Michel Michaelides

List of Publications by Year in Descending Order

267 papers	8,610 citations	47 h-index	80 g-index
287 ext. papers	10,519 ext. citations	5.1 avg, IF	6.15 L-index

#	Paper	IF	Citations
267	Long-term vision outcomes for patients with albinism and diabetic retinopathy.. *Graefe's Archive for Clinical and Experimental Ophthalmology*, **2022**, 1	3.8	
266	Leber Congenital Amaurosis/Early-Onset Severe Retinal Dystrophy **2022**, 109-115		
265	Progressive and Stationary Disorders of Cone Function: Cone and Cone-Rod Dystrophies and Cone Dysfunction Syndromes **2022**, 3937-3965		
264	RDH12 retinopathy: clinical features, biology, genetics and future directions.. *Ophthalmic Genetics*, **2022**, 1-6	1.2	
263	Pathogenic variants in the gene cause isolated autosomal dominant congenital posterior polar cataracts. *Ophthalmic Genetics*, **2021**, 1-6	1.2	0
262	A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. *Eye*, **2021**, 35, 1482-1489	4.4	2
261	Treatments for dry age-related macular degeneration: therapeutic avenues, clinical trials and future directions. *British Journal of Ophthalmology*, **2021**,	5.5	21
260	Panel-based genetic testing for inherited retinal disease screening 176 genes. *Molecular Genetics & Genomic Medicine*, **2021**, e1663	2.3	0
259	Inherited retinal diseases: Therapeutics, clinical trials and end points-A review. *Clinical and Experimental Ophthalmology*, **2021**, 49, 270-288	2.4	12
258	Leber congenital amaurosis/early-onset severe retinal dystrophy: current management and clinical trials. *British Journal of Ophthalmology*, **2021**,	5.5	7
257	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpoints-KCNV2 Study Group Report 2. *American Journal of Ophthalmology*, **2021**, 230, 1-11	4.9	1
256	Restoration of visual function in advanced disease after transplantation of purified human pluripotent stem cell-derived cone photoreceptors. *Cell Reports*, **2021**, 35, 109022	10.6	18
255	Examining Whether AOSLO-Based Foveal Cone Metrics in Achromatopsia and Albinism Are Representative of Foveal Cone Structure. *Translational Vision Science and Technology*, **2021**, 10, 22	3.3	1
254	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course-KCNV2 Study Group Report 1. *American Journal of Ophthalmology*, **2021**, 225, 95-107	4.9	7
253	Structural evaluation in inherited retinal diseases. *British Journal of Ophthalmology*, **2021**, 105, 1623-1634	5.5	3
252	CNGB1-related rod-cone dystrophy: A mutation review and update. *Human Mutation*, **2021**, 42, 641-666	4.7	6
251	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. *Npj Genomic Medicine*, **2021**, 6, 53	6.2	0

250	Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. *Ophthalmic Genetics*, **2021**, 42, 664-673	1.2	5
249	Swept-source optical coherence tomography changes and visual acuity among Palestinian retinitis Pigmentosa patients: a cross-sectional study. *BMC Ophthalmology*, **2021**, 21, 289	2.3	
248	Gene therapy for neovascular age-related macular degeneration: rationale, clinical trials and future directions. *British Journal of Ophthalmology*, **2021**, 105, 151-157	5.5	21
247	Pigmentary retinopathy can indicate the presence of pathogenic LAMP2 variants even in somatic mosaic carriers with no additional signs of Danon disease. *Acta Ophthalmologica*, **2021**, 99, 61-68	3.7	
246	Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. *Ophthalmology*, **2021**, 128, 952-955	7.3	4
245	Sector Retinitis Pigmentosa: Extending the Molecular Genetics Basis and Elucidating the Natural History. *American Journal of Ophthalmology*, **2021**, 221, 299-310	4.9	7
244	Enhanced S-Cone Syndrome: Spectrum of Clinical, Imaging, Electrophysiologic, and Genetic Findings in a Retrospective Case Series of 56 Patients. *Ophthalmology Retina*, **2021**, 5, 195-214	3.8	6
243	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. *Progress in Retinal and Eye Research*, **2021**, 82, 100898	20.5	17
242	Autosomal Recessive Bestrophinopathy: Clinical Features, Natural History, and Genetic Findings in Preparation for Clinical Trials. *Ophthalmology*, **2021**, 128, 706-718	7.3	11
241	Joubert syndrome diagnosed renally late. *CKJ: Clinical Kidney Journal*, **2021**, 14, 1017-1019	4.5	1
240	A NOVEL CASE SERIES OF NMNAT1-ASSOCIATED EARLY-ONSET RETINAL DYSTROPHY: EXTENDING THE PHENOTYPIC SPECTRUM. *Retinal Cases and Brief Reports*, **2021**, 15, 139-144	1.1	4
239	Retinal Surgical Techniques for Gene Therapy **2021**, 389-395		
238	Optical Coherence Tomography Artifacts Are Associated With Adaptive Optics Scanning Light Ophthalmoscopy Success in Achromatopsia. *Translational Vision Science and Technology*, **2021**, 10, 11	3.3	4
237	Progressive and Stationary Disorders of Cone Function: Cone and Cone-Rod Dystrophies and Cone Dysfunction Syndromes **2021**, 1-29		
236	Gene Therapy in X-linked Retinitis Pigmentosa Due to Defects in RPGR. *International Ophthalmology Clinics*, **2021**, 61, 97-108	1.7	1
235	Extending the phenotypic spectrum of PRPF8, PRPH2, RP1 and RPGR, and the genotypic spectrum of early-onset severe retinal dystrophy. *Orphanet Journal of Rare Diseases*, **2021**, 16, 128	4.2	1
234	Characterization of Retinal Function Using Microperimetry-Derived Metrics in Both Adults and Children With RPGR-Associated Retinopathy. *American Journal of Ophthalmology*, **2021**, 234, 81-90	4.9	0
233	Agreement Between Spectral-Domain and Swept-Source Optical Coherence Tomography Retinal Thickness Measurements in Macular and Retinal Disease. *Ophthalmology and Therapy*, **2021**, 10, 913-922	5	0

232	Comparing Retinal Structure in Patients with Achromatopsia and Blue Cone Monochromacy Using OCT. *Ophthalmology Science*, **2021**, 1, 100047		1
231	The Phenotypic Spectrum of Patients with PHARC Syndrome Due to Variants in : An Ophthalmic Perspective. *Genes*, **2021**, 12,	4.2	2
230	A demonstration of cone function plasticity after gene therapy in achromatopsia. *Journal of Vision*, **2021**, 21, 2444	0.4	1
229	Unilateral congenital non-syndromic retinal vessel dilation and tortuosity. *American Journal of Ophthalmology Case Reports*, **2021**, 23, 101160	1.3	1
228	ATF6 is essential for human cone photoreceptor development. *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	5
227	Photoaversion in inherited retinal diseases: clinical phenotypes, biological basis, and qualitative and quantitative assessment.. *Ophthalmic Genetics*, **2021**, 1-9	1.2	
226	SSBP1-Disease Update: Expanding the Genetic and Clinical Spectrum, Reporting Variable Penetrance and Confirming Recessive Inheritance. **2021**, 62, 12		1
225	The RUSH2A Study: Best-Corrected Visual Acuity, Full-Field Electroretinography Amplitudes, and Full-Field Stimulus Thresholds at Baseline. *Translational Vision Science and Technology*, **2020**, 9, 9	3.3	7
224	Genetic Basis of Inherited Retinal Disease in a Molecularly Characterized Cohort of More Than 3000 Families from the United Kingdom. *Ophthalmology*, **2020**, 127, 1384-1394	7.3	56
223	Whole Exome Sequencing Reveals Novel and Recurrent Disease-Causing Variants in Lens Specific Gap Junctional Protein Encoding Genes Causing Congenital Cataract. *Genes*, **2020**, 11,	4.2	2
222	The Impact of Inherited Retinal Diseases in the Republic of Ireland (ROI) and the United Kingdom (UK) from a Cost-of-Illness Perspective. *Clinical Ophthalmology*, **2020**, 14, 707-719	2.5	22
221	Nystagmus and optical coherence tomography findings in CNGB3-associated achromatopsia. *Journal of AAPOS*, **2020**, 24, 82.e1-82.e7	1.3	2
220	Photoreceptor Structure in GNAT2-Associated Achromatopsia **2020**, 61, 40		19
219	A novel missense mutation in causing isolated autosomal dominant congenital cataract. *Ophthalmic Genetics*, **2020**, 41, 131-134	1.2	2
218	Oliver McFarlane syndrome and choroidal neovascularisation: a case report. *Ophthalmic Genetics*, **2020**, 41, 451-456	1.2	2
217	Quantifying the Separation Between the Retinal Pigment Epithelium and Bruch's Membrane using Optical Coherence Tomography in Patients with Inherited Macular Degeneration. *Translational Vision Science and Technology*, **2020**, 9, 26	3.3	7
216	Longitudinal Assessment of Remnant Foveal Cone Structure in a Case Series of Early Macular Telangiectasia Type 2. *Translational Vision Science and Technology*, **2020**, 9, 27	3.3	4
215	Faster Sensitivity Loss around Dense Scotomas than for Overall Macular Sensitivity in Stargardt Disease: ProgStar Report No. 14. *American Journal of Ophthalmology*, **2020**, 216, 219-225	4.9	12

214	Interocular Symmetry of Foveal Cone Topography in Congenital Achromatopsia. *Current Eye Research*, **2020**, 45, 1257-1264	2.9	19
213	Retinal Structure in RPE65-Associated Retinal Dystrophy **2020**, 61, 47		13
212	Inherited cataracts: molecular genetics, clinical features, disease mechanisms and novel therapeutic approaches. *British Journal of Ophthalmology*, **2020**, 104, 1331-1337	5.5	25
211	Visual hallucinations in neurological and ophthalmological disease: pathophysiology and management. *Journal of Neurology, Neurosurgery and Psychiatry*, **2020**, 91, 512-519	5.5	38
210	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. *Genes*, **2020**, 11,	4.2	7
209	Incidental unilateral idiopathic maculopathy in children. *Journal of AAPOS*, **2020**, 24, 357.e1-357.e6	1.3	
208	Baseline Visual Field Findings in the RUSH2A Study: Associated Factors and Correlation With Other Measures of Disease Severity. *American Journal of Ophthalmology*, **2020**, 219, 87-100	4.9	8
207	retinopathy: clinical features, molecular genetics and directions for future therapy. *Ophthalmic Genetics*, **2020**, 41, 208-215	1.2	15
206	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. *American Journal of Ophthalmology*, **2020**, 210, 59-70	4.9	23
205	Juvenile Batten Disease (CLN3): Detailed Ocular Phenotype, Novel Observations, Delayed Diagnosis, Masquerades, and Prospects for Therapy. *Ophthalmology Retina*, **2020**, 4, 433-445	3.8	17
204	Prospective Cohort Study of Childhood-Onset Stargardt Disease: Fundus Autofluorescence Imaging, Progression, Comparison with Adult-Onset Disease, and Disease Symmetry. *American Journal of Ophthalmology*, **2020**, 211, 159-175	4.9	20
203	Macular dystrophies: clinical and imaging features, molecular genetics and therapeutic options. *British Journal of Ophthalmology*, **2020**, 104, 451-460	5.5	37
202	Clinical and functional analyses of AIPL1 variants reveal mechanisms of pathogenicity linked to different forms of retinal degeneration. *Scientific Reports*, **2020**, 10, 17520	4.9	8
201	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. *American Journal of Human Genetics*, **2020**, 107, 802-814	11	26
200	The genetic landscape of crystallins in congenital cataract. *Orphanet Journal of Rare Diseases*, **2020**, 15, 333	4.2	9
199	Validation of a Vision-Guided Mobility Assessment for -Associated Retinal Dystrophy. *Translational Vision Science and Technology*, **2020**, 9, 5	3.3	6
198	A genetic and clinical study of individuals with nonsyndromic retinopathy consequent upon sequence variants in HGSNAT, the gene associated with Sanfilippo C mucopolysaccharidosis. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 631-643	3.1	5
197	Subfoveal retinal detachment associated with dome-shaped macula in a 6 year-old child: Comparison with other case reports and systematic review of the literature regarding dome-shaped macula in children. *American Journal of Ophthalmology Case Reports*, **2020**, 19, 100821	1.3	1

196	Retinal imaging in inherited retinal diseases. *Annals of Eye Science*, **2020**, 5,	0.9	10
195	Intraobserver Repeatability and Interobserver Reproducibility of Foveal Cone Density Measurements in and -Associated Achromatopsia. *Translational Vision Science and Technology*, **2020** , 9, 37	3.3	6
194	Long-Term Investigation of Retinal Function in Patients with Achromatopsia **2020**, 61, 38		13
193	Preservation of the Foveal Avascular Zone in Achromatopsia Despite the Absence of a Fully Formed Pit **2020**, 61, 52		3
192	Macula-predominant retinopathy associated with biallelic variants in. *Ophthalmic Genetics*, **2020**, 41, 612-615	1.2	4
191	Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. *Eye*, **2020**, 34, 783-784	4.4	2
190	Asymmetric choroidal hypopigmentation in a Son and mother with Waardenburg syndrome type I. *Ophthalmic Genetics*, **2020**, 41, 284-287	1.2	0
189	Prospective exploratory study to assess the safety and efficacy of aflibercept in cystoid macular oedema associated with retinitis pigmentosa. *British Journal of Ophthalmology*, **2020**, 104, 1203-1208	5.5	4
188	Progressive cone and cone-rod dystrophies: clinical features, molecular genetics and prospects for therapy. *British Journal of Ophthalmology*, **2019**,	5.5	74
187	Loss-of-Function Mutations in the CFH Gene Affecting Alternatively Encoded Factor H-like 1 Protein Cause Dominant Early-Onset Macular Drusen. *Ophthalmology*, **2019**, 126, 1410-1421	7.3	17
186	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. *American Journal of Ophthalmology*, **2019**, 207, 87-98	4.9	13
185	Rod-cone dystrophy associated with the Gly167Asp variant in PRPH2. *Ophthalmic Genetics*, **2019**, 40, 188-189	1.2	4
184	Cross-Sectional and Longitudinal Assessment of the Ellipsoid Zone in Childhood-Onset Stargardt Disease. *Translational Vision Science and Technology*, **2019**, 8, 1	3.3	22
183	SUBCLINICAL MACULAR CHANGES AND DISEASE LATERALITY IN PEDIATRIC COATS DISEASE DETERMINED BY QUANTITATIVE OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY. *Retina*, **2019**, 39, 2392-2398	3.6	11
182	Detailed clinical characterisation, unique features and natural history of autosomal recessive -associated retinal degeneration. *British Journal of Ophthalmology*, **2019**, 103, 1789-1796	5.5	15
181	Adaptive Optics Retinal Imaging in CNGA3-Associated Achromatopsia: Retinal Characterization, Interocular Symmetry, and Intrafamilial Variability **2019**, 60, 383-396		32
180	Detailed genetic characteristics of an international large cohort of patients with Stargardt disease: ProgStar study report 8. *British Journal of Ophthalmology*, **2019**, 103, 390-397	5.5	26
179	Scotopic Microperimetric Assessment of Rod Function in Stargardt Disease (SMART) Study: Design and Baseline Characteristics (Report No. 1). *Ophthalmic Research*, **2019**, 61, 36-43	2.9	20

178	Progression of Stargardt Disease as Determined by Fundus Autofluorescence Over a 12-Month Period: ProgStar Report No. 11. *JAMA Ophthalmology*, **2019**, 137, 1134-1145	3.9	35
177	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. *Annals of Neurology*, **2019**, 86, 368-383	9.4	29
176	Characterization of Retinal Structure in ATF6-Associated Achromatopsia **2019**, 60, 2631-2640		27
175	Assessing the Interocular Symmetry of Foveal Outer Nuclear Layer Thickness in Achromatopsia. *Translational Vision Science and Technology*, **2019**, 8, 21	3.3	15
174	Deep Phenotyping of PDE6C-Associated Achromatopsia **2019**, 60, 5112-5123		31
173	Retinopathy Associated with Biallelic Mutations in PYGM (McArdle Disease). *Ophthalmology*, **2019**, 126, 320-322	7.3	10
172	Natural History Study of Retinal Structure, Progression, and Symmetry Using Ellipzoid Zone Metrics in RPGR-Associated Retinopathy. *American Journal of Ophthalmology*, **2019**, 198, 111-123	4.9	29
171	Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. *British Journal of Ophthalmology*, **2019**, 103, 1163-1166	5.5	35
170	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. *Retina*, **2019**, 39, 570-580	3.6	17
169	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY: Detailed Clinical Study of a Large Cohort. *Retina*, **2019**, 39, 514-529	3.6	9
168	Retrospective cohort study exploring whether an association exists between spatial distribution of cystoid spaces in cystoid macular oedema secondary to retinitis pigmentosa and response to treatment with carbonic anhydrase inhibitors. *British Journal of Ophthalmology*, **2019**, 103, 233-237	5.5	12
167	Jalili Syndrome: Cross-sectional and Longitudinal Features of Seven Patients With Cone-Rod Dystrophy and Amelogenesis Imperfecta. *American Journal of Ophthalmology*, **2018**, 188, 123-130	4.9	6
166	Retinal gene therapy. *British Medical Bulletin*, **2018**, 126, 13-25	5.4	35
165	A clinical and molecular characterisation of CRB1-associated maculopathy. *European Journal of Human Genetics*, **2018**, 26, 687-694	5.3	36
164	Gene therapy for Leber congenital amaurosis. *Expert Review of Ophthalmology*, **2018**, 13, 11-15	1.5	3
163	Leber Congenital Amaurosis Associated with Mutations in CEP290, Clinical Phenotype, and Natural History in Preparation for Trials of Novel Therapies. *Ophthalmology*, **2018**, 125, 894-903	7.3	46
162	Early Patterns of Macular Degeneration in ABCA4-Associated Retinopathy. *Ophthalmology*, **2018**, 125, 735-746	7.3	40
161	Achromatopsia: clinical features, molecular genetics, animal models and therapeutic options. *Ophthalmic Genetics*, **2018**, 39, 149-157	1.2	59

160	Resolution of cystoid macular edema following arginine-restricted diet and vitamin B6 supplementation in a case of gyrate atrophy. *Journal of AAPOS*, **2018**, 22, 321-323	1.3	10
159	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. *Retina*, **2018**, 38, 620-628	3.6	9
158	NORMAL ELECTROOCULOGRAPHY IN BEST DISEASE AND AUTOSOMAL RECESSIVE BESTROPHINOPATHY. *Retina*, **2018**, 38, 379-386	3.6	11
157	THE FUNDUS PHENOTYPE ASSOCIATED WITH THE p.Ala243Val BEST1 MUTATION. *Retina*, **2018**, 38, 606-613	3.6	5
156	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. *Ophthalmic Genetics*, **2018**, 39, 236-241	1.2	9
155	Assessment of the incorporation of CNV surveillance into gene panel next-generation sequencing testing for inherited retinal diseases. *Journal of Medical Genetics*, **2018**, 55, 114-121	5.8	38
154	Missense variants in the X-linked gene PRPS1 cause retinal degeneration in females. *Human Mutation*, **2018**, 39, 80-91	4.7	19
153	QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. *Retina*, **2018**, 38, 2401-2414	3.6	26
152	Bullous X linked retinoschisis: clinical features and prognosis. *British Journal of Ophthalmology*, **2018**, 102, 622-624	5.5	11
151	The Progression of the Stargardt Disease Type 4 (ProgStar-4) Study: Design and Baseline Characteristics (ProgStar-4 Report No. 1). *Ophthalmic Research*, **2018**, 60, 185-194	2.9	10
150	Transplantation of Human Embryonic Stem Cell-Derived Retinal Pigment Epithelial Cells in Macular Degeneration. *Ophthalmology*, **2018**, 125, 1765-1775	7.3	101
149	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 3-18	11	27
148	Longitudinal Changes of Fixation Location and Stability Within 12 Months in Stargardt Disease: ProgStar Report No. 12. *American Journal of Ophthalmology*, **2018**, 193, 54-61	4.9	20
147	Visual Acuity Change Over 24 Months and Its Association With Foveal Phenotype and Genotype in Individuals With Stargardt Disease: ProgStar Study Report No. 10. *JAMA Ophthalmology*, **2018**, 136, 920-928	3.9	27
146	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. *Molecular Vision*, **2018**, 24, 603-612	2.3	6
145	Clinical Course of Autosomal Recessive Bestrophinopathy Complicated by Choroidal Neovascularization. *Ophthalmic Surgery Lasers and Imaging Retina*, **2018**, 49, 888-892	1.4	3
144	Adaptive optics imaging of inherited retinal diseases. *British Journal of Ophthalmology*, **2018**, 102, 1028-1035	3.5	43
143	Factors associated with visual acuity in patients with cystoid macular oedema and Retinitis Pigmentosa. *Ophthalmic Epidemiology*, **2018**, 25, 183-186	1.9	5

142	A Cross-Sectional and Longitudinal Study of Retinal Sensitivity in RPE65-Associated Leber Congenital Amaurosis **2018**, 59, 3330-3339		16
141	Cross-Sectional and Longitudinal Assessment of Retinal Sensitivity in Patients With Childhood-Onset Stargardt Disease. *Translational Vision Science and Technology*, **2018**, 7, 10	3.3	18
140	Longitudinal Assessment of Retinal Structure in Achromatopsia Patients With Long-Term Follow-up **2018**, 59, 5735-5744		30
139	Severe Loss of Tritan Color Discrimination in RPE65 Associated Leber Congenital Amaurosis **2018**, 59, 85-93		11
138	Characterization of Visual Function, Interocular Variability and Progression Using Static Perimetry-Derived Metrics in RPGR-Associated Retinopathy **2018**, 59, 2422-2436		20
137	Residual Cone Structure in Patients With X-Linked Cone Opsin Mutations **2018**, 59, 4238-4248		21
136	Fast adaptive optics scanning light ophthalmoscope retinal montaging. *Biomedical Optics Express*, **2018**, 9, 4317-4328	3.5	14
135	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. *PLoS Genetics*, **2018**, 14, e1007504	6	13
134	Automatic Cone Photoreceptor Localisation in Healthy and Stargardt Afflicted Retinas Using Deep Learning. *Scientific Reports*, **2018**, 8, 7911	4.9	31
133	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
132	Whole-genome sequencing reveals a recurrent missense mutation in the Connexin 46 (GJA3) gene causing autosomal-dominant lamellar cataract. *Eye*, **2018**, 32, 1661-1668	4.4	3
131	Novel heterozygous mutation in YAP1 in a family with isolated ocular colobomas. *Ophthalmic Genetics*, **2017**, 38, 281-283	1.2	11
130	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. *JAMA Ophthalmology*, **2017**, 135, 137-144	3.9	17
129	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. *American Journal of Human Genetics*, **2017**, 100, 334-342	11	14
128	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. *American Journal of Human Genetics*, **2017**, 100, 592-604	11	42
127	Genome-wide linkage and haplotype sharing analysis implicates the MCDR3 locus as a candidate region for a developmental macular disorder in association with digit abnormalities. *Ophthalmic Genetics*, **2017**, 38, 511-519	1.2	1
126	Pheno4J: a gene to phenotype graph database. *Bioinformatics*, **2017**, 33, 3317-3319	7.2	6
125	Multisensory cue combination after sensory loss: Audio-visual localization in patients with progressive retinal disease. *Journal of Experimental Psychology: Human Perception and Performance*, **2017**, 43, 729-740	2.6	2

124	Detailed Clinical Phenotype and Molecular Genetic Findings in CLN3-Associated Isolated Retinal Degeneration. *JAMA Ophthalmology*, **2017**, 135, 749-760	3.9	42
123	Visual Acuity Change over 12 Months in the Prospective Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study: ProgStar Report Number 6. *Ophthalmology*, **2017**, 124, 1640-1651	7.3	34
122	Incidence of Atrophic Lesions in Stargardt Disease in the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study: Report No. 5. *JAMA Ophthalmology*, **2017**, 135, 687-695	3.9	36
121	Association of Steroid 5□Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. *JAMA Ophthalmology*, **2017**, 135, 339-347	3.9	32
120	Quantitative Analysis of Retinal Structure Using Spectral-Domain Optical Coherence Tomography in RPGR-Associated Retinopathy. *American Journal of Ophthalmology*, **2017**, 178, 18-26	4.9	24
119	Validation of copy number variation analysis for next-generation sequencing diagnostics. *European Journal of Human Genetics*, **2017**, 25, 719-724	5.3	53
118	Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509	3.9	34
117	Peripheral fundus findings in X-linked retinoschisis. *British Journal of Ophthalmology*, **2017**, 101, 1555-1559	3.9	23
116	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. *American Journal of Human Genetics*, **2017**, 100, 75-90	11	235
115	Vitamin A deficiency due to bi-allelic mutation of RBP4: There's more to it than meets the eye. *Ophthalmic Genetics*, **2017**, 38, 465-466	1.2	16
114	Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options. *British Journal of Ophthalmology*, **2017**, 101, 25-30	5.5	183
113	Progression of Stargardt Disease as Determined by Fundus Autofluorescence in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 9). *JAMA Ophthalmology*, **2017**, 135, 1232-1241	3.9	53
112	Reliability and Repeatability of Cone Density Measurements in Patients With Stargardt Disease and RPGR-Associated Retinopathy **2017**, 58, 3608-3615		28
111	A Quantitative and Qualitative Exploration of Photoaversion in Achromatopsia **2017**, 58, 3537-3546		14
110	The integrity and organization of the human AIPL1 functional domains is critical for its role as a HSP90-dependent co-chaperone for rod PDE6. *Human Molecular Genetics*, **2017**, 26, 4465-4480	5.6	9
109	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. *Scientific Reports*, **2017**, 7, 7512	4.9	17
108	Progression of Visual Acuity and Fundus Autofluorescence in Recent-Onset Stargardt Disease: ProgStar Study Report #4. *Ophthalmology Retina*, **2017**, 1, 514-523	3.8	23
107	Leber congenital amaurosis/early-onset severe retinal dystrophy: clinical features, molecular genetics and therapeutic interventions. *British Journal of Ophthalmology*, **2017**, 101, 1147-1154	5.5	146

106	FUNCTIONAL AND ANATOMICAL OUTCOMES OF CHOROIDAL NEOVASCULARIZATION COMPLICATING BEST1-RELATED RETINOPATHY. *Retina*, **2017**, 37, 1360-1370	3.6	28
105	Unsupervised identification of cone photoreceptors in non-confocal adaptive optics scanning light ophthalmoscope images. *Biomedical Optics Express*, **2017**, 8, 3081-3094	3.5	20
104	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy **2017**, 58, 2906-2914		24
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