David G Birch

List of Publications by Year in Descending Order

209 papers	14,130 citations	69 h-index	113 g-index
215 ext. papers	15,498 ext. citations	5.1 avg, IF	5.96 L-index

#	Paper	IF	Citations
209	The RUSH2A Study: Dark-Adapted Visual Fields in Patients With Retinal Degeneration Associated With Biallelic Variants in the USH2A Gene. **2022**, 63, 17		1
208	A Hybrid Model Composed of Two Convolutional Neural Networks (CNNs) for Automatic Retinal Layer Segmentation of OCT Images in Retinitis Pigmentosa (RP). *Translational Vision Science and Technology*, **2021**, 10, 9	3.3	0
207	Identification of Deep-Intronic Splice Mutations in a Large Cohort of Patients With Inherited Retinal Diseases. *Frontiers in Genetics*, **2021**, 12, 647400	4.5	7
206	Longitudinal Changes of Fixation Stability and Location Within 24 Months in Stargardt Disease: ProgStar Report No. 16. *American Journal of Ophthalmology*, **2021**, 233, 78-89	4.9	1
205	Variable expressivity in patients with autosomal recessive retinitis pigmentosa associated with the gene. *Ophthalmic Genetics*, **2021**, 42, 15-22	1.2	3
204	Endpoints for Measuring Efficacy in Clinical Trials for Inherited Retinal Disease. *International Ophthalmology Clinics*, **2021**, 61, 63-78	1.7	
203	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
202	The Effect of Attention on Fixation Stability During Dynamic Fixation Testing in Stargardt Disease. *American Journal of Ophthalmology*, **2020**, 217, 305-316	4.9	4
201	Longitudinal Microperimetric Changes of Macular Sensitivity in Stargardt Disease After 12 Months: ProgStar Report No. 13. *JAMA Ophthalmology*, **2020**, 138, 772-779	3.9	11
200	Application of a Deep Machine Learning Model for Automatic Measurement of EZ Width in SD-OCT Images of RP. *Translational Vision Science and Technology*, **2020**, 9, 15	3.3	7
199	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
198	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
197	X-Chromosome Inactivation Is a Biomarker of Clinical Severity in Female Carriers of RPGR-Associated X-Linked Retinitis Pigmentosa. *Ophthalmology Retina*, **2020**, 4, 510-520	3.8	12
196	Randomised study evaluating the pharmacodynamics of emixustat hydrochloride in subjects with macular atrophy secondary to Stargardt disease. *British Journal of Ophthalmology*, **2020**,	5.5	6
195	Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second Monaciano Symposium. *Translational Vision Science and Technology*, **2020**, 9, 2	3.3	28
194	Disease Progression in Patients with Autosomal Dominant Retinitis Pigmentosa due to a Mutation in Inosine Monophosphate Dehydrogenase 1 (IMPDH1). *Translational Vision Science and Technology*, **2020**, 9, 14	3.3	5
193	A Workshop on Measuring the Progression of Atrophy Secondary to Stargardt Disease in the ProgStar Studies: Findings and Lessons Learned. *Translational Vision Science and Technology*, **2019**, 8, 16	3.3	19

192	Regional Variations and Intra-/Intersession Repeatability for Scotopic Sensitivity in Normal Controls and Patients With Inherited Retinal Degenerations **2019**, 60, 1122-1131		17
191	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
190	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
189	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
188	Histopathologic Assessment of Optic Nerves and Retina From a Patient With Chronically Implanted Argus II Retinal Prosthesis System. *Translational Vision Science and Technology*, **2019**, 8, 31	3.3	5
187	Reliability of Semiautomated Kinetic Perimetry (SKP) and Goldmann Kinetic Perimetry in Children and Adults With Retinal Dystrophies. *Translational Vision Science and Technology*, **2019**, 8, 36	3.3	9
186	Retinal Sensitivity Using Microperimetry in Age-Related Macular Degeneration in an Amish Population. *Ophthalmic Surgery Lasers and Imaging Retina*, **2019**, 50, e236-e241	1.4	9
185	CHOROIDEREMIA: Retinal Degeneration With an Unmet Need. *Retina*, **2019**, 39, 2059-2069	3.6	12
184	Time Course of Disease Progression of PRPF31-mediated Retinitis Pigmentosa. *American Journal of Ophthalmology*, **2019**, 200, 76-84	4.9	16
183	Macular spatial distribution of preserved autofluorescence in patients with choroideremia. *British Journal of Ophthalmology*, **2019**, 103, 933-937	5.5	11
182	An early nonsense mutation facilitates the expression of a short isoform of CNGA3 by alternative translation initiation. *Experimental Eye Research*, **2018**, 171, 48-53	3.7	6
181	Molecular Findings in Families with an Initial Diagnose of Autosomal Dominant Retinitis Pigmentosa (adRP). *Advances in Experimental Medicine and Biology*, **2018**, 1074, 237-245	3.6	4
180	EVALUATION OF FULL-FIELD ELECTRORETINOGRAM REDUCTIONS AFTER OCRIPLASMIN TREATMENT: Results of the OASIS Trial ERG Substudy. *Retina*, **2018**, 38, 364-378	3.6	16
179	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
178	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
177	Effect of Oral Valproic Acid vs Placebo for Vision Loss in Patients With Autosomal Dominant Retinitis Pigmentosa: A Randomized Phase 2 Multicenter Placebo-Controlled Clinical Trial. *JAMA Ophthalmology*, **2018**, 136, 849-856	3.9	24
176	Prospective Evaluation of Patients With X-Linked Retinoschisis During 18 Months **2018**, 59, 5941-5956		9
175	Inherited Retinal Degenerations: Current Landscape and Knowledge Gaps. *Translational Vision Science and Technology*, **2018**, 7, 6	3.3	87

174	Retinal Anatomy and Electrode Array Position in Retinitis Pigmentosa Patients After Argus II Implantation: An International Study. *American Journal of Ophthalmology*, **2018**, 193, 87-99	4.9	14
173	Fixation Location and Stability Using the MP-1 Microperimeter in Stargardt Disease: ProgStar Report No. 3. *Ophthalmology Retina*, **2017**, 1, 68-76	3.8	28
172	Macular Sensitivity Measured With Microperimetry in Stargardt Disease in the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study: Report No. 7. *JAMA Ophthalmology*, **2017** , 135, 696-703	3.9	46
171	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
170	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
169	Dark-Adapted Chromatic Perimetry for Measuring Rod Visual Fields in Patients with Retinitis Pigmentosa. *Translational Vision Science and Technology*, **2017**, 6, 15	3.3	26
168	Metrics and Acquisition Modes for Fixation Stability as a Visual Function Biomarker **2017**, 58, BIO268-BIO276		12
167	A Novel Dominant Mutation in SAG, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States **2017**, 58, 2774-2784		24
166	Leveraging splice-affecting variant predictors and a minigene validation system to identify Mendelian disease-causing variants among exon-captured variants of uncertain significance. *Human Mutation*, **2017**, 38, 1521-1533	4.7	22
165	Next-generation sequencing to solve complex inherited retinal dystrophy: A case series of multiple genes contributing to disease in extended families. *Molecular Vision*, **2017**, 23, 470-481	2.3	14
164	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. *Molecular Genetics and Metabolism Reports*, **2016**, 9, 75-78	1.8	22
163	Five-Year Safety and Performance Results from the Argus II Retinal Prosthesis System Clinical Trial. *Ophthalmology*, **2016**, 123, 2248-54	7.3	209
162	Worldwide Argus II implantation: recommendations to optimize patient outcomes. *BMC Ophthalmology*, **2016**, 16, 52	2.3	32
161	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
160	Treatment of retinitis pigmentosa due to MERTK mutations by ocular subretinal injection of adeno-associated virus gene vector: results of a phase I trial. *Human Genetics*, **2016**, 135, 327-43	6.3	156
159	Identification of a Novel Gene on 10q22.1 Causing Autosomal Dominant Retinitis Pigmentosa (adRP). *Advances in Experimental Medicine and Biology*, **2016**, 854, 193-200	3.6	6
158	North Carolina macular dystrophy (MCDR1) caused by a novel tandem duplication of the gene. *Molecular Vision*, **2016**, 22, 1239-1247	2.3	21
157	Autosomal Dominant Retinal Dystrophies Caused by a Founder Splice Site Mutation, c.828+3A>T, in PRPH2 and Protein Haplotypes in trans as Modifiers **2016**, 57, 349-59		11

156	Structure/Psychophysical Relationships in X-Linked Retinoschisis **2016**, 57, 332-7		18
155	Reliability of a Manual Procedure for Marking the EZ Endpoint Location in Patients with Retinitis Pigmentosa. *Translational Vision Science and Technology*, **2016**, 5, 6	3.3	18
154	Mutation Detection in Patients with Retinal Dystrophies Using Targeted Next Generation Sequencing. *PLoS ONE*, **2016**, 11, e0145951	3.7	76
153	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
152	Quantification of Ellipsoid Zone Changes in Retinitis Pigmentosa Using en Face Spectral Domain-Optical Coherence Tomography. *JAMA Ophthalmology*, **2016**, 134, 628-35	3.9	51
151	Long-term Follow-up of Patients With Retinitis Pigmentosa Receiving Intraocular Ciliary Neurotrophic Factor Implants. *American Journal of Ophthalmology*, **2016**, 170, 10-14	4.9	67
150	Transcription factor PRDM8 is required for rod bipolar and type 2 OFF-cone bipolar cell survival and amacrine subtype identity. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E3010-9	11.5	25
149	Long-Term Results from an Epiretinal Prosthesis to Restore Sight to the Blind. *Ophthalmology*, **2015**, 122, 1547-54	7.3	183
148	Rates of decline in regions of the visual field defined by frequency-domain optical coherence tomography in patients with RPGR-mediated X-linked retinitis pigmentosa. *Ophthalmology*, **2015**, 122, 833-9	7.3	50
147	Founder Effect of a c.828+3A>T Splice Site Mutation in Peripherin 2 (PRPH2) Causing Autosomal Dominant Retinal Dystrophies. *JAMA Ophthalmology*, **2015**, 133, 511-7	3.9	10
146	Phase ii, randomized, placebo-controlled, 90-day study of emixustat hydrochloride in geographic atrophy associated with dry age-related macular degeneration. *Retina*, **2015**, 35, 1173-83	3.6	43
145	Docosahexaenoic Acid Slows Visual Field Progression in X-Linked Retinitis Pigmentosa: Ancillary Outcomes of the DHAX Trial **2015**, 56, 6646-53		36
144	Outer Segment Thickness Predicts Visual Field Response to QLT091001 in Patients with or Mutations. *Translational Vision Science and Technology*, **2015**, 4, 8	3.3	6
143	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
142	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. *Human Genetics*, **2014**, 133, 331-45	6.3	172
141	Safety assessment of docosahexaenoic acid in X-linked retinitis pigmentosa: the 4-year DHAX trial **2014**, 55, 4958-66		21
140	A comparison of progressive loss of the ellipsoid zone (EZ) band in autosomal dominant and x-linked retinitis pigmentosa **2014**, 55, 7417-22		72
139	A dominant mutation in hexokinase 1 (HK1) causes retinitis pigmentosa **2014**, 55, 7147-58		36

138	Four-year placebo-controlled trial of docosahexaenoic acid in X-linked retinitis pigmentosa (DHAX trial): a randomized clinical trial. *JAMA Ophthalmology*, **2014**, 132, 866-73	3.9	45
137	Mutations in the X-linked retinitis pigmentosa genes RPGR and RP2 found in 8.5% of families with a provisional diagnosis of autosomal dominant retinitis pigmentosa **2013**, 54, 1411-6		101
136	Normative reference ranges for the retinal nerve fiber layer, macula, and retinal layer thicknesses in children. *American Journal of Ophthalmology*, **2013**, 155, 354-360.e1	4.9	123
135	Randomized trial of ciliary neurotrophic factor delivered by encapsulated cell intraocular implants for retinitis pigmentosa. *American Journal of Ophthalmology*, **2013**, 156, 283-292.e1	4.9	133
134	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. *Journal of Medical Genetics*, **2013**, 50, 674-88	5.8	125
133	Spectral-domain optical coherence tomography measures of outer segment layer progression in patients with X-linked retinitis pigmentosa. *JAMA Ophthalmology*, **2013**, 131, 1143-50	3.9	125
132	A Comparison of Methods for Tracking Progression in X-Linked Retinitis Pigmentosa Using Frequency Domain OCT. *Translational Vision Science and Technology*, **2013**, 2, 5	3.3	35
131	Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200) cause 1.6% of autosomal dominant retinitis pigmentosa. *Molecular Vision*, **2013**, 19, 2407-17	2.3	20
130	Polymorphic variation of RPGRIP1L and IQCB1 as modifiers of X-linked retinitis pigmentosa caused by mutations in RPGR. *Advances in Experimental Medicine and Biology*, **2012**, 723, 313-20	3.6	14
129	Characterizing the phenotype and genotype of a family with occult macular dystrophy. *JAMA Ophthalmology*, **2012**, 130, 1554-9		20
128	Mutations in RPGR and RP2 account for 15% of males with simplex retinal degenerative disease **2012**, 53, 8232-7		91
127	Relationships among multifocal electroretinogram amplitude, visual field sensitivity, and SD-OCT receptor layer thicknesses in patients with retinitis pigmentosa **2012**, 53, 833-40		50
126	GPR179 is required for depolarizing bipolar cell function and is mutated in autosomal-recessive complete congenital stationary night blindness. *American Journal of Human Genetics*, **2012**, 90, 331-9	11	108
125	Loss of caveolin-1 impairs retinal function due to disturbance of subretinal microenvironment. *Journal of Biological Chemistry*, **2012**, 287, 16424-34	5.4	38
124	Safety and effect on rod function of ACU-4429, a novel small-molecule visual cycle modulator. *Retina*, **2012**, 32, 183-8	3.6	81
123	Rod photoreceptor temporal properties in retinal degenerative diseases. *Advances in Experimental Medicine and Biology*, **2012**, 723, 495-502	3.6	2
122	Rod photoreceptor temporal properties in retinitis pigmentosa. *Experimental Eye Research*, **2011**, 92, 202-8	3.7	7
121	Method for deriving visual field boundaries from OCT scans of patients with retinitis pigmentosa. *Biomedical Optics Express*, **2011**, 2, 1106-14	3.5	70

120	Phenotypic characterization of 3 families with autosomal dominant retinitis pigmentosa due to mutations in KLHL7. *JAMA Ophthalmology*, **2011**, 129, 1475-82		27
119	Identification of disease-causing mutations in autosomal dominant retinitis pigmentosa (adRP) using next-generation DNA sequencing **2011**, 52, 494-503		77
118	Pediatric Hereditary Macular Degenerations **2011**, 245-294		
117	Transition zones between healthy and diseased retina in choroideremia (CHM) and Stargardt disease (STGD) as compared to retinitis pigmentosa (RP) **2011**, 52, 9581-90		59
116	The transition zone between healthy and diseased retina in patients with retinitis pigmentosa **2011** , 52, 101-8		107
115	The inner segment/outer segment border seen on optical coherence tomography is less intense in patients with diminished cone function **2011**, 52, 9703-9		89
114	Abnormal cone structure in foveal schisis cavities in X-linked retinoschisis from mutations in exon 6 of the RS1 gene **2011**, 52, 9614-23		31
113	Rod sensitivity, cone sensitivity, and photoreceptor layer thickness in retinal degenerative diseases **2011**, 52, 7141-7		71
112	Ciliary neurotrophic factor delivered by encapsulated cell intraocular implants for treatment of geographic atrophy in age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 6241-5	11.5	218
111	ApoER2 function in the establishment and maintenance of retinal synaptic connectivity. *Journal of Neuroscience*, **2011**, 31, 14413-23	6.6	22
110	Allelic heterogeneity and genetic modifier loci contribute to clinical variation in males with X-linked retinitis pigmentosa due to RPGR mutations. *PLoS ONE*, **2011**, 6, e23021	3.7	71
109	Electroretinographic Testing in Infants and Children **2011**, 67-84		
108	A comparison of visual field sensitivity to photoreceptor thickness in retinitis pigmentosa **2010**, 51, 4213-9		107
107	Macular atrophy in birdshot retinochoroidopathy: an optical coherence tomography and multifocal electroretinography analysis. *Retina*, **2010**, 30, 930-7	3.6	34
106	Targeted high-throughput DNA sequencing for gene discovery in retinitis pigmentosa. *Advances in Experimental Medicine and Biology*, **2010**, 664, 325-31	3.6	30
105	Thickness of receptor and post-receptor retinal layers in patients with retinitis pigmentosa measured with frequency-domain optical coherence tomography **2009**, 50, 2328-36		170
104	Mutations in a BTB-Kelch protein, KLHL7, cause autosomal-dominant retinitis pigmentosa. *American Journal of Human Genetics*, **2009**, 84, 792-800	11	84
103	A novel GCAP1(N104K) mutation in EF-hand 3 (EF3) linked to autosomal dominant cone dystrophy. *Vision Research*, **2008**, 48, 2425-32	2.1	33

102	Mutation analysis identifies GUCY2D as the major gene responsible for autosomal dominant progressive cone degeneration **2008**, 49, 5015-23		54
101	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
100	Mutations in the TOPORS gene cause 1% of autosomal dominant retinitis pigmentosa. *Molecular Vision*, **2008**, 14, 922-7	2.3	32
99	Mutations in known genes account for 58% of autosomal dominant retinitis pigmentosa (adRP). *Advances in Experimental Medicine and Biology*, **2008**, 613, 203-9	3.6	22
98	Persistent cone dysfunction in acute exudative polymorphous vitelliform maculopathy. *Retina*, **2007**, 27, 109-13	3.6	11
97	Retinal pathology and skin barrier defect in mice carrying a Stargardt disease-3 mutation in elongase of very long chain fatty acids-4. *Molecular Vision*, **2007**, 13, 258-72	2.3	76
96	Age-related macular degeneration: a target for nanotechnology derived medicines. *International Journal of Nanomedicine*, **2007**, 2, 65-77	7.3	44
95	The Gly56Arg mutation in NR2E3 accounts for 1-2% of autosomal dominant retinitis pigmentosa. *Molecular Vision*, **2007**, 13, 1970-5	2.3	40
94	Spectrum and frequency of mutations in IMPDH1 associated with autosomal dominant retinitis pigmentosa and leber congenital amaurosis. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 34-42		115
93	The very large G-protein-coupled receptor VLGR1: a component of the ankle link complex required for the normal development of auditory hair bundles. *Journal of Neuroscience*, **2006**, 26, 6543-53	6.6	154
92	Prevalence of disease-causing mutations in families with autosomal dominant retinitis pigmentosa: a screen of known genes in 200 families. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 3052-64		214
91	Genomic rearrangements of the PRPF31 gene account for 2.5% of autosomal dominant retinitis pigmentosa. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 4579-88		65
90	The Iroquois homeobox gene, Irx5, is required for retinal cone bipolar cell development. *Developmental Biology*, **2005**, 287, 48-60	3.1	77
89	Phenotypic characterization of a large family with RP10 autosomal-dominant retinitis pigmentosa: an Asp226Asn mutation in the IMPDH1 gene. *American Journal of Ophthalmology*, **2005**, 140, 858-867	4.9	14
88	Visual maturation of term infants fed long-chain polyunsaturated fatty acid-supplemented or control formula for 12 mo. *American Journal of Clinical Nutrition*, **2005**, 81, 871-9	7	130
87	A randomized placebo-controlled clinical trial of docosahexaenoic acid (DHA) supplementation for X-linked retinitis pigmentosa. *Retina*, **2005**, 25, S52-S54	3.6	9
86	Outcome measures and their application in clinical trials for retinal degenerative diseases: outline, review, and perspective. *Retina*, **2005**, 25, 772-7	3.6	36
85	Retinal disease in mice lacking hypoxia-inducible transcription factor-2alpha. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 1010-6		31

84	Lipofuscin accumulation, abnormal electrophysiology, and photoreceptor degeneration in mutant ELOVL4 transgenic mice: a model for macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 4164-9	11.5	158
83	Functional characterization of mouse RDH11 as a retinol dehydrogenase involved in dark adaptation in vivo. *Journal of Biological Chemistry*, **2005**, 280, 20413-20	5.4	32
82	Dark adaptation of rod photoreceptors in normal subjects, and in patients with Stargardt disease and an ABCA4 mutation. *Investigative Ophthalmology and Visual Science*, **2004**, 45, 2447-56		16
81	Surrogate electroretinographic markers for assessing therapeutic efficacy in the retina. *Expert Review of Molecular Diagnostics*, **2004**, 4, 693-703	3.8	8
80	Control of late off-center cone bipolar cell differentiation and visual signaling by the homeobox gene Vsx1. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 1754-9	11.5	123
79	A randomized, placebo-controlled clinical trial of docosahexaenoic acid supplementation for X-linked retinitis pigmentosa. *American Journal of Ophthalmology*, **2004**, 137, 704-18	4.9	53
78	The phenotype of Leber congenital amaurosis in patients with AIPL1 mutations. *JAMA Ophthalmology*, **2004**, 122, 1029-37		94
77	Cone deactivation kinetics and GRK1/GRK7 expression in enhanced S cone syndrome caused by mutations in NR2E3. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 1268-74		32
76	Biological safety assessment of docosahexaenoic acid supplementation in a randomized clinical trial for X-linked retinitis pigmentosa. *JAMA Ophthalmology*, **2003**, 121, 1269-78		43
75	Variability of full-field electroretinogram responses in subjects without diffuse photoreceptor cell disease. *Ophthalmology*, **2003**, 110, 1159-63	7.3	44
74	Visual function in breast-fed term infants weaned to formula with or without long-chain polyunsaturates at 4 to 6 months: a randomized clinical trial. *Journal of Pediatrics*, **2003**, 142, 669-77	3.6	127
73	Cone and rod ERG phototransduction parameters in retinitis pigmentosa. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 3993-4000		21
72	Characterization of RP1L1, a highly polymorphic paralog of the retinitis pigmentosa 1 (RP1) gene. *Molecular Vision*, **2003**, 9, 129-37	2.3	25
71	A randomized controlled trial of long-chain polyunsaturated fatty acid supplementation of formula in term infants after weaning at 6 wk of age. *American Journal of Clinical Nutrition*, **2002**, 75, 570-80	7	136
70	Quantitative electroretinogram measures of phototransduction in cone and rod photoreceptors: normal aging, progression with disease, and test-retest variability. *JAMA Ophthalmology*, **2002**, 120, 1045-51		70
69	A double-blind placebo-controlled evaluation of the acute effects of sildenafil citrate (Viagra) on visual function in subjects with early-stage age-related macular degeneration. *American Journal of Ophthalmology*, **2002**, 133, 665-72	4.9	24
68	Mutations in the inosine monophosphate dehydrogenase 1 gene (IMPDH1) cause the RP10 form of autosomal dominant retinitis pigmentosa. *Human Molecular Genetics*, **2002**, 11, 559-68	5.6	151
67	A comprehensive mutation analysis of RP2 and RPGR in a North American cohort of families with X-linked retinitis pigmentosa. *American Journal of Human Genetics*, **2002**, 70, 1545-54	11	194

66	Essential fatty acids in visual and brain development. *Lipids*, **2001**, 36, 885-95	1.6	349
65	Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. *Human Mutation*, **2001**, 17, 42-51	4.7	234
64	Five novel RPGR mutations in families with X-linked retinitis pigmentosa. *Human Mutation*, **2001**, 17, 151	4.7	10
63	Deficiency of rds/peripherin causes photoreceptor death in mouse models of digenic and dominant retinitis pigmentosa. *Proceedings of the National Academy of Sciences of the United States of America*, **2001**, 98, 7718-23	11.5	62
62	Visual function in patients with cone-rod dystrophy (CRD) associated with mutations in the ABCA4(ABCR) gene. *Experimental Eye Research*, **2001**, 73, 877-86	3.7	55
61	The Southwest Eye Registry **2001**, 339-345		
60	Impaired synthesis of DHA in patients with X-linked retinitis pigmentosa. *Journal of Lipid Research*, **2001**, 42, 1395-1401	6.3	42
59	X-Linked Retinitis Pigmentosa: Current Status **2001**, 11-22		1
58	Impact of early dietary intake and blood lipid composition of long-chain polyunsaturated fatty acids on later visual development. *Journal of Pediatric Gastroenterology and Nutrition*, **2000**, 31, 540-53	2.8	102
57	Electroretinographic determination of human rod flash response in vivo. *Methods in Enzymology*, **2000**, 316, 202-23	1.7	9
56	Mutations in a new photoreceptor-pineal gene on 17p cause Leber congenital amaurosis. *Nature Genetics*, **2000**, 24, 79-83	36.3	231
55	Rom-1 is required for rod photoreceptor viability and the regulation of disk morphogenesis. *Nature Genetics*, **2000**, 25, 67-73	36.3	133
54	Mutations in NYX, encoding the leucine-rich proteoglycan nyctalopin, cause X-linked complete congenital stationary night blindness. *Nature Genetics*, **2000**, 26, 319-23	36.3	265
53	A randomized controlled trial of early dietary supply of long-chain polyunsaturated fatty acids and mental development in term infants. *Developmental Medicine and Child Neurology*, **2000**, 42, 174-81	3.3	458
52	Prevalence of AIPL1 mutations in inherited retinal degenerative disease. *Molecular Genetics and Metabolism*, **2000**, 70, 142-50	3.7	133
51	Effect of stimulus size on static visual fields in patients with retinitis pigmentosa. *Ophthalmology*, **2000**, 107, 1950-4	7.3	24
50	Visual phenotype in patients with Arg41Gln and Ala196+1bp mutations in the CRX gene. *Ophthalmic Genetics*, **2000**, 21, 89-99	1.2	20
49	Mutations in the RP1 gene causing autosomal dominant retinitis pigmentosa. *Human Molecular Genetics*, **1999**, 8, 2121-8	5.6	56

48	Fatty acid profile of buccal cheek cell phospholipids as an index for dietary intake of docosahexaenoic acid in preterm infants. *Lipids*, **1999**, 34, 337-42	1.6	35
47	The effect of sleep state on electroretinographic (ERG) activity during early human development. *Early Human Development*, **1999**, 55, 51-62	2.2	4
46	Yearly rates of rod and cone functional loss in retinitis pigmentosa and cone-rod dystrophy. *Ophthalmology*, **1999**, 106, 258-68	7.3	132
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