

Gavin Arno

List of Publications by Year in descending order

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Version: 2024-02-01

86
papers

3,663
citations

172207

29
h-index

161609

54
g-index

89
all docs

89
docs citations

89
times ranked

5290
citing authors

#	ARTICLE	IF	CITATIONS
1	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care – Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	13.9	352
2	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	2.6	343
3	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	13.7	338
4	Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. <i>Genetics in Medicine</i> , 2019, 21, 1751-1760.	1.1	147
5	Genetic Basis of Inherited Retinal Disease in a Molecularly Characterized Cohort of More Than 3000 Families from the United Kingdom. <i>Ophthalmology</i> , 2020, 127, 1384-1394.	2.5	131
6	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2016, 99, 1305-1315.	2.6	121
7	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. <i>Genome Medicine</i> , 2018, 10, 95.	3.6	111
8	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. <i>Genetics in Medicine</i> , 2019, 21, 1761-1771.	1.1	111
9	The Spectrum of PAX6 Mutations and Genotype-Phenotype Correlations in the Eye. <i>Genes</i> , 2019, 10, 1050.	1.0	111
10	Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon syndromes. <i>Journal of Medical Genetics</i> , 2015, 52, 85-94.	1.5	91
11	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2020, 107, 802-814.	2.6	75
12	Biallelic Variants in TLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 760-769.	2.6	67
13	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. <i>Progress in Retinal and Eye Research</i> , 2021, 82, 100898.	7.3	65
14	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. <i>American Journal of Human Genetics</i> , 2017, 100, 592-604.	2.6	61
15	Detailed Clinical Phenotype and Molecular Genetic Findings in <i>CLN3</i> -Associated Isolated Retinal Degeneration. <i>JAMA Ophthalmology</i> , 2017, 135, 749.	1.4	61
16	Mutations in TUBGCP4 Alter Microtubule Organization via the β -Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. <i>American Journal of Human Genetics</i> , 2015, 96, 666-674.	2.6	60
17	The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in <i>CRX</i> , With Report of a Novel Macular Dystrophy Phenotype. <i>Investigative Ophthalmology and Visual Science</i> , 2014, 55, 6934-6944.	3.3	59
18	Assessment of the incorporation of CNV surveillance into gene panel next-generation sequencing testing for inherited retinal diseases. <i>Journal of Medical Genetics</i> , 2018, 55, 114-121.	1.5	57

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19	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. <i>Molecular Therapy - Nucleic Acids</i> , 2020, 21, 412-427.	2.3	55
20	A clinical and molecular characterisation of CRB1-associated maculopathy. <i>European Journal of Human Genetics</i> , 2018, 26, 687-694.	1.4	51
21	Single-base substitutions in the <i>CHM</i> promoter as a cause of choroideremia. <i>Human Mutation</i> , 2017, 38, 704-715.	1.1	45
22	Association of Steroid 5 α -Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. <i>JAMA Ophthalmology</i> , 2017, 135, 339.	1.4	43
23	Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation of <i>RBP3</i> Associated With High Myopia and Retinal Dystrophy. , 2015, 56, 2358.		42
24	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. <i>Genes</i> , 2020, 11, 460.	1.0	42
25	<i>SSBP1</i> mutations in dominant optic atrophy with variable retinal degeneration. <i>Annals of Neurology</i> , 2019, 86, 368-383.	2.8	41
26	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. <i>Bioinformatics</i> , 2017, 33, 2421-2423.	1.8	40
27	Clinical and Molecular Characterization of Enhanced S-Cone Syndrome in Children. <i>JAMA Ophthalmology</i> , 2014, 132, 1341.	1.4	39
28	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. <i>American Journal of Ophthalmology</i> , 2020, 210, 59-70.	1.7	39
29	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. <i>Genetics in Medicine</i> , 2020, 22, 2041-2051.	1.1	38
30	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. <i>Scientific Reports</i> , 2021, 11, 20607.	1.6	37
31	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. <i>Ophthalmology</i> , 2016, 123, 668-671.e2.	2.5	29
32	Expanding the Phenotype of <i>TRNT1</i> -Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. <i>JAMA Ophthalmology</i> , 2016, 134, 1049.	1.4	29
33	Clinical and Genetic Characteristics of East Asian Patients with Occult Macular Dystrophy (Miyake) Tj ETQq1 1 0.784314 rgBTJ/Overlock	2.5	28
34	Somatic mosaicism of a novel <i>IKBKKG</i> mutation in a male patient with incontinentia pigmenti. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1601-1604.	0.7	27
35	Enhanced S-Cone Syndrome. <i>Ophthalmology Retina</i> , 2021, 5, 195-214.	1.2	27
36	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26

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37	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of <i>RGR</i> With the Discovery of a Cis-Acting Mutation in <i>CDHR1</i> . , 2016, 57, 4806.		25
38	Mutation in the intracellular chloride channel <i>CLCC1</i> associated with autosomal recessive retinitis pigmentosa. <i>PLoS Genetics</i> , 2018, 14, e1007504.	1.5	25
39	Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with <i>CRX</i> -associated retinal disorder: Identification of Genotype-phenotype Association. <i>Scientific Reports</i> , 2020, 10, 9531.	1.6	24
40	The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in <i>MFRP</i> and <i>PRSS56</i> . <i>Scientific Reports</i> , 2020, 10, 1289.	1.6	24
41	Clinical Characterization of <i>CNGB1</i> -Related Autosomal Recessive Retinitis Pigmentosa. <i>JAMA Ophthalmology</i> , 2017, 135, 137.	1.4	23
42	Duplication events downstream of <i>IRX1</i> cause North Carolina macular dystrophy at the <i>MCDR3</i> locus. <i>Scientific Reports</i> , 2017, 7, 7512.	1.6	23
43	Missense variants in the X-linked gene <i>PRPS1</i> cause retinal degeneration in females. <i>Human Mutation</i> , 2018, 39, 80-91.	1.1	23
44	Mutations in <i>AGBL5</i> , Encoding β -Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
45	Missense mutations in the WD40 domain of <i>AHI1</i> cause non-syndromic retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2017, 54, 624-632.	1.5	21
46	Vitamin A deficiency due to bi-allelic mutation of <i>RBP4</i> : There's more to it than meets the eye. <i>Ophthalmic Genetics</i> , 2017, 38, 465-466.	0.5	21
47	Clinical and genetic characteristics of 10 Japanese patients with <i>PROM1</i> -associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 656-674.	0.7	21
48	Genetic Spectrum of <i>EYS</i> -associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency. <i>Scientific Reports</i> , 2020, 10, 5497.	1.6	21
49	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. <i>American Journal of Ophthalmology</i> , 2019, 207, 87-98.	1.7	20
50	Clinical and molecular findings in a cohort of 152 Brazilian severe early onset inherited retinal dystrophy patients. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 728-752.	0.7	20
51	Unique noncoding variants upstream of <i>PRDM13</i> are associated with a spectrum of developmental retinal dystrophies including progressive bifocal chorioretinal atrophy. <i>Human Mutation</i> , 2019, 40, 578-587.	1.1	19
52	Phenotypical Characteristics of <i>POC1B</i> -Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Fundusoscopic Appearance. , 2019, 60, 3432.		18
53	Practical guide to genetic screening for inherited eye diseases. <i>Therapeutic Advances in Ophthalmology</i> , 2020, 12, 251584142095459.	0.8	17
54	<i>KCNV2</i> -Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course— <i>KCNV2</i> Study Group Report 1. <i>American Journal of Ophthalmology</i> , 2021, 225, 95-107.	1.7	17

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55	Clinical and Genetic Characteristics of 15 Affected Patients From 12 Japanese Families with <i>GUCY2D</i> -Associated Retinal Disorder. <i>Translational Vision Science and Technology</i> , 2020, 9, 2.	1.1	15
56	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. <i>Retina</i> , 2018, 38, 620-628.	1.0	13
57	A recurrent splice-site mutation in <i>EPHA2</i> causing congenital posterior nuclear cataract. <i>Ophthalmic Genetics</i> , 2018, 39, 236-241.	0.5	13
58	Whole genome sequencing reveals novel mutations causing autosomal dominant inherited macular degeneration. <i>Ophthalmic Genetics</i> , 2018, 39, 763-770.	0.5	13
59	Benign Yellow Dot Maculopathy. <i>Ophthalmology</i> , 2017, 124, 1004-1013.	2.5	12
60	A genetic and clinical study of individuals with nonsyndromic retinopathy consequent upon sequence variants in <i>HGSNAT</i> , the gene associated with Sanfilippo C mucopolysaccharidosis. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 631-643.	0.7	12
61	Macula-predominant retinopathy associated with biallelic variants in <i>RDH12</i> . <i>Ophthalmic Genetics</i> , 2020, 41, 612-615.	0.5	12
62	<i>KCNV2</i> -Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpoints— <i>KCNV2</i> Study Group Report 2. <i>American Journal of Ophthalmology</i> , 2021, 230, 1-11.	1.7	11
63	Delineating the expanding phenotype associated with <i>SCAPER</i> gene mutation. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1665-1671.	0.7	10
64	Spatial Functional Characteristics of East Asian Patients With Occult Macular Dystrophy (Miyake) <i>Investigative Ophthalmology and Visual Science</i> , 2020, 61, 10.	1.7	10
65	Clinical Features of a Retinopathy Associated With a Dominant Allele of the <i>RGR</i> Gene. <i>Investigative Ophthalmology and Visual Science</i> , 2018, 59, 4812.		9
66	Isolated rod dysfunction associated with a novel genotype of <i>CNGB1</i> . <i>American Journal of Ophthalmology Case Reports</i> , 2019, 14, 83-86.	0.4	9
67	Expanding the phenotypic spectrum consequent upon de novo <i>WDR37</i> missense variants. <i>Clinical Genetics</i> , 2020, 98, 191-197.	1.0	8
68	Clinical and Genetic Findings in <i>CTNNA1</i> -Associated Macular Pattern Dystrophy. <i>Ophthalmology</i> , 2021, 128, 952-955.	2.5	8
69	Broadening <i>INPP5E</i> phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. <i>Npj Genomic Medicine</i> , 2021, 6, 53.	1.7	8
70	<i>WFS1</i> -Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. <i>American Journal of Ophthalmology</i> , 2022, 241, 9-27.	1.7	8
71	Ceramide synthase <i>TLCD3B</i> is a novel gene associated with human recessive retinal dystrophy. <i>Genetics in Medicine</i> , 2021, 23, 488-497.	1.1	7
72	Phenogenon: Gene to phenotype associations for rare genetic diseases. <i>PLoS ONE</i> , 2020, 15, e0230587.	1.1	6

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73	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. <i>Molecular Vision</i> , 2018, 24, 603-612.	1.1	6
74	Reanalysis of Association of Pro50Leu Substitution in Guanylate Cyclase Activating Protein-1 With Dominant Retinal Dystrophy. <i>JAMA Ophthalmology</i> , 2020, 138, 200.	1.4	5
75	A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. <i>Eye</i> , 2021, 35, 1482-1489.	1.1	5
76	RP2 -associated retinal disorder in a Japanese cohort: Report of novel variants and a literature review, identifying a genotype-phenotype association. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 675-693.	0.7	5
77	Identification of autosomal recessive novel genes and retinal phenotypes in members of the solute carrier (SLC) superfamily. <i>Genetics in Medicine</i> , 2022, 24, 1523-1535.	1.1	5
78	Exome sequencing reveals ADAM9 mutations in a child with cone-rod dystrophy. <i>Acta Ophthalmologica</i> , 2015, 93, e392-e393.	0.6	3
79	Macular maldevelopment in <i>ATF6</i> -mediated retinal dysfunction. <i>Ophthalmic Genetics</i> , 2019, 40, 564-569.	0.5	3
80	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. <i>Ophthalmic Genetics</i> , 2017, 38, 511-519.	0.5	2
81	Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. <i>Eye</i> , 2020, 34, 783-784.	1.1	2
82	Ocular genetics in the genomics age. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 860-868.	0.7	2
83	A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. <i>Ophthalmic Genetics</i> , 2022, 43, 110-115.	0.5	2
84	Introduction to the special issue on <i>Ophthalmic Genetics: Vision in 2020</i> . <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 535-537.	0.7	1
85	Genome Analysis for Inherited Retinal Disease: The State of the Art. <i>Essentials in Ophthalmology</i> , 2021, , 153-168.	0.0	1
86	Variability of retinopathy consequent upon novel mutations in LAMA1. <i>Ophthalmic Genetics</i> , 2022, 43, 671-678.	0.5	1