Michel Michaelides

List of Publications by Year
in descending order

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277	12,527	34016	43802
papers	citations	52 h-index	91 g-index

287	287	287	9301
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Long-Term Effect of Gene Therapy on Leberâ€™s Congenital Amaurosis. New England Journal of Medicine, 2015, 372, 1887-1897.	13.9	635
2	A Prospective Randomized Trial of Intravitreal Bevacizumab or Laser Therapy in the Management of Diabetic Macular Edema (BOLT Study). Ophthalmology, 2010, 117, 1078-1086.e2.	2.5	473
3	A comparison of the causes of blindness certifications in England and Wales in working age adults (16â€“64â€…years), 1999â€“2000 with 2009â€“2010. BMJ Open, 2014, 4, e004015.	0.8	465
4	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.	2.6	343
5	Stargardt disease: clinical features, molecular genetics, animal models and therapeutic options. British Journal of Ophthalmology, 2017, 101, 25-30.	2.1	265
6	Leber congenital amaurosis/early-onset severe retinal dystrophy: clinical features, molecular genetics and therapeutic interventions. British Journal of Ophthalmology, 2017, 101, 1147-1154.	2.1	231
7	Progressive Cone and Cone-Rod Dystrophies: Phenotypes and Underlying Molecular Genetic Basis. Survey of Ophthalmology, 2006, 51, 232-258.	1.7	208
8	Mutant prominin 1 found in patients with macular degeneration disrupts photoreceptor disk morphogenesis in mice. Journal of Clinical Investigation, 2008, 118, 2908-16.	3.9	194
9	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765.	9.4	183
10	Transplantation of Human Embryonic Stem Cell-Derived Retinal Pigment Epithelial Cells in Macular Degeneration. Ophthalmology, 2018, 125, 1765-1775.	2.5	177
11	The cone dysfunction syndromes: TableÂ1. British Journal of Ophthalmology, 2016, 100, 115-121.	2.1	170
12	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.	7.3	159
13	Clinical and Molecular Characteristics ofÂChildhood-Onset Stargardt Disease. Ophthalmology, 2015, 122, 326-334.	2.5	146
14	Retinal Structure and Function in Achromatopsia. Ophthalmology, 2014, 121, 234-245.	2.5	145
15	Mutations in CNNM4 Cause Jalili Syndrome, Consisting of Autosomal-Recessive Cone-Rod Dystrophy and Amelogenesis Imperfecta. American Journal of Human Genetics, 2009, 84, 266-273.	2.6	143
16	Progressive cone and cone-rod dystrophies: clinical features, molecular genetics and prospects for therapy. British Journal of Ophthalmology, 2019, 103, 711-720.	2.1	140
17	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, 3647-3654.	1.4	133
18	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.	2.5	131

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19	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.	1.0	129
20	The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Studies. Ophthalmology, 2016, 123, 817-828.	2.5	126
21	A Longitudinal Study of Stargardt Disease: Clinical and Electrophysiologic Assessment, Progression, and Genotype Correlations. American Journal of Ophthalmology, 2013, 155, 1075-1088.e13.	1.7	121
22	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	2.6	121
23	A Longitudinal Study of Stargardt Disease: Quantitative Assessment of Fundus Autofluorescence, Progression, and Genotype Correlations. , 2013, 54, 8181.		119
24	Mutations in the Gene KCNV2 Encoding a Voltage-Gated Potassium Channel Subunit Cause â€œCone Dystrophy with Supernormal Rod Electroretinogramâ€•in Humans. American Journal of Human Genetics, 2006, 79, 574-579.	2.6	112
25	Phenotypic Variation in Enhanced S-cone Syndrome. , 2008, 49, 2082.		107
26	Identification of NovelRPGRORF15 Mutations in X-linked Progressive Cone-Rod Dystrophy (XLCORD) Families. , 2005, 46, 1891.		104
27	Clinical and Molecular Analysis of Stargardt Disease With Preserved Foveal Structure and Function. American Journal of Ophthalmology, 2013, 156, 487-501.e1.	1.7	100
28	Human Cone Visual Pigment Deletions Spare Sufficient Photoreceptors to Warrant Gene Therapy. Human Gene Therapy, 2013, 24, 993-1006.	1.4	97
29	ThePROM1Mutation p.R373C Causes an Autosomal Dominant Bull's Eye Maculopathy Associated with Rod, Rodâ€“Cone, and Macular Dystrophy. , 2010, 51, 4771.		96
30	Treatments for dry age-related macular degeneration: therapeutic avenues, clinical trials and future directions. British Journal of Ophthalmology, 2022, 106, 297-304.	2.1	86
31	The Effect of Cone Opsin Mutations on Retinal Structure and the Integrity of the Photoreceptor Mosaic. , 2012, 53, 8006.		85
32	Achromatopsia: clinical features, molecular genetics, animal models and therapeutic options. Ophthalmic Genetics, 2018, 39, 149-157.	0.5	82
33	Detailed Phenotypic and Genotypic Characterization of Bietti Crystalline Dystrophy. Ophthalmology, 2014, 121, 1174-1184.	2.5	79
34	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.	1.4	77
35	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.	2.6	75
36	Visual hallucinations in neurological and ophthalmological disease: pathophysiology and management. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 512-519.	0.9	75

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37	Progressive Cone Dystrophy Associated with Mutation inCNGB3. , 2004, 45, 1975.		74
38	Coneâ€“Rod Dystrophy, Intrafamilial Variability, and Incomplete Penetrance Associated with the R172W Mutation in the Peripherin/RDS Gene. Ophthalmology, 2005, 112, 1592-1598.	2.5	72
39	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). Retina, 2010, 30, 781-786.	1.0	72
40	Validation of copy number variation analysis for next-generation sequencing diagnostics. European Journal of Human Genetics, 2017, 25, 719-724.	1.4	72
41	Macular dystrophies: clinical and imaging features, molecular genetics and therapeutic options. British Journal of Ophthalmology, 2020, 104, 451-460.	2.1	72
42	â€œCONE DYSTROPHY WITH SUPERNORMAL ROD ELECTRORETINOGRAMâ€• A COMPREHENSIVE GENOTYPE/PHENOTYPE STUDY INCLUDING FUNDUS AUTOFLUORESCENCE AND EXTENSIVE ELECTROPHYSIOLOGY. Retina, 2010, 30, 51-62.	1.0	71
43	A Prospective Longitudinal Study of Retinal Structure and Function in Achromatopsia. , 2014, 55, 5733.		68
44	RPGR-associated retinopathy: clinical features, molecular genetics, animal models and therapeutic options. British Journal of Ophthalmology, 2016, 100, 1022-1027.	2.1	68
45	Inherited retinal diseases: Therapeutics, clinical trials and end pointsâ€”A review. Clinical and Experimental Ophthalmology, 2021, 49, 270-288.	1.3	68
46	Genotype-Dependent Variability in Residual Cone Structure in Achromatopsia: Toward Developing Metrics for Assessing Cone Health. , 2014, 55, 7303.		67
47	The Phenotype of Severe Early Childhood Onset Retinal Dystrophy (SECORD) from Mutation ofRPE65and Differentiation from Leber Congenital Amaurosis. , 2011, 52, 292.		65
48	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. Progress in Retinal and Eye Research, 2021, 82, 100898.	7.3	65
49	Restoration of visual function in advanced disease after transplantation of purified human pluripotent stem cell-derived cone photoreceptors. Cell Reports, 2021, 35, 109022.	2.9	65
50	Clinical and Genetic Features of Choroideremia in Childhood. Ophthalmology, 2016, 123, 2158-2165.	2.5	63
51	Guanylate cyclases and associated activator proteins in retinal disease. Molecular and Cellular Biochemistry, 2010, 334, 157-168.	1.4	62
52	Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509.	2.1	62
53	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
54	Detailed Clinical Phenotype and Molecular Genetic Findings in CLN3-Associated Isolated Retinal Degeneration. JAMA Ophthalmology, 2017, 135, 749.	1.4	61

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55	Adaptive optics imaging of inherited retinal diseases. British Journal of Ophthalmology, 2018, 102, 1028-1035.	2.1	61
56	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, Volume 14, 707-719.	0.9	61
57	An Early-Onset Autosomal Dominant Macular Dystrophy (MCDR3) Resembling North Carolina Macular Dystrophy Maps to Chromosome 5. , 2003, 44, 2178.		60
58	Potential of Handheld Optical Coherence Tomography to Determine Cause of Infantile Nystagmus in Children by Using Foveal Morphology. Ophthalmology, 2013, 120, 2714-2724.	2.5	60
59	Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). Ophthalmology, 2016, 123, 1887-1897.	2.5	59
60	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.	2.5	58
61	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.	1.5	57
62	Progression of Stargardt Disease as Determined by Fundus Autofluorescence Over a 12-Month Period. JAMA Ophthalmology, 2019, 137, 1134.	1.4	57
63	The Clinical Effect of Homozygous ABCA4 Alleles in 18 Patients. Ophthalmology, 2013, 120, 2324-2331.	2.5	56
64	Gene therapy for neovascular age-related macular degeneration: rationale, clinical trials and future directions. British Journal of Ophthalmology, 2021, 105, 151-157.	2.1	56
65	Assessing Retinal Structure in Complete Congenital Stationary Night Blindness and Oguchi Disease. American Journal of Ophthalmology, 2012, 154, 987-1001.e1.	1.7	55
66	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.	1.1	55
67	Early Patterns of Macular Degeneration in ABCA4-Associated Retinopathy. Ophthalmology, 2018, 125, 735-746.	2.5	55
68	Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. British Journal of Ophthalmology, 2019, 103, 1163-1166.	2.1	55
69	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, 1531-1536.	3.3	54
70	X-Linked Cone Dysfunction Syndrome with Myopia and Protanopia. Ophthalmology, 2005, 112, 1448-1454.	2.5	53
71	Three Different Cone Opsin Gene Array Mutational Mechanisms; Genotype-Phenotype Correlation and Functional Investigation of Cone Opsin Variants. Human Mutation, 2014, 35, n/a-n/a.	1.1	53
72	Retinal gene therapy. British Medical Bulletin, 2018, 126, 13-25.	2.7	52

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73	The extended clinical phenotype of dome-shaped macula. Graefe's Archive for Clinical and Experimental Ophthalmology, 2014, 252, 499-508.	1.0	51
74	A clinical and molecular characterisation of CRB1-associated maculopathy. European Journal of Human Genetics, 2018, 26, 687-694.	1.4	51
75	Mutation in the Gene GUCA1A, Encoding Guanylate Cyclase-Activating Protein 1, Causes Cone, Cone-Rod, and Macular Dystrophy. Ophthalmology, 2005, 112, 1442-1447.	2.5	50
76	Variations in Opsin Coding Sequences Cause X-Linked Cone Dysfunction Syndrome with Myopia and Dichromacy. , 2013, 54, 1361.		50
77	Macular Dystrophy Associated With the A3243G Mitochondrial DNA Mutation. JAMA Ophthalmology, 2008, 126, 320.	2.6	49
78	High-resolution optical coherence tomography imaging inKCNV2retinopathy. British Journal of Ophthalmology, 2012, 96, 213-217.	2.1	49
79	Automatic Cone Photoreceptor Localisation in Healthy and Stargardt Afflicted Retinas Using Deep Learning. Scientific Reports, 2018, 8, 7911.	1.6	49
80	Inherited cataracts: molecular genetics, clinical features, disease mechanisms and novel therapeutic approaches. British Journal of Ophthalmology, 2020, 104, 1331-1337.	2.1	49
81	Maculopathy Due to the R345W Substitution in Fibulin-3: Distinct Clinical Features, Disease Variability, and Extent of Retinal Dysfunction. , 2006, 47, 3085.		48
82	ABCA4Gene Screening by Next-Generation Sequencing in a British Cohort. , 2013, 54, 6662.		47
83	Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis. American Journal of Human Genetics, 2016, 99, 1338-1352.	2.6	47
84	Incidence of Atrophic Lesions in Stargardt Disease in the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study. JAMA Ophthalmology, 2017, 135, 687.	1.4	47
85	Glaucoma following congenital cataract surgery â€“ the role of early surgery and posterior capsulotomy. BMC Ophthalmology, 2007, 7, 13.	0.6	46
86	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.	2.6	46
87	X-Linked Cone Dystrophy Caused by Mutation of the Red and Green Cone Opsins. American Journal of Human Genetics, 2010, 87, 26-39.	2.6	45
88	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.	2.1	45
89	Blue cone monochromacy: causative mutations and associated phenotypes. Molecular Vision, 2009, 15, 876-84.	1.1	45
90	An Autosomal Dominant Bullâ€™s-Eye Macular Dystrophy (MCDR2) that Maps to the Short Arm of Chromosome 4. , 2003, 44, 1657.		44

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91	Visual Acuity Change Over 24 Months and Its Association With Foveal Phenotype and Genotype in Individuals With Stargardt Disease. JAMA Ophthalmology, 2018, 136, 920.	1.4	44
92	Deep Phenotyping ofPDE6C-Associated Achromatopsia. , 2019, 60, 5112.		44
93	Visual Acuity Change over 12 Months in the Prospective Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study. Ophthalmology, 2017, 124, 1640-1651.	2.5	43
94	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	1.4	43
95	Characterization of Retinal Structure in ATF6-Associated Achromatopsia. , 2019, 60, 2631.		43
96	Adaptive Optics Retinal Imaging inCNGA3-Associated Achromatopsia: Retinal Characterization, Interocular Symmetry, and Intrafamilial Variability. , 2019, 60, 383.		43
97	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.	1.7	43
98	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	1.0	42
99	The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients. , 2016, 57, 5963.		41
100	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	2.8	41
101	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.	1.7	41
102	Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4. , 2016, 57, 4668.		40
103	Peripheral fundus findings in X-linked retinoschisis. British Journal of Ophthalmology, 2017, 101, 1555-1559.	2.1	40
104	Cross-Sectional and Longitudinal Assessment of the Ellipsoid Zone in Childhood-Onset Stargardt Disease. Translational Vision Science and Technology, 2019, 8, 1.	1.1	40
105	Longitudinal Assessment of Retinal Structure in Achromatopsia Patients With Long-Term Follow-up. , 2018, 59, 5735.		39
106	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.	1.7	39
107	Reliability and Repeatability of Cone Density Measurements in Patients with Congenital Achromatopsia. Advances in Experimental Medicine and Biology, 2016, 854, 277-283.	0.8	39
108	Molecular and Clinical Findings in Patients With Knobloch Syndrome. JAMA Ophthalmology, 2016, 134, 753.	1.4	37

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109	Cone Photoreceptor Structure in Patients With X-Linked Cone Dysfunction and Red-Green Color Vision Deficiency. , 2016, 57, 3853.		36
110	Reliability and Repeatability of Cone Density Measurements in Patients With Stargardt Disease and RPGR-Associated Retinopathy. , 2017, 58, 3608.		36
111	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
112	Novel Mutations and Electrophysiologic Findings in RGS9- and R9AP-Associated Retinal Dysfunction (Bradyopsia). Ophthalmology, 2010, 117, 120-127.e1.	2.5	35
113	Phenotypic findings in C1QTNF5 retinopathy (lateâ€onset retinal degeneration). Acta Ophthalmologica, 2013, 91, e191-5.	0.6	35
114	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906.		35
115	Leber congenital amaurosis/early-onset severe retinal dystrophy: current management and clinical trials. British Journal of Ophthalmology, 2022, 106, 445-451.	2.1	35
116	FUNCTIONAL AND ANATOMICAL OUTCOMES OF CHOROIDAL NEOVASCULARIZATION COMPLICATING BEST1-RELATED RETINOPATHY. Retina, 2017, 37, 1360-1370.	1.0	34
117	Integrity of the Cone Photoreceptor Mosaic in Oligocone Trichromacy. , 2011, 52, 4757.		33
118	Effects of Intraframe Distortion on Measures of Cone Mosaic Geometry from Adaptive Optics Scanning Light Ophthalmoscopy. Translational Vision Science and Technology, 2016, 5, 10.	1.1	33
119	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140. , 2016, 57, 1053.		33
120	QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. Retina, 2018, 38, 2401-2414.	1.0	33
121	Preserved Outer Retina in AIPL1 Leber's Congenital Amaurosis: Implications for Gene Therapy. Ophthalmology, 2015, 122, 862-864.	2.5	31
122	Juvenile Batten Disease (CLN3): Detailed Ocular Phenotype, Novel Observations, Delayed Diagnosis, Masquerades, and Prospects for Therapy. Ophthalmology Retina, 2020, 4, 433-445.	1.2	31
123	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.	1.1	31
124	Autosomal Recessive Bestrophinopathy. Ophthalmology, 2021, 128, 706-718.	2.5	31
125	ATF6 is essential for human cone photoreceptor development. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	31
126	Quantitative Analysis of Retinal Structure Using Spectral-Domain Optical Coherence Tomography in RPGR -Associated Retinopathy. American Journal of Ophthalmology, 2017, 178, 18-26.	1.7	30

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127	Characterization of Visual Function, Interocular Variability and Progression Using Static Perimetryâ€“Derived Metrics inRPGR-Associated Retinopathy. , 2018, 59, 2422.		30
128	Comparison of Short-Wavelength Reduced-Illuminance and Conventional Autofluorescence Imaging in Stargardt Macular Dystrophy. American Journal of Ophthalmology, 2016, 168, 269-278.	1.7	29
129	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2.	2.5	29
130	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	1.4	29
131	Residual Cone Structure in Patients With X-Linked Cone Opsin Mutations. , 2018, 59, 4238.		29
132	Progression of Visual Acuity and Fundus Autofluorescence in Recent-Onset Stargardt Disease: ProgStar Study Report #4. Ophthalmology Retina, 2017, 1, 514-523.	1.2	28
133	Leber Congenital Amaurosis Associated with AIPL1: Challenges in Ascribing Disease Causation, Clinical Findings, and Implications for Gene Therapy. PLoS ONE, 2012, 7, e32330.	1.1	28
134	Unsupervised identification of cone photoreceptors in non-confocal adaptive optics scanning light ophthalmoscope images. Biomedical Optics Express, 2017, 8, 3081.	1.5	27
135	Retinal Structure inRPE65-Associated Retinal Dystrophy. , 2020, 61, 47.		27
136	Enhanced S-Cone Syndrome. Ophthalmology Retina, 2021, 5, 195-214.	1.2	27
137	Unilateral BEST1 -Associated Retinopathy. American Journal of Ophthalmology, 2016, 169, 24-32.	1.7	26
138	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.	2.6	26
139	Cross-Sectional and Longitudinal Assessment of Retinal Sensitivity in Patients With Childhood-Onset Stargardt Disease. Translational Vision Science and Technology, 2018, 7, 10.	1.1	26
140	Scotopic Microperimetric Assessment of Rod Function in Stargardt Disease (SMART) Study: Design and Baseline Characteristics (Report No. 1). Ophthalmic Research, 2019, 61, 36-43.	1.0	26
141	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1. , 2016, 57, 4806.		25
142	Preserved visual function in retinal dystrophy due to hypomorphicRPE65mutations. British Journal of Ophthalmology, 2016, 100, 1499-1505.	2.1	25
143	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
144	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.	2.5	25

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145	CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. Retina, 2019, 39, 570-580.	1.0	25
146	The genetic landscape of crystallins in congenital cataract. Orphanet Journal of Rare Diseases, 2020, 15, 333.	1.2	25
147	Photoreceptor Structure inGNAT2-Associated Achromatopsia. , 2020, 61, 40.		25
148	KCNV2 retinopathy: clinical features, molecular genetics and directions for future therapy. Ophthalmic Genetics, 2020, 41, 208-215.	0.5	25
149	Extended extraocular phenotype of PROM1 mutation in kindreds with known autosomal dominant macular dystrophy. European Journal of Human Genetics, 2011, 19, 131-137.	1.4	24
150	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.	1.7	24
151	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	1.4	23
152	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512.	1.6	23
153	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	1.1	23
154	Fast adaptive optics scanning light ophthalmoscope retinal montaging. Biomedical Optics Express, 2018, 9, 4317.	1.5	23
155	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY. Retina, 2019, 39, 514-529.	1.0	23
156	Interocular Symmetry of Foveal Cone Topography in Congenital Achromatopsia. Current Eye Research, 2020, 45, 1257-1264.	0.7	23
157	Evidence of Genetic Heterogeneity in MRCS (Microcornea, Rod-Cone Dystrophy, Cataract, and) Tj ETQq1 1 0.784314 rgBT /Overlock	1.7	22
158	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.	1.7	22
159	Cone Dystrophy With â€œSupernormalâ€•Rod ERG: Psychophysical Testing Shows Comparable Rod and Cone Temporal Sensitivity Losses With No Gain in Rod Function. , 2014, 55, 832.		21
160	Vitamin A deficiency due to bi-allelic mutation of RBP4: Thereâ€™s more to it than meets the eye. Ophthalmic Genetics, 2017, 38, 465-466.	0.5	21
161	Vision in Observers With Enhanced S-Cone Syndrome: An Excess of S-Cones but Connected Mainly to Conventional S-Cone Pathways. , 2014, 55, 963.		20
162	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	1.7	20

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