# Andrew R Webster

List of Publications by Year
in descending order



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|---|---|---|---|
| 145 papers | 6,817 citations | 100601 38 h-index | 93651 72 g-index |
| 149 all docs | 149 docs citations | 149 times ranked | 8071 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Characterization of Retinal Function Using Microperimetry-Derived Metrics in Both Adults and Children With RPGR-Associated Retinopathy. American Journal of Ophthalmology, 2022, 234, 81-90. | 1.7 | 8 |
| 2 | A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. Ophthalmic Genetics, 2022, 43, 110-115. | 0.5 | 2 |
| 3 | Collaborative Research and Development of a Novel, Patient-Centered Digital Platform (MyEyeSite) for Rare Inherited Retinal Disease Data: Acceptability and Feasibility Study. JMIR Formative Research, 2022, 6, e21341. | 0.7 | 2 |
| 4 | X-Linked Retinoschisis. Ophthalmology, 2022, 129, 542-551. | 2.5 | 19 |
| 5 | WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27. | 1.7 | 8 |
| 6 | Electrical responses from human retinal cone pathways associate with a common genetic polymorphism implicated in myopia. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, . | 3.3 | 8 |
| 7 | Variability of retinopathy consequent upon novel mutations in LAMA1. Ophthalmic Genetics, 2022, 43, 671-678. | 0.5 | 1 |
| 8 | Axial Length Distributions in Patients With Genetically Confirmed Inherited Retinal Diseases. , 2022, 63, 15. | | 6 |
| 9 | A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. Eye, 2021, 35, 1482-1489. | 1.1 | 5 |
| 10 | Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. Ophthalmology, 2021, 128, 952-955. | 2.5 | 8 |
| 11 | Sector Retinitis Pigmentosa: Extending the Molecular Genetics Basis and Elucidating the Natural History. American Journal of Ophthalmology, 2021, 221, 299-310. | 1.7 | 20 |
| 12 | 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 |
| 13 | Ceramide synthase TLCD3B is a novel gene associated with human recessive retinal dystrophy. Genetics in Medicine, 2021, 23, 488-497. | 1.1 | 7 |
| 14 | Autosomal Recessive Bestrophinopathy. Ophthalmology, 2021, 128, 706-718. | 2.5 | 31 |
| 15 | New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176. | 1.1 | 7 |
| 16 | KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€"KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11. | 1.7 | 11 |
| 17 | KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107. | 1.7 | 17 |
| 18 | CNGB1 â€related rodâ€cone dystrophy: A mutation review and update. Human Mutation, 2021, 42, 641-666. | 1.1 | 16 |

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| 19 | Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53. | 1.7 | 8 |
| 20 | Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. Ophthalmic Genetics, 2021, 42, 664-673. | 0.5 | 14 |
| 21 | Prevalence of electronegative electroretinograms in a healthy adult cohort. BMJ Open Ophthalmology, 2021, 6, e000751. | 0.8 | 3 |
| 22 | Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607. | 1.6 | 37 |
| 23 | 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880. | 13.9 | 352 |
| 24 | Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. Eye, 2020, 34, 783-784. | 1.1 | 2 |
| 25 | A new paradigm for delivering personalised care: integrating genetics with surgical interventions in BEST1 mutations. Eye, 2020, 34, 577-583. | 1.1 | 11 |
| 26 | 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 |
| 27 | Transcorneal Electrical Stimulation for the Treatment of Retinitis Pigmentosa: A Multicenter Safety Study of the OkuStimÂ® System (TESOLA-Study). Ophthalmic Research, 2020, 63, 234-243. | 1.0 | 18 |
| 28 | Reanalysis of Association of Pro50Leu Substitution in Guanylate Cyclase Activating Protein-1 With Dominant Retinal Dystrophy. JAMA Ophthalmology, 2020, 138, 200. | 1.4 | 5 |
| 29 | Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427. | 2.3 | 55 |
| 30 | 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 |
| 31 | A genetic and clinical study of individuals with nonsyndromic retinopathy consequent upon sequence variants in <scp>HGSNAT</scp>, the gene associated with Sanfilippo C mucopolysaccharidosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 631-643. | 0.7 | 12 |
| 32 | Long-term follow-up of a case of posterior microphthalmos (PRSS56) with hyperautofluorescent retinal pigment epithelial deposits. European Journal of Ophthalmology, 2020, , 112067212094975. | 0.7 | 2 |
| 33 | Macula-predominant retinopathy associated with biallelic variants in RDH12. Ophthalmic Genetics, 2020, 41, 612-615. | 0.5 | 12 |
| 34 | 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 |
| 35 | Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102. | 13.7 | 338 |
| 36 | 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. | 1.1 | 15 |

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|---|---|---|---|
| 37 | Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. Human Molecular Genetics, 2020, 29, 1882-1899. | 1.4 | 24 |
| 38 | The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in MFRP and PRSS56. Scientific Reports, 2020, 10, 1289. | 1.6 | 24 |
| 39 | Characterisation of microvascular abnormalities using OCT angiography in patients with biallelic variants in USH2A and MYO7A. British Journal of Ophthalmology, 2020, 104, 480-486. | 2.1 | 12 |
| 40 | 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 |
| 41 | Monozygotic twins discordant for asymmetric pigmented paravenous chorioretinal atrophy. Retinal Cases and Brief Reports, 2020, Publish Ahead of Print, . | 0.3 | 4 |
| 42 | SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383. | 2.8 | 41 |
| 43 | Missense variants in the conserved transmembrane M2 protein domain of KCNJ13 associated with retinovascular changes in humans and zebrafish. Experimental Eye Research, 2019, 189, 107852. | 1.2 | 13 |
| 44 | Benefit of an electronic headâ€mounted low vision aid. Ophthalmic and Physiological Optics, 2019, 39, 422-431. | 1.0 | 23 |
| 45 | ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. Genetics in Medicine, 2019, 21, 1761-1771. | 1.1 | 111 |
| 46 | Nonsense-mediated mRNA decay efficiency varies in choroideremia providing a target to boost small molecule therapeutics. Human Molecular Genetics, 2019, 28, 1865-1871. | 1.4 | 25 |
| 47 | Unique noncoding variants upstream of PRDM13 are associated with a spectrum of developmental retinal dystrophies including progressive bifocal chorioretinal atrophy. Human Mutation, 2019, 40, 578-587. | 1.1 | 19 |
| 48 | Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671. | 0.7 | 10 |
| 49 | Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, . | 6.0 | 178 |
| 50 | Isolated rod dysfunction associated with a novel genotype of CNGB1. American Journal of Ophthalmology Case Reports, 2019, 14, 83-86. | 0.4 | 9 |
| 51 | 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 |
| 52 | Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98. | 1.7 | 20 |
| 53 | Rod-cone dystrophy associated with the Gly167Asp variant in PRPH2. Ophthalmic Genetics, 2019, 40, 188-189. | 0.5 | 6 |
| 54 | Phagosomal and mitochondrial alterations in RPE may contribute to KCNJ13 retinopathy. Scientific Reports, 2019, 9, 3793. | 1.6 | 29 |

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| 55 | Detailed clinical characterisation, unique features and natural history of autosomal recessive RDH12-associated retinal degeneration. British Journal of Ophthalmology, 2019, 103, bjophthalmol-2018-313580. | 2.1 | 20 |
| 56 | Macular maldevelopment inATF6-mediated retinal dysfunction. Ophthalmic Genetics, 2019, 40, 564-569. | 0.5 | 3 |
| 57 | Deep Phenotyping ofPDE6C-Associated Achromatopsia. , 2019, 60, 5112. | | 44 |
| 58 | Unilateral pigmentary retinopathy: a retrospective case series. Acta Ophthalmologica, 2019, 97, e601-e617. | 0.6 | 17 |
| 59 | Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. Genetics in Medicine, 2019, 21, 1751-1760. | 1.1 | 147 |
| 60 | Retinopathy Associated with Biallelic Mutations in PYGM (McArdle Disease). Ophthalmology, 2019, 126, 320-322. | 2.5 | 12 |
| 61 | Prevalence of cystoid macular oedema, epiretinal membrane and cataract in retinitis pigmentosa. British Journal of Ophthalmology, 2019, 103, 1163-1166. | 2.1 | 55 |
| 62 | CELLULAR IMAGING OF THE TAPETAL-LIKE REFLEX IN CARRIERS OF RPGR-ASSOCIATED RETINOPATHY. Retina, 2019, 39, 570-580. | 1.0 | 25 |
| 63 | PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY. Retina, 2019, 39, 514-529. | 1.0 | 23 |
| 64 | A clinical and molecular characterisation of CRB1-associated maculopathy. European Journal of Human Genetics, 2018, 26, 687-694. | 1.4 | 51 |
| 65 | 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 |
| 66 | DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. Retina, 2018, 38, 620-628. | 1.0 | 13 |
| 67 | NORMAL ELECTROOCULOGRAPHY IN BEST DISEASE AND AUTOSOMAL RECESSIVE BESTROPHINOPATHY. Retina, 2018, 38, 379-386. | 1.0 | 16 |
| 68 | 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 |
| 69 | Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91. | 1.1 | 23 |
| 70 | QUANTITATIVE ANALYSIS OF HYPERAUTOFLUORESCENT RINGS TO CHARACTERIZE THE NATURAL HISTORY AND PROGRESSION IN RPGR-ASSOCIATED RETINOPATHY. Retina, 2018, 38, 2401-2414. | 1.0 | 33 |
| 71 | Factors associated with visual acuity in patients with cystoid macular oedema and Retinitis Pigmentosa. Ophthalmic Epidemiology, 2018, 25, 183-186. | 0.8 | 8 |
| 72 | Whole genome sequencing reveals novel mutations causing autosomal dominant inherited macular degeneration. Ophthalmic Genetics, 2018, 39, 763-770. | 0.5 | 13 |

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| 73 | Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95. | 3.6 | 111 |
| 74 | Clinical Features of a Retinopathy Associated With a Dominant Allele of the RGR Gene. , 2018, 59, 4812. | | 9 |
| 75 | Retinal findings in a patient with mutations in ABCC6 and ABCA4. Eye, 2018, 32, 1542-1543. | 1.1 | 5 |
| 76 | Identification and characterization of the VAX2 p.Leu139Arg variant: possible involvement of VAX2 in cone dystrophy. Ophthalmic Genetics, 2018, 39, 539-543. | 0.5 | 1 |
| 77 | The treatment of refractory angle-closure glaucoma in a patient with X-linked juvenile retinoschisis. Ophthalmic Genetics, 2018, 39, 625-627. | 0.5 | 6 |
| 78 | Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612. | 1.1 | 6 |
| 79 | Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137. | 1.4 | 23 |
| 80 | Mechanism and evidence of nonsense suppression therapy for genetic eye disorders. Experimental Eye Research, 2017, 155, 24-37. | 1.2 | 40 |
| 81 | 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 |
| 82 | 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 |
| 83 | Measurement and Reproducibility of Preserved Ellipsoid Zone Area and Preserved Retinal Pigment Epithelium Area in Eyes With Choroideremia. American Journal of Ophthalmology, 2017, 179, 110-117. | 1.7 | 51 |
| 84 | 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. | 0.5 | 2 |
| 85 | Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423. | 1.8 | 40 |
| 86 | Single-base substitutions in theCHMpromoter as a cause of choroideremia. Human Mutation, 2017, 38, 704-715. | 1.1 | 45 |
| 87 | Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339. | 1.4 | 43 |
| 88 | 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 |
| 89 | Rescue of the MERTK phagocytic defect in a human iPSC disease model using translational read-through inducing drugs. Scientific Reports, 2017, 7, 51. | 1.6 | 55 |
| 90 | Childhood-onset Leber hereditary optic neuropathy. British Journal of Ophthalmology, 2017, 101, 1505-1509. | 2.1 | 62 |

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|---|---|---|---|
| 91 | Benign Yellow Dot Maculopathy. Ophthalmology, 2017, 124, 1004-1013. | 2.5 | 12 |
| 92 | 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 |
| 93 | Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512. | 1.6 | 23 |
| 94 | Association of C-Reactive Protein Genetic Polymorphisms With Late Age-Related Macular Degeneration. JAMA Ophthalmology, 2017, 135, 909. | 1.4 | 13 |
| 95 | Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906. | | 35 |
| 96 | Psychophysical measures of visual function and everyday perceptual experience in a case of congenital stationary night blindness. Clinical Ophthalmology, 2016, Volume 10, 1593-1606. | 0.9 | 3 |
| 97 | 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 |
| 98 | Phenotype and Progression of Retinal Degeneration Associated With Nullizigosity of ABCA4. , 2016, 57, 4668. | | 40 |
| 99 | Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180. | | 21 |
| 100 | The Effect on Retinal Structure and Function of 15 Specific ABCA4 Mutations: A Detailed Examination of 82 Hemizygous Patients. , 2016, 57, 5963. | | 41 |
| 101 | Preserved visual function in retinal dystrophy due to hypomorphicRPE65mutations. British Journal of Ophthalmology, 2016, 100, 1499-1505. | 2.1 | 25 |
| 102 | Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2. | 2.5 | 29 |
| 103 | Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626. | 2.5 | 19 |
| 104 | Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315. | 2.6 | 121 |
| 105 | Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049. | 1.4 | 29 |
| 106 | Mislocalisation of BEST1 in iPSC-derived retinal pigment epithelial cells from a family with autosomal dominant vitreoretinochoroidopathy (ADVIRC). Scientific Reports, 2016, 6, 33792. | 1.6 | 25 |
| 107 | Functional rescue of REP1 following treatment with PTC124 and novel derivative PTC-414 in human choroideremia fibroblasts and the nonsense-mediated zebrafish model. Human Molecular Genetics, 2016, 25, 3416-3431. | 1.4 | 69 |
| 108 | Investigation of SLA4A3 as a candidate gene for human retinal disease. Journal of Negative Results in BioMedicine, 2016, 15, 11. | 1.4 | 1 |

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| 109 | The clinical features of retinal disease due to a dominant mutation in RPE65. Molecular Vision, 2016, 22, 626-35. | 1.1 | 32 |
| 110 | Retinal Architecture in â€‹RGS9- and â€‹R9AP-Associated Retinal Dysfunction (Bradyopsia). American Journal of Ophthalmology, 2015, 160, 1269-1275.e1. | 1.7 | 15 |
| 111 | Exome sequencing revealsADAM9mutations in a child with cone-rod dystrophy. Acta Ophthalmologica, 2015, 93, e392-e393. | 0.6 | 3 |
| 112 | Somatic mosaicism of a novel IKBKG mutation in a male patient with incontinentia pigmenti. American Journal of Medical Genetics, Part A, 2015, 167, 1601-1604. | 0.7 | 27 |
| 113 | Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083. | | 13 |
| 114 | Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation ofRBP3Is Associated With High Myopia and Retinal Dystrophy. , 2015, 56, 2358. | | 42 |
| 115 | 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 |
| 116 | Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia. Nature Genetics, 2015, 47, 757-765. | 9.4 | 183 |
| 117 | Clinical Heterogeneity in a Family With Mutations inUSH2A. JAMA Ophthalmology, 2015, 133, 352. | 1.4 | 14 |
| 118 | Predominantly Cone-System Dysfunction as Rare Form of Retinal Degeneration in Patients With Molecularly Confirmed Bardet-Biedl Syndrome. American Journal of Ophthalmology, 2015, 160, 364-372.e1. | 1.7 | 26 |
| 119 | A detailed clinical and molecular survey of subjects with nonsyndromic USH2A retinopathy reveals an allelic hierarchy of disease-causing variants. European Journal of Human Genetics, 2015, 23, 1318-1327. | 1.4 | 89 |
| 120 | Author reply. Ophthalmology, 2015, 122, e22. | 2.5 | 0 |
| 121 | Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954. | 2.6 | 42 |
| 122 | Genotype-Dependent Variability in Residual Cone Structure in Achromatopsia: Toward Developing Metrics for Assessing Cone Health. , 2014, 55, 7303. | | 67 |
| 123 | Dark-Adaptation Functions in Molecularly Confirmed Achromatopsia and the Implications for Assessment in Retinal Therapy Trials. , 2014, 55, 6340. | | 14 |
| 124 | Clinical utility gene card for: Choroideremia. European Journal of Human Genetics, 2014, 22, 572-572. | 1.4 | 37 |
| 125 | Differential Light-induced Responses in Sectorial Inherited Retinal Degeneration. Journal of Biological Chemistry, 2014, 289, 35918-35928. | 1.6 | 32 |
| 126 | A Prospective Longitudinal Study of Retinal Structure and Function in Achromatopsia. , 2014, 55, 5733. | | 68 |

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|---|---|---|---|
| 127 | 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 |
| 128 | Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996. | 1.4 | 15 |
| 129 | Detailed Phenotypic and Genotypic Characterization of Bietti Crystalline Dystrophy. Ophthalmology, 2014, 121, 1174-1184. | 2.5 | 79 |
| 130 | Retinal Structure and Function in Achromatopsia. Ophthalmology, 2014, 121, 234-245. | 2.5 | 145 |
| 131 | Retinal gene therapy in patients with choroideremia: initial findings from a phase 1/2 clinical trial. Lancet, The, 2014, 383, 1129-1137. | 6.3 | 689 |
| 132 | The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in CRX, With Report of a Novel Macular Dystrophy Phenotype. Investigative Ophthalmology and Visual Science, 2014, 55, 6934-6944. | 3.3 | 59 |
| 133 | The effect of the common c.2299delG mutation in USH2A on RNA splicing. Experimental Eye Research, 2014, 122, 9-12. | 1.2 | 23 |
| 134 | Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769. | 2.6 | 67 |
| 135 | Natural History and Retinal Structure in Patients with Usher Syndrome Type 1 Owing to MYO7A Mutation. Ophthalmology, 2014, 121, 580-587. | 2.5 | 27 |
| 136 | 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 |
| 137 | Phenotypic findings in C1QTNF5 retinopathy (lateâ€onset retinal degeneration). Acta Ophthalmologica, 2013, 91, e191-5. | 0.6 | 35 |
| 138 | Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel LCA5 Mutations and New Genotype-Phenotype Correlations. Human Mutation, 2013, 34, 1537-1546. | 1.1 | 32 |
| 139 | Comprehensive sequence analysis of nine Usher syndrome genes in the UK National Collaborative Usher Study. Journal of Medical Genetics, 2012, 49, 27-36. | 1.5 | 152 |
| 140 | SERIAL IMAGING AND STRUCTURE-FUNCTION CORRELATES OF HIGH-DENSITY RINGS OF FUNDUS AUTOFLUORESCENCE IN RETINITIS PIGMENTOSA. Retina, 2011, 31, 1670-1679. | 1.0 | 79 |
| 141 | Autosomal dominant Best disease with an unusual electrooculographic light rise and risk of angle-closure glaucoma: a clinical and molecular genetic study. Molecular Vision, 2011, 17, 2272-82. | 1.1 | 16 |
| 142 | RDH12 retinopathy: novel mutations and phenotypic description. Molecular Vision, 2011, 17, 2706-16. | 1.1 | 47 |
| 143 | Biallelic Mutation of BEST1 Causes a Distinct Retinopathy in Humans. American Journal of Human Genetics, 2008, 82, 19-31. | 2.6 | 257 |
| 144 | Infrared Fundus Photography in a Case of Acute Macular Neuroretinopathy. Neuro-Ophthalmology, 2008, 32, 200-202. | 0.4 | 2 |

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|---|---|---|---|
| 145 | Phenotypic Variation in Enhanced S-cone Syndrome. , 2008, 49, 2082. | | 107 |