Michel Michaelides

List of Publications by Citations

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 effect of gene therapy on Leber congenital amaurosis. *New England Journal of Medicine*, **2015**, 372, 1887-97	59.2	489
266	A prospective randomized trial of intravitreal bevacizumab or laser therapy in the management of diabetic macular edema (BOLT study) 12-month data: report 2. *Ophthalmology*, **2010**, 117, 1078-1086.e2	7.3	379
265	A comparison of the causes of blindness certifications in England and Wales in working age adults (16-64 years), 1999-2000 with 2009-2010. *BMJ Open*, **2014**, 4, e004015	3	313
264	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
263	Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options. *British Journal of Ophthalmology*, **2017**, 101, 25-30	5.5	183
262	Progressive cone and cone-rod dystrophies: phenotypes and underlying molecular genetic basis. *Survey of Ophthalmology*, **2006**, 51, 232-58	6.1	182
261	Mutant prominin 1 found in patients with macular degeneration disrupts photoreceptor disk morphogenesis in mice. *Journal of Clinical Investigation*, **2008**, 118, 2908-16	15.9	154
260	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
259	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. *Nature Genetics*, **2015**, 47, 757-65	36.3	143
258	Retinal structure and function in achromatopsia: implications for gene therapy. *Ophthalmology*, **2014**, 121, 234-245	7.3	119
257	Mutations in CNNM4 cause Jalili syndrome, consisting of autosomal-recessive cone-rod dystrophy and amelogenesis imperfecta. *American Journal of Human Genetics*, **2009**, 84, 266-73	11	117
256	Deep intronic mutation in OFD1, identified by targeted genomic next-generation sequencing, causes a severe form of X-linked retinitis pigmentosa (RP23). *Human Molecular Genetics*, **2012**, 21, 3647-54	5.6	115
255	Clinical and molecular characteristics of childhood-onset Stargardt disease. *Ophthalmology*, **2015**, 122, 326-34	7.3	111
254	Functional characteristics of patients with retinal dystrophy that manifest abnormal parafoveal annuli of high density fundus autofluorescence; a review and update. *Documenta Ophthalmologica*, **2008**, 116, 79-89	2.2	111
253	Differentiating drusen: Drusen and drusen-like appearances associated with ageing, age-related macular degeneration, inherited eye disease and other pathological processes. *Progress in Retinal and Eye Research*, **2016**, 53, 70-106	20.5	109
252	Transplantation of Human Embryonic Stem Cell-Derived Retinal Pigment Epithelial Cells in Macular Degeneration. *Ophthalmology*, **2018**, 125, 1765-1775	7.3	101
251	The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Studies: Design and Baseline Characteristics: ProgStar Report No. 1. *Ophthalmology*, **2016**, 123, 817-28	7.3	94

250	A longitudinal study of Stargardt disease: quantitative assessment of fundus autofluorescence, progression, and genotype correlations **2013**, 54, 8181-90		94
249	The cone dysfunction syndromes. *British Journal of Ophthalmology*, **2016**, 100, 115-21	5.5	91
248	Phenotypic variation in enhanced S-cone syndrome. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 2082-93		91
247	Mutations in the gene KCNV2 encoding a voltage-gated potassium channel subunit cause "cone dystrophy with supernormal rod electroretinogram" in humans. *American Journal of Human Genetics*, **2006**, 79, 574-9	11	91
246	A longitudinal study of stargardt disease: clinical and electrophysiologic assessment, progression, and genotype correlations. *American Journal of Ophthalmology*, **2013**, 155, 1075-1088.e13	4.9	88
245	Identification of novel RPGR ORF15 mutations in X-linked progressive cone-rod dystrophy (XLCORD) families. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 1891-8		87
244	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. *American Journal of Human Genetics*, **2016**, 99, 1305-1315	11	84
243	Clinical and molecular analysis of Stargardt disease with preserved foveal structure and function. *American Journal of Ophthalmology*, **2013**, 156, 487-501.e1	4.9	84
242	Human cone visual pigment deletions spare sufficient photoreceptors to warrant gene therapy. *Human Gene Therapy*, **2013**, 24, 993-1006	4.8	77
241	Progressive cone and cone-rod dystrophies: clinical features, molecular genetics and prospects for therapy. *British Journal of Ophthalmology*, **2019**,	5.5	74
240	The PROM1 mutation p.R373C causes an autosomal dominant bull's eye maculopathy associated with rod, rod-cone, and macular dystrophy **2010**, 51, 4771-80		70
239	The effect of cone opsin mutations on retinal structure and the integrity of the photoreceptor mosaic **2012**, 53, 8006-15		69
238	Progressive cone dystrophy associated with mutation in CNGB3. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 1975-82		61
237	Macular perfusion determined by fundus fluorescein angiography at the 4-month time point in a prospective randomized trial of intravitreal bevacizumab or laser therapy in the management of diabetic macular edema (Bolt Study): Report 1. *Retina*, **2010**, 30, 781-6	3.6	60
236	Achromatopsia: clinical features, molecular genetics, animal models and therapeutic options. *Ophthalmic Genetics*, **2018**, 39, 149-157	1.2	59
235	Detailed phenotypic and genotypic characterization of bietti crystalline dystrophy. *Ophthalmology*, **2014**, 121, 1174-84	7.3	59
234	"Cone dystrophy with supernormal rod electroretinogram": a comprehensive genotype/phenotype study including fundus autofluorescence and extensive electrophysiology. *Retina*, **2010**, 30, 51-62	3.6	57
233	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

232	Genotype-dependent variability in residual cone structure in achromatopsia: toward developing metrics for assessing cone health **2014**, 55, 7303-11		56
231	Cone-rod dystrophy, intrafamilial variability, and incomplete penetrance associated with the R172W mutation in the peripherin/RDS gene. *Ophthalmology*, **2005**, 112, 1592-8	7.3	56
230	A prospective longitudinal study of retinal structure and function in achromatopsia **2014**, 55, 5733-43		55
229	RPGR-associated retinopathy: clinical features, molecular genetics, animal models and therapeutic options. *British Journal of Ophthalmology*, **2016**, 100, 1022-7	5.5	54
228	The phenotype of Severe Early Childhood Onset Retinal Dystrophy (SECORD) from mutation of RPE65 and differentiation from Leber congenital amaurosis **2011**, 52, 292-302		54
227	Validation of copy number variation analysis for next-generation sequencing diagnostics. *European Journal of Human Genetics*, **2017**, 25, 719-724	5.3	53
226	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	7.9	53
225	Potential of handheld optical coherence tomography to determine cause of infantile nystagmus in children by using foveal morphology. *Ophthalmology*, **2013**, 120, 2714-2724	7.3	53
224	Guanylate cyclases and associated activator proteins in retinal disease. *Molecular and Cellular Biochemistry*, **2010**, 334, 157-68	4.2	53
223	Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). *Ophthalmology*, **2016**, 123, 1887-97	7.3	52
222	An early-onset autosomal dominant macular dystrophy (MCDR3) resembling North Carolina macular dystrophy maps to chromosome 5. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 2178-83		51
221	The clinical effect of homozygous ABCA4 alleles in 18 patients. *Ophthalmology*, **2013**, 120, 2324-31	7.3	50
220	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
219	Clinical and Genetic Features of Choroideremia in Childhood. *Ophthalmology*, **2016**, 123, 2158-65	7.3	46
218	Assessing retinal structure in complete congenital stationary night blindness and Oguchi disease. *American Journal of Ophthalmology*, **2012**, 154, 987-1001.e1	4.9	45
217	Safety and Proof-of-Concept Study of Oral QLT091001 in Retinitis Pigmentosa Due to Inherited Deficiencies of Retinal Pigment Epithelial 65 Protein (RPE65) or Lecithin:Retinol Acyltransferase (LRAT). *PLoS ONE*, **2015**, 10, e0143846	3.7	43
216	Macular dystrophy associated with the A3243G mitochondrial DNA mutation. Distinct retinal and associated features, disease variability, and characterization of asymptomatic family members. *JAMA Ophthalmology*, **2008**, 126, 320-8		43
215	Adaptive optics imaging of inherited retinal diseases. *British Journal of Ophthalmology*, **2018**, 102, 1028-1035	3.5	43

214	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
213	Detailed Clinical Phenotype and Molecular Genetic Findings in CLN3-Associated Isolated Retinal Degeneration. *JAMA Ophthalmology*, **2017**, 135, 749-760	3.9	42
212	The extended clinical phenotype of dome-shaped macula. *Graefe's Archive for Clinical and Experimental Ophthalmology*, **2014**, 252, 499-508	3.8	42
211	X-linked cone dysfunction syndrome with myopia and protanopia. *Ophthalmology*, **2005**, 112, 1448-54	7.3	42
210	Variations in opsin coding sequences cause x-linked cone dysfunction syndrome with myopia and dichromacy **2013**, 54, 1361-9		41
209	High-resolution optical coherence tomography imaging in KCNV2 retinopathy. *British Journal of Ophthalmology*, **2012**, 96, 213-7	5.5	41
208	Early Patterns of Macular Degeneration in ABCA4-Associated Retinopathy. *Ophthalmology*, **2018**, 125, 735-746	7.3	40
207	Mutation in the gene GUCA1A, encoding guanylate cyclase-activating protein 1, causes cone, cone-rod, and macular dystrophy. *Ophthalmology*, **2005**, 112, 1442-7	7.3	40
206	Efficacy and prognostic factors of response to carbonic anhydrase inhibitors in management of cystoid macular edema in retinitis pigmentosa. *Investigative Ophthalmology and Visual Science*, **2015**, 56, 1531-6		39
205	Visual hallucinations in neurological and ophthalmological disease: pathophysiology and management. *Journal of Neurology, Neurosurgery and Psychiatry*, **2020**, 91, 512-519	5.5	38
204	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
203	Maculopathy due to the R345W substitution in fibulin-3: distinct clinical features, disease variability, and extent of retinal dysfunction. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 3085-97		38
202	An autosomal dominant bull's-eye macular dystrophy (MCDR2) that maps to the short arm of chromosome 4. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 1657-62		38
201	Three different cone opsin gene array mutational mechanisms with genotype-phenotype correlation and functional investigation of cone opsin variants. *Human Mutation*, **2014**, 35, 1354-62	4.7	37
200	Blue cone monochromacy: causative mutations and associated phenotypes. *Molecular Vision*, **2009**, 15, 876-84	2.3	37
199	Macular dystrophies: clinical and imaging features, molecular genetics and therapeutic options. *British Journal of Ophthalmology*, **2020**, 104, 451-460	5.5	37
198	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
197	A clinical and molecular characterisation of CRB1-associated maculopathy. *European Journal of Human Genetics*, **2018**, 26, 687-694	5.3	36

196	ABCA4 gene screening by next-generation sequencing in a British cohort **2013**, 54, 6662-74		36
195	Retinal gene therapy. *British Medical Bulletin*, **2018**, 126, 13-25	5.4	35
194	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
193	X-linked cone dystrophy caused by mutation of the red and green cone opsins. *American Journal of Human Genetics*, **2010**, 87, 26-39	11	35
192	Reliability and Repeatability of Cone Density Measurements in Patients with Congenital Achromatopsia. *Advances in Experimental Medicine and Biology*, **2016**, 854, 277-83	3.6	35
191	Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. *British Journal of Ophthalmology*, **2019**, 103, 1163-1166	5.5	35
190	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
189	Childhood-onset Leber hereditary optic neuropathy. *British Journal of Ophthalmology*, **2017**, 101, 1505-1509	3.9	34
188	Association of Steroid 5Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. *JAMA Ophthalmology*, **2017**, 135, 339-347	3.9	32
187	Adaptive Optics Retinal Imaging in CNGA3-Associated Achromatopsia: Retinal Characterization, Interocular Symmetry, and Intrafamilial Variability **2019**, 60, 383-396		32
186	Novel mutations and electrophysiologic findings in RGS9- and R9AP-associated retinal dysfunction (Bradyopsia). *Ophthalmology*, **2010**, 117, 120-127.e1	7.3	31
185	Glaucoma following congenital cataract surgery--the role of early surgery and posterior capsulotomy. *BMC Ophthalmology*, **2007**, 7, 13	2.3	31
184	Deep Phenotyping of PDE6C-Associated Achromatopsia **2019**, 60, 5112-5123		31
183	Automatic Cone Photoreceptor Localisation in Healthy and Stargardt Afflicted Retinas Using Deep Learning. *Scientific Reports*, **2018**, 8, 7911	4.9	31
182	Integrity of the cone photoreceptor mosaic in oligocone trichromacy **2011**, 52, 4757-64		30
181	Longitudinal Assessment of Retinal Structure in Achromatopsia Patients With Long-Term Follow-up **2018**, 59, 5735-5744		30
180	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. *Annals of Neurology*, **2019**, 86, 368-383	9.4	29
179	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

178	Reliability and Repeatability of Cone Density Measurements in Patients With Stargardt Disease and RPGR-Associated Retinopathy **2017**, 58, 3608-3615		28
177	Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis. *American Journal of Human Genetics*, **2016**, 99, 1338-1352	11	28
176	FUNCTIONAL AND ANATOMICAL OUTCOMES OF CHOROIDAL NEOVASCULARIZATION COMPLICATING BEST1-RELATED RETINOPATHY. *Retina*, **2017**, 37, 1360-1370	3.6	28
175	Preserved outer retina in AIPL1 Leber congenital amaurosis: implications for gene therapy. *Ophthalmology*, **2015**, 122, 862-4	7.3	27
174	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
173	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
172	Characterization of Retinal Structure in ATF6-Associated Achromatopsia **2019**, 60, 2631-2640		27
171	QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. *Retina*, **2018**, 38, 2401-2414	3.6	26
170	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
169	Leber congenital amaurosis associated with AIPL1: challenges in ascribing disease causation, clinical findings, and implications for gene therapy. *PLoS ONE*, **2012**, 7, e32330	3.7	26
168	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
167	Cone Photoreceptor Structure in Patients With X-Linked Cone Dysfunction and Red-Green Color Vision Deficiency **2016**, 57, 3853-63		26
166	Inherited cataracts: molecular genetics, clinical features, disease mechanisms and novel therapeutic approaches. *British Journal of Ophthalmology*, **2020**, 104, 1331-1337	5.5	25
165	Molecular and Clinical Findings in Patients With Knobloch Syndrome. *JAMA Ophthalmology*, **2016**, 134, 753-62	3.9	25
164	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140 **2016**, 57, 1053-62		25
163	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
162	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
161	Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4 **2016**, 57, 4668-78		24

160	Peripheral fundus findings in X-linked retinoschisis. *British Journal of Ophthalmology*, **2017**, 101, 1555-1559	3.3	23
159	Expanding the Phenotype of TRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. *JAMA Ophthalmology*, **2016**, 134, 1049-53	3.9	23
158	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
157	Phenotypic findings in C1QTNF5 retinopathy (late-onset retinal degeneration). *Acta Ophthalmologica*, **2013**, 91, e191-5	3.7	23
156	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
155	Comparison of Short-Wavelength Reduced-Illuminance and Conventional Autofluorescence Imaging in Stargardt Macular Dystrophy. *American Journal of Ophthalmology*, **2016**, 168, 269-278	4.9	23
154	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. *Ophthalmology*, **2016**, 123, 668-71.e2	7.3	23
153	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
152	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
151	Extended extraocular phenotype of PROM1 mutation in kindreds with known autosomal dominant macular dystrophy. *European Journal of Human Genetics*, **2011**, 19, 131-7	5.3	22
150	Effects of Intraframe Distortion on Measures of Cone Mosaic Geometry from Adaptive Optics Scanning Light Ophthalmoscopy. *Translational Vision Science and Technology*, **2016**, 5, 10	3.3	22
149	The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients **2016**, 57, 5963-5973		22
148	Unilateral BEST1-Associated Retinopathy. *American Journal of Ophthalmology*, **2016**, 169, 24-32	4.9	21
147	Treatments for dry age-related macular degeneration: therapeutic avenues, clinical trials and future directions. *British Journal of Ophthalmology*, **2021**,	5.5	21
146	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
145	Residual Cone Structure in Patients With X-Linked Cone Opsin Mutations **2018**, 59, 4238-4248		21
144	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
143	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

142	Unsupervised identification of cone photoreceptors in non-confocal adaptive optics scanning light ophthalmoscope images. *Biomedical Optics Express*, **2017**, 8, 3081-3094	3.5	20
141	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
140	Characterization of Visual Function, Interocular Variability and Progression Using Static Perimetry-Derived Metrics in RPGR-Associated Retinopathy **2018**, 59, 2422-2436		20
139	Photoreceptor Structure in GNAT2-Associated Achromatopsia **2020**, 61, 40		19
138	Interocular Symmetry of Foveal Cone Topography in Congenital Achromatopsia. *Current Eye Research*, **2020**, 45, 1257-1264	2.9	19
137	Missense variants in the X-linked gene PRPS1 cause retinal degeneration in females. *Human Mutation*, **2018**, 39, 80-91	4.7	19
136	Evidence of genetic heterogeneity in MRCS (microcornea, rod-cone dystrophy, cataract, and posterior staphyloma) syndrome. *American Journal of Ophthalmology*, **2006**, 141, 418-20	4.9	19
135	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
134	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
133	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
132	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. *JAMA Ophthalmology*, **2017**, 135, 137-144	3.9	17
131	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
130	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. *Scientific Reports*, **2017**, 7, 7512	4.9	17
129	Cone dystrophy with "supernormal" rod ERG: psychophysical testing shows comparable rod and cone temporal sensitivity losses with no gain in rod function **2014**, 55, 832-40		17
128	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
127	Preserved visual function in retinal dystrophy due to hypomorphic mutations. *British Journal of Ophthalmology*, **2016**, 100, 1499-1505	5.5	17
126	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. *Retina*, **2019**, 39, 570-580	3.6	17
125	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

124	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
123	Fine central macular dots associated with childhood-onset Stargardt Disease. *Acta Ophthalmologica* , **2014**, 92, e157-9	3.7	16
122	A Cross-Sectional and Longitudinal Study of Retinal Sensitivity in RPE65-Associated Leber Congenital Amaurosis **2018**, 59, 3330-3339		16
121	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
120	retinopathy: clinical features, molecular genetics and directions for future therapy. *Ophthalmic Genetics*, **2020**, 41, 208-215	1.2	15
119	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1 **2016**, 57, 4806-13		15
118	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. *Ophthalmology*, **2016**, 123, 1624-6	7.3	15
117	Assessing the Interocular Symmetry of Foveal Outer Nuclear Layer Thickness in Achromatopsia. *Translational Vision Science and Technology*, **2019**, 8, 21	3.3	15
116	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
115	A Quantitative and Qualitative Exploration of Photoaversion in Achromatopsia **2017**, 58, 3537-3546		14
114	Vision in observers with enhanced S-cone syndrome: an excess of s-cones but connected mainly to conventional s-cone pathways **2014**, 55, 963-76		14
113	Fast adaptive optics scanning light ophthalmoscope retinal montaging. *Biomedical Optics Express*, **2018**, 9, 4317-4328	3.5	14
112	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. *American Journal of Ophthalmology*, **2019**, 207, 87-98	4.9	13
111	Retinal Structure in RPE65-Associated Retinal Dystrophy **2020**, 61, 47		13
110	Long-Term Investigation of Retinal Function in Patients with Achromatopsia **2020**, 61, 38		13
109	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. *PLoS Genetics*, **2018**, 14, e1007504	6	13
108	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
107	Developmental macular disorders: phenotypes and underlying molecular genetic basis. *British Journal of Ophthalmology*, **2012**, 96, 917-24	5.5	12

106	Retinal vein occlusion and angle closure: a retrospective case series. *Journal of Glaucoma*, **2010**, 19, 643-9	2.1	12
105	Inherited retinal diseases: Therapeutics, clinical trials and end points-A review. *Clinical and Experimental Ophthalmology*, **2021**, 49, 270-288	2.4	12
104	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
103	Novel heterozygous mutation in YAP1 in a family with isolated ocular colobomas. *Ophthalmic Genetics*, **2017**, 38, 281-283	1.2	11
102	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
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