Gavin Arno

List of Publications by Year
in descending order

86 papers	3,663 citations	172457 29 h-index	161849 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 â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
2	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.	6.2	343
3	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
4	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.	2.4	147
5	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.	5.2	131
6	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
7	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	8.2	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. Genetics in Medicine, 2019, 21, 1761-1771.	2.4	111
9	The Spectrum of PAX6 Mutations and Genotype-Phenotype Correlations in the Eye. Genes, 2019, 10, 1050.	2.4	111
10	Neuropathy target esterase impairments cause Oliverâ€"McFarlane and Laurenceâ€"Moon syndromes. Journal of Medical Genetics, 2015, 52, 85-94.	3.2	91
11	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.	6.2	75
12	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
13	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. Progress in Retinal and Eye Research, 2021, 82, 100898.	15.5	65
14	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
15	Detailed Clinical Phenotype and Molecular Genetic Findings in CLN3-Associated Isolated Retinal Degeneration. JAMA Ophthalmology, 2017, 135, 749.	2.5	61
16	Mutations in TUBGCP4 Alter Microtubule Organization via the Î³-Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. American Journal of Human Genetics, 2015, 96, 666-674.	6.2	60
17	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
18	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.	3.2	57

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19	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	5.1	55
20	A clinical and molecular characterisation of CRB1-associated maculopathy. European Journal of Human Genetics, 2018, 26, 687-694.	2.8	51
21	Single-base substitutions in theCHMpromoter as a cause of choroideremia. Human Mutation, 2017, 38, 704-715.	2.5	45
22	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	2.5	43
23	Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation ofRBP3Is 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. Genes, 2020, 11, 460.	2.4	42
25	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	5.3	41
26	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
27	Clinical and Molecular Characterization of Enhanced S-Cone Syndrome in Children. JAMA Ophthalmology, 2014, 132, 1341.	2.5	39
28	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.	3.3	39
29	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. Genetics in Medicine, 2020, 22, 2041-2051.	2.4	38
30	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	3.3	37
31	Mutations in CACNA2D4 Cause Distinctive Retinal Dysfunction in Humans. Ophthalmology, 2016, 123, 668-671.e2.	5.2	29
32	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	2.5	29
33	Clinical and Genetic Characteristics of East Asian Patients with Occult Macular Dystrophy (Miyake) Tj ETQq1 1 0.784314 rgBT /Overlo	5.2	28
34	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.	1.2	27
35	Enhanced S-Cone Syndrome. Ophthalmology Retina, 2021, 5, 195-214.	2.4	27
36	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.	6.2	26

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37	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
38	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
39	Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association. Scientific Reports, 2020, 10, 9531.	3.3	24
40	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.	3.3	24
41	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	2.5	23
42	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512.	3.3	23
43	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
44	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
45	Missense mutations in the WD40 domain ofAHI1cause non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2017, 54, 624-632.	3.2	21
46	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.	1.2	21
47	Clinical and genetic characteristics of 10 Japanese patients with PROM1 â€associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 656-674.	1.6	21
48	Genetic Spectrum of EYS-associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency. Scientific Reports, 2020, 10, 5497.	3.3	21
49	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	3.3	20
50	Clinical and molecular findings in a cohort of 152 Brazilian severe early onset inherited retinal dystrophy patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 728-752.	1.6	20
51	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.	2.5	19
52	Phenotypical Characteristics ofPOC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance. , 2019, 60, 3432.		18
53	Practical guide to genetic screening for inherited eye diseases. Therapeutic Advances in Ophthalmology, 2020, 12, 251584142095459.	1.4	17
54	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€”KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	3.3	17

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55	Clinical and Genetic Characteristics of 15 Affected Patients From 12 Japanese Families with GUCY2D-Associated Retinal Disorder. Translational Vision Science and Technology, 2020, 9, 2.	2.2	15
56	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. Retina, 2018, 38, 620-628.	1.7	13
57	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. Ophthalmic Genetics, 2018, 39, 236-241.	1.2	13
58	Whole genome sequencing reveals novel mutations causing autosomal dominant inherited macular degeneration. Ophthalmic Genetics, 2018, 39, 763-770.	1.2	13
59	Benign Yellow Dot Maculopathy. Ophthalmology, 2017, 124, 1004-1013.	5.2	12
60	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.	1.6	12
61	Macula-predominant retinopathy associated with biallelic variants in RDH12. Ophthalmic Genetics, 2020, 41, 612-615.	1.2	12
62	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€"KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11.	3.3	11
63	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
64	Spatial Functional Characteristics of East Asian Patients With Occult Macular Dystrophy (Miyake) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	3.3	10
65	Clinical Features of a Retinopathy Associated With a Dominant Allele of the RGR Gene. , 2018, 59, 4812.		9
66	Isolated rod dysfunction associated with a novel genotype of CNGB1. American Journal of Ophthalmology Case Reports, 2019, 14, 83-86.	0.7	9
67	Expanding the phenotypic spectrum consequent upon de novo <scp>WDR37</scp> missense variants. Clinical Genetics, 2020, 98, 191-197.	2.0	8
68	Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. Ophthalmology, 2021, 128, 952-955.	5.2	8
69	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
70	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	3.3	8
71	Ceramide synthase TLCD3B is a novel gene associated with human recessive retinal dystrophy. Genetics in Medicine, 2021, 23, 488-497.	2.4	7
72	Phenogenon: Gene to phenotype associations for rare genetic diseases. PLoS ONE, 2020, 15, e0230587.	2.5	6

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73	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6
74	Reanalysis of Association of Pro50Leu Substitution in Guanylate Cyclase Activating Protein-1 With Dominant Retinal Dystrophy. JAMA Ophthalmology, 2020, 138, 200.	2.5	5
75	A clinical study of patients with novel CDHR1 genotypes associated with late-onset macular dystrophy. Eye, 2021, 35, 1482-1489.	2.1	5
76	RP2 â€associated retinal disorder in a Japanese cohort: Report of novel variants and a literature review, identifying a genotypeâ€“phenotype association. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 675-693.	1.6	5
77	Identification of autosomal recessive novel genes and retinal phenotypes in members of the solute carrier (SLC) superfamily. Genetics in Medicine, 2022, 24, 1523-1535.	2.4	5
78	Exome sequencing revealsADAM9mutations in a child with cone-rod dystrophy. Acta Ophthalmologica, 2015, 93, e392-e393.	1.1	3
79	Macular maldevelopment inATF6-mediated retinal dysfunction. Ophthalmic Genetics, 2019, 40, 564-569.	1.2	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. Ophthalmic Genetics, 2017, 38, 511-519.	1.2	2
81	Awareness of olfactory impairment in a cohort of patients with CNGB1-associated retinitis pigmentosa. Eye, 2020, 34, 783-784.	2.1	2
82	Ocular genetics in the genomics age. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 860-868.	1.6	2
83	A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. Ophthalmic Genetics, 2022, 43, 110-115.	1.2	2
84	Introduction to the special issue on Ophthalmic Genetics: Vision in 2020. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 535-537.	1.6	1
85	Genome Analysis for Inherited Retinal Disease: The State of the Art. Essentials in Ophthalmology, 2021, , 153-168.	0.1	1
86	Variability of retinopathy consequent upon novel mutations in LAMA1. Ophthalmic Genetics, 2022, 43, 671-678.	1.2	1