitis Pigmentosa: A Proposal of Genetic Pathogenic Mechanisms. European Journal of Ophthalmology, 2012, 22, 654-660. | 0.7 | 27 |
| 314 | STRUCTURAL AND FUNCTIONAL CHANGES ASSOCIATED WITH NORMAL AND ABNORMAL FUNDUS AUTOFLUORESCENCE IN PATIENTS WITH RETINITIS PIGMENTOSA. Retina, 2012, 32, 349-357. | 1.0 | 57 |
| 315 | Autofluorescence Imaging and Spectral-Domain Optical Coherence Tomography in Incomplete Congenital Stationary Night Blindness and Comparison With Retinitis Pigmentosa. American Journal of Ophthalmology, 2012, 153, 143-154.e2. | 1.7 | 34 |
| 316 | Progressive Constriction of the Hyperautofluorescent Ring in Retinitis Pigmentosa. American Journal of Ophthalmology, 2012, 153, 718-727.e2. | 1.7 | 75 |
| 317 | Mice with a D190N Mutation in the Gene Encoding Rhodopsin: A Model for Human Autosomal-Dominant Retinitis Pigmentosa. Molecular Medicine, 2012, 18, 549-555. | 1.9 | 21 |
| 318 | Long-term Safety and Efficacy of Human-Induced Pluripotent Stem Cell (iPS) Grafts in a Preclinical Model of Retinitis Pigmentosa. Molecular Medicine, 2012, 18, 1312-1319. | 1.9 | 162 |
| 319 | Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2012, 53, 4458. | | 81 |
| 320 | Effect of the ILE86TER mutation in the Î³ subunit of cGMP phosphodiesterase (PDE6) on rod photoreceptor signaling. Cellular Signalling, 2012, 24, 181-188. | 1.7 | 9 |
| 321 | Vigabatrin-Induced Retinal Toxicity Is Partially Mediated by Signaling in Rod and Cone Photoreceptors. PLoS ONE, 2012, 7, e43889. | 1.1 | 22 |
| 322 | Functional Analysis of Retinal Flecks in Stargardt Disease. Journal of Clinical & Experimental Ophthalmology, 2012, 03, . | 0.1 | 17 |
| 323 | Familial discordance in Stargardt disease. Molecular Vision, 2012, 18, 227-33. | 1.1 | 15 |
| 324 | Allelic and phenotypic heterogeneity inABCA4mutations. Ophthalmic Genetics, 2011, 32, 165-174. | 0.5 | 85 |

| # | Article | IF | Citations |
|---|---|---|---|
| 325 | Quantification of Peripapillary Sparing and Macular Involvement in Stargardt Disease (STGD1). , 2011, 52, 8006. | | 45 |
| 326 | Mouse Eye Enucleation for Remote High-throughput Phenotyping. Journal of Visualized Experiments, 2011, , . | 0.2 | 18 |
| 327 | shRNA knockdown ofâ€‚guanylate cyclase 2eâ€‚,orâ€‚,cyclic nucleotide gated channel alpha 1â€‚,increases photoreceptor survival in a cGMPâ€‚,phosphodiesteraseâ€‚,mouse model of retinitis pigmentosa. Journal of Cellular and Molecular Medicine, 2011, 15, 1778-1787. | 1.6 | 25 |
| 328 | Function of the asparagine 74 residue of the inhibitory Î³-subunit of retinal rod cGMP-phophodiesterase (PDE) in vivo. Cellular Signalling, 2011, 23, 1584-1589. | 1.7 | 5 |
| 329 | Autofluorescence Imaging in a Case of Benign Familial Fleck Retina. JAMA Ophthalmology, 2011, 125, 714. | 2.6 | 12 |
| 330 | Lentivirus-mediated expression of cDNA and shRNA slows degeneration in retinitis pigmentosa. Experimental Biology and Medicine, 2011, 236, 1211-1217. | 1.1 | 26 |
| 331 | Transplantation of Reprogrammed Embryonic Stem Cells Improves Visual Function in a Mouse Model for Retinitis Pigmentosa. Transplantation, 2010, 89, 911-919. | 0.5 | 71 |
| 332 | FUNDUS AUTOFLUORESCENCE AND OPTICAL COHERENCE TOMOGRAPHY OF CONGENITAL GROUPED ALBINOTIC SPOTS. Retina, 2010, 30, 1217-1222. | 1.0 | 10 |
| 333 | Rapid and Noninvasive Imaging of Retinal Ganglion Cells in Live Mouse Models of Glaucoma. Molecular Imaging and Biology, 2010, 12, 386-393. | 1.3 | 6 |
| 334 | Loss of peripapillary sparing in non-group I Stargardt disease. Experimental Eye Research, 2010, 91, 592-600. | 1.2 | 23 |
| 335 | Transplantation of Reprogrammed Embryonic Stem Cells Improves Visual Function in a Mouse Model for Retinitis Pigmentosa. Annals of Neurosciences, 2010, 17, 185-6. | 0.9 | 0 |
| 336 | A Comparison of Fundus Autofluorescence and Retinal Structure in Patients with Stargardt Disease. , 2009, 50, 3953. | | 128 |
| 337 | Autofluorescence Imaging in Rubella Retinopathy. Ocular Immunology and Inflammation, 2009, 17, 400-402. | 1.0 | 9 |
| 338 | Fundus autofluorescence in cone dystrophy. Documenta Ophthalmologica, 2009, 119, 141-144. | 1.0 | 23 |
| 339 | G1961E mutant allele in the Stargardt disease gene ABCA4 causes bull's eye maculopathy. Experimental Eye Research, 2009, 89, 16-24. | 1.2 | 90 |
| 340 | Light-dependent phosphorylation of the gamma subunit of cGMP-phophodiesterase (PDE6Î³) at residue threonine 22 in intact photoreceptor neurons. Biochemical and Biophysical Research Communications, 2009, 390, 1149-1153. | 1.0 | 12 |
| 341 | Cellular and Molecular Origin of Circumpapillary Dysgenesis of the Pigment Epithelium. Ophthalmology, 2009, 116, 971-980. | 2.5 | 6 |
| 342 | Case Report: Autofluorescence Imaging and Phenotypic Variance in a Sibling Pair with Early-Onset Retinal Dystrophy Due to Defective CRB1 Function. Current Eye Research, 2009, 34, 395-400. | 0.7 | 19 |

| # | Article | IF | Citations |
|---|---|---|---|
| 343 | STRUCTURAL ASSESSMENT OF HYPERAUTOFLUORESCENT RING IN PATIENTS WITH RETINITIS PIGMENTOSA. Retina, 2009, 29, 1025-1031. | 1.0 | 98 |
| 344 | Electronegative electroretinogram associated with topiramate toxicity and vitelliform maculopathy. Documenta Ophthalmologica, 2008, 116, 57-60. | 1.0 | 17 |
| 345 | Non-vascular vision loss in pseudoxanthoma elasticum. Documenta Ophthalmologica, 2008, 117, 65-67. | 1.0 | 9 |
| 346 | A Novel Mutation and Phenotypes in Phosphodiesterase 6 Deficiency. American Journal of Ophthalmology, 2008, 146, 780-788.e1. | 1.7 | 51 |
| 347 | Phenotype-Genotype Correlations in Autosomal Dominant Retinitis Pigmentosa Caused by RHO, D190N. Current Eye Research, 2008, 33, 1014-1022. | 0.7 | 20 |
| 348 | Modulation of Phosphodiesterase6 Turnoff during Background Illumination in Mouse Rod Photoreceptors. Journal of Neuroscience, 2008, 28, 2064-2074. | 1.7 | 59 |
| 349 | PREFERRED RETINAL LOCUS IN MACULAR DISEASE. Retina, 2008, 28, 1234-1240. | 1.0 | 61 |
| 350 | Functional Rescue of Degenerating Photoreceptors in Mice Homozygous for a Hypomorphic cGMP Phosphodiesterase 6 b Allele (Pde6b <sup>H620Q</sup>). , 2008, 49, 5067. | | 57 |
| 351 | Novel Phenotypic and Genotypic Findings in X-Linked Retinoschisis. JAMA Ophthalmology, 2007, 125, 259. | 2.6 | 62 |
| 352 | Transgenic mice carrying theH258N mutation in the gene encoding the Î²-subunit of phosphodiesterase-6 (PDE6B) provide a model for human congenital stationary night blindness. Human Mutation, 2007, 28, 243-254. | 1.1 | 40 |
| 353 | Stationary night blindness or progressive retinal degeneration in mice carrying different alleles of PDE gamma. Frontiers in Bioscience - Landmark, 2003, 8, s666-675. | 3.0 | 12 |
| 354 | The positive role of the carboxyl terminus of the Î³ subunit of retinal cGMP-phosphodiesterase in maintaining phosphodiesterase activity in vivo. Vision Research, 2002, 42, 439-445. | 0.7 | 17 |
| 355 | In vivo studies of the Î³ subunit of retinal cGMP-phophodiesterase with a substitution of tyrosine-84. Biochemical Journal, 2001, 353, 467. | 1.7 | 13 |
| 356 | In vivo studies of the Î³ subunit of retinal cGMP-phophodiesterase with a substitution of tyrosine-84. Biochemical Journal, 2001, 353, 467-474. | 1.7 | 13 |