## Stephen H Tsang

# List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

342 6,728 papers citations

44 h-index 65 g-index

361 ext. papers

8,127 ext. citations

**5.1** avg, IF

6.24 L-index

#	Paper	IF	Citations
342	Comparisons Among Optical Coherence Tomography and Fundus Autofluorescence Modalities as Measurements of Atrophy in ABCA4-Associated Disease <i>Translational Vision Science and Technology</i> , <b>2022</b> , 11, 36	3.3	
341	Late-stage rescue of visually guided behavior in the context of a significantly remodeled retinitis pigmentosa mouse model <i>Cellular and Molecular Life Sciences</i> , <b>2022</b> , 79, 148	10.3	
340	Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease <i>PLoS Genetics</i> , <b>2022</b> , 18, e1010129	6	
339	Expanding the phenotype of TTLL5-associated retinal dystrophy: a case series <i>Orphanet Journal of Rare Diseases</i> , <b>2022</b> , 17, 146	4.2	1
338	Multimodal imaging reveals retinoschisis masquerading as retinal detachment in patients with choroideremia <i>American Journal of Ophthalmology Case Reports</i> , <b>2022</b> , 26, 101543	1.3	
337	Clinical and genetic findings in Italian patients with sector retinitis pigmentosa. <i>Molecular Vision</i> , <b>2021</b> , 27, 78-94	2.3	O
336	Phase transition specified by a binary code patterns the vertebrate eye cup. <i>Science Advances</i> , <b>2021</b> , 7, eabj9846	14.3	3
335	Telegenetics for inherited retinal diseases in the COVID-19 environment. <i>International Journal of Retina and Vitreous</i> , <b>2021</b> , 7, 25	2.9	1
334	Bardet-Biedl syndrome proteins regulate intracellular signaling and neuronal function in patient-specific iPSC-derived neurons. <i>Journal of Clinical Investigation</i> , <b>2021</b> , 131,	15.9	6
333	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> , <b>2021</b> , 30, 1293-1304	5.6	5
332	A novel KCNV2 mutation in a patient taking hydroxychloroquine associated with cone dystrophy with supernormal rod response. <i>Ophthalmic Genetics</i> , <b>2021</b> , 42, 458-463	1.2	O
331	CNGB1-related rod-cone dystrophy: A mutation review and update. <i>Human Mutation</i> , <b>2021</b> , 42, 641-666	4.7	6
330	Impaired cholesterol efflux in retinal pigment epithelium of individuals with juvenile macular degeneration. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 903-918	11	3
329	Overcoming translational barriers in modeling macular degenerations. <i>Cell Stem Cell</i> , <b>2021</b> , 28, 781-783	18	O
328	Distinct expression requirements and rescue strategies for loss- and gain-of-function mutations. <i>ELife</i> , <b>2021</b> , 10,	8.9	2
327	Reply. <i>Ophthalmology Retina</i> , <b>2021</b> , 5, e7-e8	3.8	
326	Shared Features in Retinal Disorders With Involvement of Retinal Pigment Epithelium <b>2021</b> , 62, 15		1

#### (2020-2021)

325	Stage-dependent choriocapillaris impairment in Best vitelliform macular dystrophy characterized by optical coherence tomography angiography. <i>Scientific Reports</i> , <b>2021</b> , 11, 14300	4.9	1
324	Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. <i>Ophthalmic Genetics</i> , <b>2021</b> , 42, 664-673	1.2	5
323	CHORIORETINAL CHANGES IN A GENETICALLY CONFIRMED CASE OF BOUCHER-NEUHÜSER SYNDROME. <i>Retinal Cases and Brief Reports</i> , <b>2021</b> , 15, 179-184	1.1	7
322	Whole-Exome Sequencing of Patients With Posterior Segment Uveitis. <i>American Journal of Ophthalmology</i> , <b>2021</b> , 221, 246-259	4.9	3
321	Retinal pigment epithelium lipid metabolic demands and therapeutic restoration. <i>Taiwan Journal of Ophthalmology</i> , <b>2021</b> , 11, 216-220	1.4	
320	Retinal Pigment Epithelium Atrophy in Recessive Stargardt Disease as Measured by Short-Wavelength and Near-Infrared Autofluorescence. <i>Translational Vision Science and Technology</i> , <b>2021</b> , 10, 3	3.3	4
319	Nutrigenetic reprogramming of oxidative stress. <i>Taiwan Journal of Ophthalmology</i> , <b>2021</b> , 11, 207-215	1.4	1
318	Central serous chorioretinopathy treatment with a systemic PDE5 and PDE6 inhibitor (sildenafil). <i>American Journal of Ophthalmology Case Reports</i> , <b>2021</b> , 21, 100998	1.3	1
317	PROGRESSION OF SCOTOPIC SINGLE-FLASH ELECTRORETINOGRAPHY IN THE STAGES OF CAPN5 VITREORETINOPATHY. <i>Retinal Cases and Brief Reports</i> , <b>2021</b> , 15, 473-478	1.1	4
316	Gene therapy for inherited retinal diseases. <i>Annals of Translational Medicine</i> , <b>2021</b> , 9, 1278	3.2	5
315	Precision Medicine Trials in Retinal Degenerations. <i>Annual Review of Vision Science</i> , <b>2021</b> , 7, 851-865	8.2	1
314	Stickler Syndrome Genotype (COL2A1 mutation) with Retinitis Pigmentosa Phenotype. <i>Ophthalmology Retina</i> , <b>2020</b> , 4, 522	3.8	2
313	Sequential multiple retinal vein occlusions and transient ischemic attack in MTHFR polymorphism and protein S deficiency. <i>Molecular Genetics &amp; Enomic Medicine</i> , <b>2020</b> , 8, e1273	2.3	2
312	PKM2 ablation enhanced retinal function and survival in a preclinical model of retinitis pigmentosa. <i>Mammalian Genome</i> , <b>2020</b> , 31, 77-85	3.2	4
311	Phenotypic variance in Calpain-5 retinal degeneration. <i>American Journal of Ophthalmology Case Reports</i> , <b>2020</b> , 18, 100627	1.3	3
310	Optical coherence tomography in the evaluation of retinitis pigmentosa. <i>Ophthalmic Genetics</i> , <b>2020</b> , 41, 413-419	1.2	1
309	Stargardt Juvenile Macular Degeneration. New England Journal of Medicine, 2020, 382, 2353	59.2	
308	Quasidominance in autosomal recessive -Leber congenital amaurosis. <i>Ophthalmic Genetics</i> , <b>2020</b> , 41, 198-200	1.2	

307	Phenotypic expansion of autosomal dominant retinitis pigmentosa associated with the D477G mutation in. <i>Journal of Physical Education and Sports Management</i> , <b>2020</b> , 6,	2.8	7
306	Prospective Impact of Sildenafil on Chronic cEntral Serous Chorioretinopathy: PISCES Trial. <i>Ophthalmology Retina</i> , <b>2020</b> , 4, 1119-1123	3.8	6
305	Short-Wavelength and Near-Infrared Autofluorescence in Patients with Deficiencies of the Visual Cycle and Phototransduction. <i>Scientific Reports</i> , <b>2020</b> , 10, 8998	4.9	3
304	Metabolite therapy guided by liquid biopsy proteomics delays retinal neurodegeneration. <i>EBioMedicine</i> , <b>2020</b> , 52, 102636	8.8	15
303	Disease asymmetry and hyperautofluorescent ring shape in retinitis pigmentosa patients. <i>Scientific Reports</i> , <b>2020</b> , 10, 3364	4.9	1
302	Progressive RPE atrophy and photoreceptor death in -associated autosomal recessive retinitis pigmentosa. <i>Ophthalmic Genetics</i> , <b>2020</b> , 41, 26-30	1.2	2
301	Inhibition of Ca channel surface expression by mutant bestrophin-1 in RPE cells. <i>FASEB Journal</i> , <b>2020</b> , 34, 4055-4071	0.9	6
300	Comparative Analysis of Functional and Structural Decline in Retinitis Pigmentosas. <i>International Journal of Molecular Sciences</i> , <b>2020</b> , 21,	6.3	2
299	Multiexon deletion alleles of ATF6 linked to achromatopsia. JCI Insight, 2020, 5,	9.9	3
298	Precision metabolome reprogramming for imprecision therapeutics in retinitis pigmentosa. <i>Journal of Clinical Investigation</i> , <b>2020</b> , 130, 3971-3973	15.9	5
297	Fundoscopy-directed genetic testing to re-evaluate negative whole exome sequencing results. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 32	4.2	3
296	Optical Gap Biomarker in Cone-Dominant Retinal Dystrophy. <i>American Journal of Ophthalmology</i> , <b>2020</b> , 218, 40-53	4.9	2
295	Differences in Intraretinal Pigment Migration Across Inherited Retinal Dystrophies. <i>American Journal of Ophthalmology</i> , <b>2020</b> , 217, 252-260	4.9	2
294	Effects of deficiency in the -encoded visual cycle protein CRALBP on visual dysfunction in humans and mice. <i>Journal of Biological Chemistry</i> , <b>2020</b> , 295, 6767-6780	5.4	15
293	Therapy in Rhodopsin-Mediated Autosomal Dominant Retinitis Pigmentosa. <i>Molecular Therapy</i> , <b>2020</b> , 28, 2139-2149	11.7	8
292	Retinal Manifestations of Mitochondrial Oxidative Phosphorylation Disorders <b>2020</b> , 61, 12		3
291	Compound heterozygous inheritance of two novel COQ2 variants results in familial coenzyme Q deficiency. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 320	4.2	2
290	Treatment-Emergent Adverse Events in Gene Therapy Trials for Inherited Retinal Diseases: A Narrative Review. <i>Ophthalmology and Therapy</i> , <b>2020</b> , 9, 709-724	5	12

289	Quantitative Autofluorescence Following Gene Therapy With Voretigene Neparvovec. <i>JAMA Ophthalmology</i> , <b>2020</b> , 138, 919-921	3.9	4
288	Allele-Specific Chromosome Removal after Cas9 Cleavage in Human Embryos. <i>Cell</i> , <b>2020</b> , 183, 1650-166	45 <b>6</b> .125	75
287	Dark noise and retinal degeneration from D190N-rhodopsin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2020</b> , 117, 23033-23043	11.5	2
286	Quantitative Fundus Autofluorescence in HCQ Retinopathy <b>2020</b> , 61, 41		4
285	A mutation in causing pigmented paravenous retinochoroidal atrophy. <i>European Journal of Ophthalmology</i> , <b>2020</b> , 1120672120957599	1.9	
284	Presumed Chloroquine Retinopathy With Short-term Therapy for Glioblastoma Multiforme. <i>JAMA Ophthalmology</i> , <b>2020</b> , 138, 1215-1217	3.9	1
283	Spectral-Domain Optical Coherence Tomography Is More Sensitive for Hydroxychloroquine-Related Structural Abnormalities Than Short-Wavelength and Near-Infrared Autofluorescence. <i>Translational Vision Science and Technology</i> , <b>2020</b> , 9, 8	3.3	3
282	Novel REEP6 gene mutation associated with autosomal recessive retinitis pigmentosa. <i>Documenta Ophthalmologica</i> , <b>2020</b> , 140, 67-75	2.2	4
281	Progressive Choriocapillaris Impairment in ABCA4 Maculopathy Is Secondary to Retinal Pigment Epithelium Atrophy <b>2020</b> , 61, 13		3
280	Perspectives on Gene Therapy: Choroideremia Represents a Challenging Model for the Treatment of Other Inherited Retinal Degenerations. <i>Translational Vision Science and Technology</i> , <b>2020</b> , 9, 17	3.3	6
279	HMGB1 and Caveolin-1 related to RPE cell senescence in age-related macular degeneration. <i>Aging</i> , <b>2019</b> , 11, 4323-4337	5.6	10
278	CLIC4 regulates late endosomal trafficking and matrix degradation activity of MMP14 at focal adhesions in RPE cells. <i>Scientific Reports</i> , <b>2019</b> , 9, 12247	4.9	9
277	Hypoxic drive caused type 3 neovascularization in a preclinical model of exudative age-related macular degeneration. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 3475-3485	5.6	5
276	Distinct Imprinting Signatures and Biased Differentiation of Human Androgenetic and Parthenogenetic Embryonic Stem Cells. <i>Cell Stem Cell</i> , <b>2019</b> , 25, 419-432.e9	18	14
275	Modification of the disease phenotype by a mutation in. <i>Ophthalmic Genetics</i> , <b>2019</b> , 40, 369-375	1.2	9
274	Fundus autofluorescence and ellipsoid zone (EZ) line width can be an outcome measurement in RHO-associated autosomal dominant retinitis pigmentosa. <i>Graefels Archive for Clinical and Experimental Ophthalmology</i> , <b>2019</b> , 257, 725-731	3.8	13
273	VCAN Canonical Splice Site Mutation is Associated With Vitreoretinal Degeneration and Disrupts an MMP Proteolytic Site <b>2019</b> , 60, 282-293		7
272	Proteomic insight into the pathogenesis of CAPN5-vitreoretinopathy. <i>Scientific Reports</i> , <b>2019</b> , 9, 7608	4.9	5

271	Spectrum of Disease Severity and Phenotype in Choroideremia Carriers. <i>American Journal of Ophthalmology</i> , <b>2019</b> , 207, 77-86	4.9	11
270	Multimodal Imaging in Best Vitelliform Macular Dystrophy <b>2019</b> , 60, 2012-2022		14
269	Correlation between B-scan optical coherence tomography, en face thickness map ring and hyperautofluorescent ring in retinitis pigmentosa patients. <i>Graefeks Archive for Clinical and Experimental Ophthalmology</i> , <b>2019</b> , 257, 1601-1609	3.8	О
268	Multi-platform imaging in ABCA4-Associated Disease. <i>Scientific Reports</i> , <b>2019</b> , 9, 6436	4.9	12
267	Therapeutic Window for Phosphodiesterase 6-Related Retinitis Pigmentosa. <i>JAMA Ophthalmology</i> , <b>2019</b> , 137, 679-680	3.9	3
266	Mechanisms of neurodegeneration in a preclinical autosomal dominant retinitis pigmentosa knock-in model with a Rho mutation. <i>Cellular and Molecular Life Sciences</i> , <b>2019</b> , 76, 3657-3665	10.3	5
265	A case-control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic ABCA4 variants. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2336-2344	8.1	22
264	Non-paraneoplastic related retinopathy: clinical challenges and review. <i>Ophthalmic Genetics</i> , <b>2019</b> , 40, 293-297	1.2	1
263	Choroidal neovascularization in an adolescent with -associated retinal degeneration. <i>Ophthalmic Genetics</i> , <b>2019</b> , 40, 362-364	1.2	3
262	CAPN5 genetic inactivation phenotype supports therapeutic inhibition trials. <i>Human Mutation</i> , <b>2019</b> , 40, 2377-2392	4.7	5
261	Significant Vision Recovery after Early Treatment of Diffuse Unilateral Subacute Neuroretinitis. <i>Ophthalmology Retina</i> , <b>2019</b> , 3, 709	3.8	
260	Comparison of structural progression between ciliopathy and non-ciliopathy associated with autosomal recessive retinitis pigmentosa. <i>Orphanet Journal of Rare Diseases</i> , <b>2019</b> , 14, 187	4.2	8
259	Multimodal structural disease progression of retinitis pigmentosa according to mode of inheritance. <i>Scientific Reports</i> , <b>2019</b> , 9, 10712	4.9	13
258	Characterization of Retinal Structure in ATF6-Associated Achromatopsia <b>2019</b> , 60, 2631-2640		27
257	CRISPR Base Editing in Induced Pluripotent Stem Cells. <i>Methods in Molecular Biology</i> , <b>2019</b> , 2045, 337-3	3 <b>46</b> 4	7
256	Hyperautofluorescent Dots are Characteristic in Ceramide Kinase Like-associated Retinal Degeneration. <i>Scientific Reports</i> , <b>2019</b> , 9, 876	4.9	5
255	Investigation and Restoration of BEST1 Activity in Patient-derived RPEs with Dominant Mutations. <i>Scientific Reports</i> , <b>2019</b> , 9, 19026	4.9	15
254	Compound heterozygous novel frameshift variants in the gene result in Leber congenital amaurosis. <i>Journal of Physical Education and Sports Management</i> , <b>2019</b> , 5,	2.8	4

#### (2018-2019)

253	Novel mutations in the 3-box motif of the BACK domain of KLHL7 associated with nonsyndromic autosomal dominant retinitis pigmentosa. <i>Orphanet Journal of Rare Diseases</i> , <b>2019</b> , 14, 295	4.2	2
252	VITAMIN A DEFICIENCY MONITORED BY QUANTITATIVE SHORT WAVELENGTH FUNDUS AUTOFLUORESCENCE IN A CASE OF BARIATRIC SURGERY. <i>Retinal Cases and Brief Reports</i> , <b>2019</b> ,	1.1	2
251	SCAPER-associated nonsyndromic autosomal recessive retinitis pigmentosa. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 312-316	2.5	6
250	Attenuation of Inherited and Acquired Retinal Degeneration Progression with Gene-based Techniques. <i>Molecular Diagnosis and Therapy</i> , <b>2019</b> , 23, 113-120	4.5	4
249	Viral Delivery Systems for CRISPR. <i>Viruses</i> , <b>2019</b> , 11,	6.2	92
248	Adeno-Associated Viral Gene Therapy for Inherited Retinal Disease. <i>Pharmaceutical Research</i> , <b>2019</b> , 36, 34	4.5	27
247	Macular spatial distribution of preserved autofluorescence in patients with choroideremia. <i>British Journal of Ophthalmology</i> , <b>2019</b> , 103, 933-937	5.5	11
246	Phenotypic expansion and progression of SPATA7-associated retinitis pigmentosa. <i>Documenta Ophthalmologica</i> , <b>2018</b> , 136, 125-133	2.2	2
245	A novel de novo mutation in a patient with inflammatory vitreoretinopathy, hearing loss, and developmental delay. <i>Journal of Physical Education and Sports Management</i> , <b>2018</b> , 4,	2.8	16
244	The unfolded protein response regulator ATF6 promotes mesodermal differentiation. <i>Science Signaling</i> , <b>2018</b> , 11,	8.8	34
243	Autologous stem cell therapy for inherited and acquired retinal disease. <i>Regenerative Medicine</i> , <b>2018</b> , 13, 89-96	2.5	9
242	Personalized Proteomics in Proliferative Vitreoretinopathy Implicate Hematopoietic Cell Recruitment and mTOR as a Therapeutic Target. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 186, 152-163	3 <sup>4.9</sup>	22
241	Reprogramming the metabolome rescues retinal degeneration. <i>Cellular and Molecular Life Sciences</i> , <b>2018</b> , 75, 1559-1566	10.3	11
240	Success of Gene Therapy in Late-Stage Treatment. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1074, 101-107	3.6	2
239	Treatment of Macular Degeneration with Sildenafil: Results of a Two-Year Trial. <i>Ophthalmologica</i> , <b>2018</b> , 240, 45-54	3.7	12
238	A Distinct Phenotype of Eyes Shut Homolog (EYS)-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 190, 99-112	4.9	14
237	CHOROIDEREMIA ASSOCIATED WITH A NOVEL SYNONYMOUS MUTATION IN GENE ENCODING REP-1. <i>Retinal Cases and Brief Reports</i> , <b>2018</b> , 12 Suppl 1, S67-S71	1.1	6
236	HYPERREFLECTIVE DEPOSITION IN THE BACKGROUND OF ADVANCED STARGARDT DISEASE. <i>Retina</i> , <b>2018</b> , 38, 2214-2219	3.6	5

235	The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. <i>Ophthalmology</i> , <b>2018</b> , 125, 89-99	7.3	24
234	CRISPR/Cas9 genome surgery for retinal diseases. <i>Drug Discovery Today: Technologies</i> , <b>2018</b> , 28, 23-32	7.1	7
233	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 195, 16-25	4.9	8
232	Gene therapy in inherited retinal degenerative diseases, a review. <i>Ophthalmic Genetics</i> , <b>2018</b> , 39, 560-5	68.2	42
231	Genetic Rescue Reverses Microglial Activation in Preclinical Models of Retinitis Pigmentosa. <i>Molecular Therapy</i> , <b>2018</b> , 26, 1953-1964	11.7	10
230	Translation of CRISPR Genome Surgery to the Bedside for Retinal Diseases. <i>Frontiers in Cell and Developmental Biology</i> , <b>2018</b> , 6, 46	5.7	15
229	CRISPR GENOME SURGERY IN THE RETINA IN LIGHT OF OFF-TARGETING. Retina, 2018, 38, 1443-1455	3.6	9
228	Clustered Regularly Interspaced Short Palindromic Repeats-Based Genome Surgery for the Treatment of Autosomal Dominant Retinitis Pigmentosa. <i>Ophthalmology</i> , <b>2018</b> , 125, 1421-1430	7.3	65
227	HTRA1, an age-related macular degeneration protease, processes extracellular matrix proteins EFEMP1 and TSP1. <i>Aging Cell</i> , <b>2018</b> , 17, e12710	9.9	38
226	Mutations in GPR143/OA1 and ABCA4 Inform Interpretations of Short-Wavelength and Near-Infrared Fundus Autofluorescence <b>2018</b> , 59, 2459-2469		21
225	Quantitative Comparison of Near-infrared Versus Short-wave Autofluorescence Imaging in Monitoring Progression of Retinitis Pigmentosa. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 194, 120-12	5 <sup>4.9</sup>	13
224	Rates of Bone Spicule Pigment Appearance in Patients With Retinitis Pigmentosa Sine Pigmento. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 195, 176-180	4.9	9
223	Proteomic analysis of the human retina reveals region-specific susceptibilities to metabolic- and oxidative stress-related diseases. <i>PLoS ONE</i> , <b>2018</b> , 13, e0193250	3.7	24
222	Gene therapy and genome surgery in the retina. <i>Journal of Clinical Investigation</i> , <b>2018</b> , 128, 2177-2188	15.9	76
221	Patients and animal models of CNGI-deficient retinitis pigmentosa support gene augmentation approach. <i>Journal of Clinical Investigation</i> , <b>2018</b> , 128, 190-206	15.9	30
220	Blue Cone Monochromatism. Advances in Experimental Medicine and Biology, 2018, 1085, 65-66	3.6	1
219	Late-Onset Retinal Degeneration. Advances in Experimental Medicine and Biology, 2018, 1085, 115-116	3.6	
218	Retinal Histology and Anatomical Landmarks. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 3-5	3.6	1

#### (2018-2018)

217	Pigmented Paravenous Chorioretinal Atrophy (PPCRA). <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 111-113	3.6	1
216	Rubella Retinopathy. Advances in Experimental Medicine and Biology, 2018, 1085, 215-217	3.6	
215	Diffuse Unilateral Subacute Neuroretinitis (DUSN). <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 239-241	3.6	
214	Inborn Errors of Metabolism: Pseudoxanthoma Elasticum. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 187-189	3.6	1
213	Acute Zonal Occult Outer Retinopathy (AZOOR) and Related Diseases. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 233-237	3.6	1
212	Doyne Honeycomb Retinal Dystrophy (Malattia Leventinese, Autosomal Dominant Drusen). <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 97-102	3.6	2
211	X-linked Juvenile Retinoschisis. Advances in Experimental Medicine and Biology, 2018, 1085, 43-48	3.6	4
<b>21</b> 0	Progressive Cone Dystrophy and Cone-Rod Dystrophy (XL, AD, and AR). <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 53-60	3.6	10
209	Congenital Stationary Night Blindness. Advances in Experimental Medicine and Biology, 2018, 1085, 61-6	43.6	5
208	Pattern Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 91-96	3.6	5
207	North Carolina Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 109-110	3.6	O
206	Enhanced S-Cone Syndrome (Goldmann-Favre Syndrome). <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 153-156	3.6	5
205	Best Vitelliform Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 157-158	3.6	6
204	Mitochondrial Disorder: Kearns-Sayre Syndrome. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 161-162	3.6	12
203	Ciliopathy: Senior-Lilen Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 175-178	3.6	10
202	Von Hippel-Lindau Disease. Advances in Experimental Medicine and Biology, 2018, 1085, 201-203	3.6	3
201	Electroretinography. Advances in Experimental Medicine and Biology, 2018, 1085, 17-20	3.6	8
200	A Practical Approach to Retinal Dystrophies. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 245-259	3.6	3

199	Electrooculography. Advances in Experimental Medicine and Biology, 2018, 1085, 21-22	3.6	1
198	CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa: A Brief Methodology. <i>Methods in Molecular Biology</i> , <b>2018</b> , 1715, 191-205	1.4	3
197	Stem cell therapy and regenerative medicine in RPE degenerative disease: advances and challenges. <i>Expert Review of Ophthalmology</i> , <b>2018</b> , 13, 321-327	1.5	
196	Caring for Hereditary Childhood Retinal Blindness. <i>Asia-Pacific Journal of Ophthalmology</i> , <b>2018</b> , 7, 183-1	<b>9:1</b> 5	11
195	Personalized Proteomics for Precision Health: Identifying Biomarkers of Vitreoretinal Disease. Translational Vision Science and Technology, <b>2018</b> , 7, 12	3.3	21
194	Extracellular Matrix: Alport Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 197-	1986	O
193	Glossary of Relevant Genetic and Molecular/Cell Biology. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 23-28	3.6	
192	Autosomal Dominant Retinitis Pigmentosa. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 69-77	3.6	15
191	Best Vitelliform Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 79-90	3.6	6
190	Occult Macular Dystrophy. Advances in Experimental Medicine and Biology, 2018, 1085, 103-104	3.6	2
189	Sorsby Pseudoinflammatory Fundus Dystrophy. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 105-108	3.6	1
188	Retinitis Pigmentosa (Non-syndromic). Advances in Experimental Medicine and Biology, 2018, 1085, 125-	13.66	24
187	Stargardt Disease. Advances in Experimental Medicine and Biology, 2018, 1085, 139-151	3.6	19
186	Optical Coherence Tomography. Advances in Experimental Medicine and Biology, 2018, 1085, 11-13	3.6	2
185	Mitochondrial Disorder: Maternally Inherited Diabetes and Deafness. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 163-165	3.6	7
184	Ciliopathy: AlstrEn Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 179-180	3.6	8
183	Ciliopathy: Sjigren-Larsson Syndrome. Advances in Experimental Medicine and Biology, <b>2018</b> , 1085, 181-18	3 <b>3</b> .6	1
182	Inborn Errors of Metabolism: Gyrate Atrophy. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 183-185	3.6	6

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181	Inborn Errors of Metabolism: Refsum Disease. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 191-192	3.6	2
180	Inborn Errors of Metabolism: Bietti Crystalline Dystrophy. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 193-195	3.6	1
179	Neurofibromatosis. Advances in Experimental Medicine and Biology, 2018, 1085, 209-211	3.6	
178	Syphilis. Advances in Experimental Medicine and Biology, 2018, 1085, 219-221	3.6	2
177	Drug-Induced Retinal Toxicity. Advances in Experimental Medicine and Biology, 2018, 1085, 227-232	3.6	4
176	Genetic Testing for Inherited Retinal Dystrophy: Basic Understanding. <i>Advances in Experimental Medicine and Biology</i> , <b>2018</b> , 1085, 261-268	3.6	2
175	X-linked Choroideremia. Advances in Experimental Medicine and Biology, 2018, 1085, 37-42	3.6	2
174	Rod Monochromatism (Achromatopsia). Advances in Experimental Medicine and Biology, <b>2018</b> , 1085, 119	)- <u>1</u> .83	7
173	Fundus Autofluorescence. Advances in Experimental Medicine and Biology, 2018, 1085, 15-16	3.6	4
172	Tuberous Sclerosis. Advances in Experimental Medicine and Biology, <b>2018</b> , 1085, 205-207	3.6	4
171	X-linked Ocular Albinism. Advances in Experimental Medicine and Biology, 2018, 1085, 49-52	3.6	2
170	Fluorescein Angiography. Advances in Experimental Medicine and Biology, 2018, 1085, 7-10	3.6	3
169	Leber Congenital Amaurosis. Advances in Experimental Medicine and Biology, 2018, 1085, 131-137	3.6	23
168	Ciliopathy: Usher Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 167-170	3.6	17
167	Ciliopathy: Bardet-Biedl Syndrome. Advances in Experimental Medicine and Biology, 2018, 1085, 171-174	3.6	28
166	Autoimmune Retinopathy. Advances in Experimental Medicine and Biology, 2018, 1085, 223-226	3.6	2
165	X-linked Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2018, 1085, 31-35	3.6	9
164	Revolution in Gene Medicine Therapy and Genome Surgery. <i>Genes</i> , <b>2018</b> , 9,	4.2	17

163	Quantitative progression of retinitis pigmentosa by optical coherence tomography angiography. <i>Scientific Reports</i> , <b>2018</b> , 8, 13130	4.9	25
162	Structural disease progression in PDE6-associated autosomal recessive retinitis pigmentosa. <i>Ophthalmic Genetics</i> , <b>2018</b> , 39, 610-614	1.2	13
161	Missense mutation in SLIT2 associated with congenital myopia, anisometropia, connective tissue abnormalities, and obesity. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 138	4.2	3
160	Deferoxamine-induced electronegative ERG responses. <i>Documenta Ophthalmologica</i> , <b>2018</b> , 137, 15-23	2.2	2
159	Congenital grouped albinotic spots of the retinal pigment epithelium in a patient with hemihypertrophy and caffau lait spots. <i>Documenta Ophthalmologica</i> , <b>2018</b> , 137, 9-14	2.2	1
158	Multimodal characterization of a novel mutation causing vitamin B6-responsive gyrate atrophy. <i>Ophthalmic Genetics</i> , <b>2018</b> , 39, 512-516	1.2	7
157	Extremely hypomorphic and severe deep intronic variants in the locus result in varying Stargardt disease phenotypes. <i>Journal of Physical Education and Sports Management</i> , <b>2018</b> , 4,	2.8	45
156	Two-year progression analysis of RPE65 autosomal dominant retinitis pigmentosa. <i>Ophthalmic Genetics</i> , <b>2018</b> , 39, 544-549	1.2	16
155	Extracellular superoxide dismutase (SOD3) regulates oxidative stress at the vitreoretinal interface. <i>Free Radical Biology and Medicine</i> , <b>2018</b> , 124, 408-419	7.8	24
154	Multimodal analysis of the Preferred Retinal Location and the Transition Zone in patients with Stargardt Disease. <i>Graefeks Archive for Clinical and Experimental Ophthalmology</i> , <b>2017</b> , 255, 1307-1317	3.8	11
153	Measurement and Reproducibility of Preserved Ellipsoid Zone Area and Preserved Retinal Pigment Epithelium Area in Eyes With Choroideremia. <i>American Journal of Ophthalmology</i> , <b>2017</b> , 179, 110-117	4.9	43
152	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> , <b>2017</b> , 114, 5259-5264	11.5	17
151	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> , <b>2017</b> , 54, 404-412	5.8	97
150	Peripapillary sparing in RDH12-associated Leber congenital amaurosis. <i>Ophthalmic Genetics</i> , <b>2017</b> , 38, 575-579	1.2	15
149	Disease in a Dish Modeling of Retinal Diseases 2017, 107-115		
148	Unexpected mutations after CRISPR-Cas9 editing in vivo. <i>Nature Methods</i> , <b>2017</b> , 14, 547-548	21.6	233
147	Evaluating Structural Progression of Retinitis Pigmentosa After Cataract Surgery. <i>American Journal of Ophthalmology</i> , <b>2017</b> , 180, 117-123	4.9	7
146	Proteomic Analysis of Elevated Intraocular Pressure with Retinal Detachment. <i>American Journal of Ophthalmology Case Reports</i> , <b>2017</b> , 5, 107-110	1.3	8

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145	Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 735-743	5.3	26	
144	CRISPR applications in ophthalmologic genome surgery. <i>Current Opinion in Ophthalmology</i> , <b>2017</b> , 28, 252-259	5.1	20	
143	Two pathways of rod photoreceptor cell death induced by elevated cGMP. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 2299-2306	5.6	33	
142	Structural modeling of a novel mutation that causes foveal hypoplasia. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2017</b> , 5, 202-209	2.3	18	
141	Achromatopsia mutations target sequential steps of ATF6 activation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, 400-405	11.5	34	
140	Quantitative Autofluorescence Intensities in Acute Zonal Occult Outer Retinopathy vs Healthy Eyes. <i>JAMA Ophthalmology</i> , <b>2017</b> , 135, 1330-1338	3.9	19	
139	CRISPR-mediated Ophthalmic Genome Surgery. Current Ophthalmology Reports, 2017, 5, 199-206	1.8	10	
138	Patient-specific mutations impair BESTROPHIN1's essential role in mediating Ca-dependent Cl currents in human RPE. <i>ELife</i> , <b>2017</b> , 6,	8.9	30	
137	Efficacy of rituximab in non-paraneoplastic autoimmune retinopathy. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 129	4.2	11	
136	Calpain-5 gene expression in the mouse eye and brain. <i>BMC Research Notes</i> , <b>2017</b> , 10, 602	2.3	3	
135	A Comparison of En Face Optical Coherence Tomography and Fundus Autofluorescence in Stargardt Disease <b>2017</b> , 58, 5227-5236		20	
134	CRISPR-Cas Genome Surgery in Ophthalmology. <i>Translational Vision Science and Technology</i> , <b>2017</b> , 6, 13	3.3	12	
133	Quantifying Fundus Autofluorescence in Patients With Retinitis Pigmentosa <b>2017</b> , 58, 1843-1855		48	
132	Electroretinography Reveals Difference in Cone Function between Syndromic and Nonsyndromic USH2A Patients. <i>Scientific Reports</i> , <b>2017</b> , 7, 11170	4.9	17	
131	Retrospective Analysis of Structural Disease Progression in Retinitis Pigmentosa Utilizing Multimodal Imaging. <i>Scientific Reports</i> , <b>2017</b> , 7, 10347	4.9	39	
130	Gene Therapy Restores Mfrp and Corrects Axial Eye Length. Scientific Reports, 2017, 7, 16151	4.9	28	
129	Viral Vectors, Engineered Cells and the CRISPR Revolution. <i>Advances in Experimental Medicine and Biology</i> , <b>2017</b> , 1016, 3-27	3.6	11	
128	CRISPR in the Retina: Evaluation of Future Potential. <i>Advances in Experimental Medicine and Biology</i> , <b>2017</b> , 1016, 147-155	3.6	3	

127	Dissection of Human Retina and RPE-Choroid for Proteomic Analysis. <i>Journal of Visualized Experiments</i> , <b>2017</b> ,	1.6	5
126	PHENOTYPING CHOROIDEREMIA AND ITS CARRIER STATE WITH MULTIMODAL IMAGING TECHNIQUES. <i>Retinal Cases and Brief Reports</i> , <b>2017</b> , 11 Suppl 1, S178-S181	1.1	10
125	Correction of Monogenic and Common Retinal Disorders with Gene Therapy. <i>Genes</i> , <b>2017</b> , 8,	4.2	29
124	Stem Cell Therapies in Retinal Disorders. <i>Cells</i> , <b>2017</b> , 6,	7.9	28
123	Therapeutic drug repositioning using personalized proteomics of liquid biopsies. <i>JCI Insight</i> , <b>2017</b> , 2,	9.9	19
122	Genome Surgery and Gene Therapy in Retinal Disorders. <i>Yale Journal of Biology and Medicine</i> , <b>2017</b> , 90, 523-532	2.4	8
121	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. <i>BMJ Case Reports</i> , <b>2017</b> , 2017,	0.9	2
120	Skin Biopsy and Patient-Specific Stem Cell Lines. <i>Methods in Molecular Biology</i> , <b>2016</b> , 1353, 77-88	1.4	10
119	Genome Editing in the Retina: A Case Study in CRISPR for a Patient-Specific Autosomal Dominant Retinitis Pigmentosa Model <b>2016</b> , 149-162		
118	Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4201-4210	5.6	7
117	Reprogramming towards anabolism impedes degeneration in a preclinical model of retinitis pigmentosa. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4244-4255	5.6	18
116	Gene and cell-based therapies for inherited retinal disorders: An update. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics,</i> <b>2016</b> , 172, 349-366	3.1	50
115	Neuroretinal hypoxic signaling in a new preclinical murine model for proliferative diabetic retinopathy. <i>Signal Transduction and Targeted Therapy</i> , <b>2016</b> , 1,	21	20
114	Precision Medicine: Genetic Repair of Retinitis Pigmentosa in Patient-Derived Stem Cells. <i>Scientific Reports</i> , <b>2016</b> , 6, 19969	4.9	112
113	MULTIMODAL IMAGING OF DISEASE-ASSOCIATED PIGMENTARY CHANGES IN RETINITIS PIGMENTOSA. <i>Retina</i> , <b>2016</b> , 36 Suppl 1, S147-S158	3.6	20
112	Precision Medicine: Personalized Proteomics for the Diagnosis and Treatment of Idiopathic Inflammatory Disease. <i>JAMA Ophthalmology</i> , <b>2016</b> , 134, 444-8	3.9	44
111	Phototransduction Influences Metabolic Flux and Nucleotide Metabolism in Mouse Retina. <i>Journal of Biological Chemistry</i> , <b>2016</b> , 291, 4698-710	5.4	58
110	Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. <i>Human Genetics</i> , <b>2016</b> , 135, 9-19	6.3	33

109	Photopsia and a temporal visual field defect. Survey of Ophthalmology, 2016, 61, 363-7	6.1	2
108	Personalized Medicine: Cell and Gene Therapy Based on Patient-Specific iPSC-Derived Retinal Pigment Epithelium Cells. <i>Advances in Experimental Medicine and Biology</i> , <b>2016</b> , 854, 549-55	3.6	22
107	Reprogramming metabolism by targeting sirtuin 6 attenuates retinal degeneration. <i>Journal of Clinical Investigation</i> , <b>2016</b> , 126, 4659-4673	15.9	52
106	Calpain-5 Expression in the Retina Localizes to Photoreceptor Synapses <b>2016</b> , 57, 2509-21		20
105	Quantitative Autofluorescence and ABCA4 Disease <b>2016</b> , 57, 3297-8		
104	Simultaneous Expression of ABCA4 and GPR143 Mutations: A Complex Phenotypic Manifestation <b>2016</b> , 57, 3409-15		6
103	Secondary glaucoma in CAPN5-associated neovascular inflammatory vitreoretinopathy. <i>Clinical Ophthalmology</i> , <b>2016</b> , 10, 1187-97	2.5	7
102	Complication of Autologous Stem Cell Transplantation in Retinitis Pigmentosa. <i>JAMA Ophthalmology</i> , <b>2016</b> , 134, 711-2	3.9	12
101	CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa. <i>Molecular Therapy</i> , <b>2016</b> , 24, 1388-94	11.7	74
100	BESTROPHIN1 mutations cause defective chloride conductance in patient stem cell-derived RPE. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2672-2680	5.6	33
99	Laser-Induced Photic Injury Phenocopies Macular Dystrophy. <i>Ophthalmic Genetics</i> , <b>2016</b> , 37, 59-67	1.2	22
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97	CAPN5 mutation in hereditary uveitis: the R243L mutation increases calpain catalytic activity and triggers intraocular inflammation in a mouse model. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4584-98	5.6	30
96	Rapid resolution of retinoschisis with acetazolamide. <i>Documenta Ophthalmologica</i> , <b>2015</b> , 131, 63-70	2.2	11
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93	Bilateral Concordance of the Fundus Hyperautofluorescent Ring in Typical Retinitis Pigmentosa Patients. <i>Ophthalmic Genetics</i> , <b>2015</b> , 36, 113-22	1.2	19
92	Rod metabolic demand drives progression in retinopathies. <i>Taiwan Journal of Ophthalmology</i> , <b>2015</b> , 5, 105-108	1.4	11

91	Quantitative fundus autofluorescence distinguishes ABCA4-associated and non-ABCA4-associated bull's-eye maculopathy. <i>Ophthalmology</i> , <b>2015</b> , 122, 345-55	7.3	60
90	Quantitative Fundus Autofluorescence and Optical Coherence Tomography in ABCA4 Carriers <b>2015</b> , 56, 7274-85		23
89	Quantitative Fundus Autofluorescence and Optical Coherence Tomography in PRPH2/RDS- and ABCA4-Associated Disease Exhibiting Phenotypic Overlap <b>2015</b> , 56, 3159-70		43
88	Patient-Specific iPSC-Derived RPE for Modeling of Retinal Diseases. <i>Journal of Clinical Medicine</i> , <b>2015</b> , 4, 567-78	5.1	22
87	Structural modeling of a novel CAPN5 mutation that causes uveitis and neovascular retinal detachment. <i>PLoS ONE</i> , <b>2015</b> , 10, e0122352	3.7	29
86	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. <i>Nature Genetics</i> , <b>2015</b> , 47, 757-65	36.3	143
85	Quantitative autofluorescence as a clinical tool for expedited differential diagnosis of retinal degeneration. <i>JAMA Ophthalmology</i> , <b>2015</b> , 133, 219-20	3.9	6
84	Personalized therapeutic strategies for patients with retinitis pigmentosa. <i>Expert Opinion on Biological Therapy</i> , <b>2015</b> , 15, 391-402	5.4	38
83	Differentiation of hypothalamic-like neurons from human pluripotent stem cells. <i>Journal of Clinical Investigation</i> , <b>2015</b> , 125, 796-808	15.9	84
82	Halting progressive neurodegeneration in advanced retinitis pigmentosa. <i>Journal of Clinical Investigation</i> , <b>2015</b> , 125, 3704-13	15.9	49
81	Emerging Treatments for Retinitis Pigmentosa: Genes and stem cells, as well as new electronic and medical therapies, are gaining ground <b>2015</b> , 12, 52-70		17
80	Validation of genome-wide association study (GWAS)-identified disease risk alleles with patient-specific stem cell lines. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 3445-55	5.6	74
79	The role of fundus autofluorescence in late-onset retinitis pigmentosa (LORP) diagnosis. <i>Ophthalmic Genetics</i> , <b>2014</b> , 35, 170-9	1.2	7
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77	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> , <b>2014</b> , 55, 8134-43		62
76	A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. <i>Cell</i> , <b>2014</b> , 159, 200-214	56.2	239
75	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 5774-80	5.6	27
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72	Quantitative fundus autofluorescence and optical coherence tomography in best vitelliform macular dystrophy <b>2014</b> , 55, 1471-82		74
71	Quantitative fundus autofluorescence in recessive Stargardt disease <b>2014</b> , 55, 2841-52		127
70	Retinal damage in chloroquine maculopathy, revealed by high resolution imaging: a case report utilizing adaptive optics scanning laser ophthalmoscopy. <i>Korean Journal of Ophthalmology: KJO</i> , <b>2014</b> , 28, 100-7	1.2	10
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64	General pathophysiology in retinal degeneration. <i>Developments in Ophthalmology</i> , <b>2014</b> , 53, 33-43		44
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59	Disease progression in autosomal dominant cone-rod dystrophy caused by a novel mutation (D100G) in the GUCA1A gene. <i>Documenta Ophthalmologica</i> , <b>2014</b> , 128, 59-67	2.2	22
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57	Cone photoreceptor abnormalities correlate with vision loss in a case of acute posterior multifocal placoid pigment epitheliopathy. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , <b>2014</b> , 45, 74-8	1.4	5
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53	Abnormality in the external limiting membrane in early Stargardt disease. <i>Ophthalmic Genetics</i> , <b>2013</b> , 34, 75-7	1.2	22
52	Evaluation of multimodal imaging in carriers of X-linked retinitis pigmentosa. <i>Experimental Eye Research</i> , <b>2013</b> , 113, 41-8	3.7	35
51	Cellular imaging demonstrates genetic mosaicism in heterozygous carriers of an X-linked ciliopathy gene. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 1240-8	5.3	22
50	Gene therapy provides long-term visual function in a pre-clinical model of retinitis pigmentosa.  Human Molecular Genetics, <b>2013</b> , 22, 558-67	5.6	47
49	Comparison of near-infrared and short-wavelength autofluorescence in retinitis pigmentosa <b>2013</b> , 54, 585-91		71
48	Therapeutic margins in a novel preclinical model of retinitis pigmentosa. <i>Journal of Neuroscience</i> , <b>2013</b> , 33, 13475-83	6.6	24
47	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> , <b>2012</b> , 153, 143-54.e2	4.9	26
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44	Long-term safety and efficacy of human-induced pluripotent stem cell (iPS) grafts in a preclinical model of retinitis pigmentosa. <i>Molecular Medicine</i> , <b>2012</b> , 18, 1312-9	6.2	136
43	Retinal phenotypes in patients homozygous for the G1961E mutation in the ABCA4 gene <b>2012</b> , 53, 4458	3-67	61
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41	Disruption in Bruch membrane in patients with Stargardt disease. <i>Ophthalmic Genetics</i> , <b>2012</b> , 33, 49-52	1.2	11
40	Subretinal injection of gene therapy vectors and stem cells in the perinatal mouse eye. <i>Journal of Visualized Experiments</i> , <b>2012</b> ,	1.6	18
39	Unilateral retinitis pigmentosa: a proposal of genetic pathogenic mechanisms. <i>European Journal of Ophthalmology</i> , <b>2012</b> , 22, 654-60	1.9	18
38	Structural and functional changes associated with normal and abnormal fundus autofluorescence in patients with retinitis pigmentosa. <i>Retina</i> , <b>2012</b> , 32, 349-57	3.6	46

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37	Vigabatrin-induced retinal toxicity is partially mediated by signaling in rod and cone photoreceptors. <i>PLoS ONE</i> , <b>2012</b> , 7, e43889	3.7	12
36	Familial discordance in Stargardt disease. <i>Molecular Vision</i> , <b>2012</b> , 18, 227-33	2.3	12
35	Functional Analysis of Retinal Flecks in Stargardt Disease. <i>Journal of Clinical &amp; Experimental Ophthalmology</i> , <b>2012</b> , 3,	Ο	14
34	Quantification of peripapillary sparing and macular involvement in Stargardt disease (STGD1) <b>2011</b> , 52, 8006-15		39
33	Mouse eye enucleation for remote high-throughput phenotyping. <i>Journal of Visualized Experiments</i> , <b>2011</b> ,	1.6	16
32	shRNA knockdown of guanylate cyclase 2e or 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> , <b>2011</b> , 15, 1778-87	5.6	22
31	Function of the asparagine 74 residue of the inhibitory Eubunit of retinal rod cGMP-phophodiesterase (PDE) in vivo. <i>Cellular Signalling</i> , <b>2011</b> , 23, 1584-9	4.9	5
30	Allelic and phenotypic heterogeneity in ABCA4 mutations. <i>Ophthalmic Genetics</i> , <b>2011</b> , 32, 165-74	1.2	73
29	Lentivirus-mediated expression of cDNA and shRNA slows degeneration in retinitis pigmentosa. <i>Experimental Biology and Medicine</i> , <b>2011</b> , 236, 1211-7	3.7	23
28	Loss of peripapillary sparing in non-group I Stargardt disease. Experimental Eye Research, 2010, 91, 592-	69 <del>9</del>	23
27	Transplantation of reprogrammed embryonic stem cells improves visual function in a mouse model for retinitis pigmentosa. <i>Transplantation</i> , <b>2010</b> , 89, 911-9	1.8	62
26	Fundus autofluorescence and optical coherence tomography of congenital grouped albinotic spots. <i>Retina</i> , <b>2010</b> , 30, 1217-22	3.6	6
25	Rapid and noninvasive imaging of retinal ganglion cells in live mouse models of glaucoma. <i>Molecular Imaging and Biology</i> , <b>2010</b> , 12, 386-93	3.8	6
24	Transplantation of reprogrammed embryonic stem cells improves visual function in a mouse model for Retinitis Pigmentosa: Transplantation 2010 April 27;89 (8): 911-919. <i>Annals of Neurosciences</i> , <b>2010</b> , 17, 185-6	1.1	
23	A comparison of fundus autofluorescence and retinal structure in patients with Stargardt disease <b>2009</b> , 50, 3953-9		111
22	Autofluorescence imaging in rubella retinopathy. Ocular Immunology and Inflammation, 2009, 17, 400-2	2.8	8
21	Fundus autofluorescence in cone dystrophy. <i>Documenta Ophthalmologica</i> , <b>2009</b> , 119, 141-4	2.2	21
20	G1961E mutant allele in the Stargardt disease gene ABCA4 causes bull's eye maculopathy. <i>Experimental Eye Research</i> , <b>2009</b> , 89, 16-24	3.7	76

19	Light-dependent phosphorylation of the gamma subunit of cGMP-phophodiesterase (PDE6gamma) at residue threonine 22 in intact photoreceptor neurons. <i>Biochemical and Biophysical Research Communications</i> , <b>2009</b> , 390, 1149-53	3.4	9
18	Cellular and molecular origin of circumpapillary dysgenesis of the pigment epithelium. <i>Ophthalmology</i> , <b>2009</b> , 116, 971-80	7:3	6
17	Case report: autofluorescence imaging and phenotypic variance in a sibling pair with early-onset retinal dystrophy due to defective CRB1 function. <i>Current Eye Research</i> , <b>2009</b> , 34, 395-400	2.9	17
16	Structural assessment of hyperautofluorescent ring in patients with retinitis pigmentosa. <i>Retina</i> , <b>2009</b> , 29, 1025-31	3.6	89
15	A novel mutation and phenotypes in phosphodiesterase 6 deficiency. <i>American Journal of Ophthalmology</i> , <b>2008</b> , 146, 780-8	4.9	45
14	Phenotype-genotype correlations in autosomal dominant retinitis pigmentosa caused by RHO, D190N. <i>Current Eye Research</i> , <b>2008</b> , 33, 1014-22	2.9	18
13	Modulation of phosphodiesterase6 turnoff during background illumination in mouse rod photoreceptors. <i>Journal of Neuroscience</i> , <b>2008</b> , 28, 2064-74	6.6	54
12	Preferred retinal locus in macular disease: characteristics and clinical implications. <i>Retina</i> , <b>2008</b> , 28, 123	43.460	52
11	Functional rescue of degenerating photoreceptors in mice homozygous for a hypomorphic cGMP phosphodiesterase 6 b allele (Pde6bH620Q) <b>2008</b> , 49, 5067-76		50
10	Electronegative electroretinogram associated with topiramate toxicity and vitelliform maculopathy. <i>Documenta Ophthalmologica</i> , <b>2008</b> , 116, 57-60	2.2	12
9	Non-vascular vision loss in pseudoxanthoma elasticum. <i>Documenta Ophthalmologica</i> , <b>2008</b> , 117, 65-7	2.2	9
8	Autofluorescence imaging in a case of benign familial fleck retina. JAMA Ophthalmology, 2007, 125, 714	1-5	10
7	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> , <b>2007</b> , 28, 243-54	4.7	33
6	Novel phenotypic and genotypic findings in X-linked retinoschisis. <i>JAMA Ophthalmology</i> , <b>2007</b> , 125, 259	9-67	53
5	Stationary night blindness or progressive retinal degeneration in mice carrying different alleles of PDE gamma. <i>Frontiers in Bioscience - Landmark</i> , <b>2003</b> , 8, s666-75	2.8	9
4	The positive role of the carboxyl terminus of the gamma subunit of retinal cGMP-phosphodiesterase in maintaining phosphodiesterase activity in vivo. <i>Vision Research</i> , <b>2002</b> , 42, 439-45	2.1	13
3	In vivo studies of the gamma subunit of retinal cGMP-phophodiesterase with a substitution of tyrosine-84. <i>Biochemical Journal</i> , <b>2001</b> , 353, 467-74	3.8	10
2	In vivo studies of the Bubunit of retinal cGMP-phophodiesterase with a substitution of tyrosine-84. <i>Biochemical Journal</i> , <b>2001</b> , 353, 467-474	3.8	11

#### LIST OF PUBLICATIONS

Gene Therapy and Surgery for Retinal Diseases1-10