Brian P Brooks

List of Publications by Year
in descending order

55 papers	1,274 citations	361413 20 h-index	395702 33 g-index
57 all docs	57 docs citations	57 times ranked	2143 citing authors

#	Article	IF	Citations
1	Novel ophthalmic findings and deep phenotyping in Williams-Beuren syndrome. British Journal of Ophthalmology, 2023, 107, 1554-1559.	3.9	3
2	DDX58(RIG-I)-related disease is associated with tissue-specific interferon pathway activation. Journal of Medical Genetics, 2022, 59, 294-304.	3.2	16
3	InÂvitro disease modeling of oculocutaneous albinism type 1 and 2 using human induced pluripotent stem cell-derived retinal pigment epithelium. Stem Cell Reports, 2022, 17, 173-186.	4.8	12
4	Identification of 4 novel human ocular coloboma genes ANK3, BMPR1B, PDGFRA, and CDH4 through evolutionary conserved vertebrate gene analysis. Genetics in Medicine, 2022, 24, 1073-1084.	2.4	4
5	Genotypic and Phenotypic Spectrum of Foveal Hypoplasia. Ophthalmology, 2022, 129, 708-718.	5.2	29
6	Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	1
7	De novo frameshift mutation in YAP1 associated with bilateral uveal coloboma and microphthalmia. Ophthalmic Genetics, 2022, 43, 513-517.	1.2	4
8	ABCA4c.859-25A>G, a Frequent Palestinian Founder Mutation Affecting the Intron 7 Branchpoint, Is Associated With Early-Onset Stargardt Disease. , 2022, 63, 20.		3
9	Clinical Phenotypes of CDHR1-Associated Retinal Dystrophies. Genes, 2022, 13, 925.	2.4	3
10	Response to Finstererâ€™s â€œExclude hereditary and acquired differential disorders before attributing retinoschisis to Kears-Sayre syndromeâ€• Ophthalmic Genetics, 2021, 42, 100-100.	1.2	0
11	Clinical utility gene card for oculocutaneous (OCA) and ocular albinism (OA)â€”an update. European Journal of Human Genetics, 2021, 29, 1577-1583.	2.8	4
12	The evolving role of genetics in ophthalmology. Ophthalmic Genetics, 2021, 42, 110-113.	1.2	5
13	Gene Therapy of Dominant CRX-Leber Congenital Amaurosis using Patient Stem Cell-Derived Retinal Organoids. Stem Cell Reports, 2021, 16, 252-263.	4.8	53
14	Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. Ophthalmic Genetics, 2021, 42, 320-325.	1.2	2
15	Reply. Ophthalmology, 2021, 128, e214-e215.	5.2	0
16	In vivo assessment of neurodegeneration in Spinocerebellar Ataxia type 7. NeuroImage: Clinical, 2021, 29, 102561.	2.7	4
17	A Comprehensive Assessment of Co-occurring Birth Defects among Infants with Non-Syndromic Anophthalmia or Microphthalmia. Ophthalmic Epidemiology, 2021, 28, 428-435.	1.7	4
18	Review of evidence for environmental causes of uveal coloboma. Survey of Ophthalmology, 2021, , .	4.0	1

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19	Anophthalmia including next-generation sequencing-based approaches. European Journal of Human Genetics, 2020, 28, 388-398.	2.8	6
20	Highâ€throughput custom capture sequencing identifies novel mutations in colobomaâ€associated genes: Mutation in DNAâ€binding domain of retinoic acid receptor beta affects nuclear localization causing ocular coloboma. Human Mutation, 2020, 41, 678-695.	2.5	13
21	PDE6C: Novel Mutations, Atypical Phenotype, and Differences Among Children and Adults. , 2020, 61, 1.		8
22	Genetics of syndromic ocular coloboma: CHARGE and COACH syndromes. Experimental Eye Research, 2020, 193, 107940.	2.6	23
23	Retinoschisis associated with Kearns-Sayre syndrome. Ophthalmic Genetics, 2020, 41, 497-500.	1.2	7
24	Novel TMEM98, MFRP, PRSS56 variants in a large United States high hyperopia and nanophthalmos cohort. Scientific Reports, 2020, 10, 19986.	3.3	17
25	Ocular and Systemic Findings in Adults with Uveal Coloboma. Ophthalmology, 2020, 127, 1772-1774.	5.2	8
26	Atypical and ultra-rare Usher syndrome: a review. Ophthalmic Genetics, 2020, 41, 401-412.	1.2	20
27	A novel frameshift mutation in SOX10 causes Waardenburg syndrome with peripheral demyelinating neuropathy, visual impairment and the absence of Hirschsprung disease. American Journal of Medical Genetics, Part A, 2020, 182, 1278-1283.	1.2	7
28	Nolz1 expression is required in dopaminergic axon guidance and striatal innervation. Nature Communications, 2020, 11, 3111.	12.8	8
29	Brain phenotyping in Moebius syndrome and other congenital facial weakness disorders by diffusion MRI morphometry. Brain Communications, 2020, 2, fcaa014.	3.3	9
30	Survey of practice patterns for the management of ophthalmic genetic disorders among AAPOS members: report by the AAPOS Genetic Eye Disease Task Force. Journal of AAPOS, 2019, 23, 226-228.e1.	0.3	6
31	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180.	12.8	27
32	One-year pilot study on the effects of nitisinone on melanin in patients with OCA-1B. JCI Insight, 2019, 4, .	5.0	25
33	Minimal Efficacy of Nitisinone Treatment in a Novel Mouse Model of Oculocutaneous Albinism, Type 3. , 2018, 59, 4945.		10
34	Antisense oligonucleotides targeting mutant Ataxin-7 restore visual function in a mouse model of spinocerebellar ataxia type 7. Science Translational Medicine, 2018, 10, .	12.4	63
35	Identifying core biological processes distinguishing human eye tissues with precise systems-level gene expression analyses and weighted correlation networks. Human Molecular Genetics, 2018, 27, 3325-3339.	2.9	46
36	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. Ophthalmology, 2018, 125, 1937-1952.	5.2	43

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37	Clinical and molecular phenotyping of a child with Hermansky-Pudlak syndrome-7, an uncommon genetic type of HPS. Molecular Genetics and Metabolism, 2017, 120, 378-383.	1.1	25
38	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	2.4	100
39	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. American Journal of Medical Genetics, Part A, 2017, 173, 1796-1812.	1.2	26
40	Genetic background-dependent role of Egr1 for eyelid development. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E7131-E7139.	7.1	6
41	Cellular and molecular defects in a patient with Hermansky-Pudlak syndrome type 5. PLoS ONE, 2017, 12, e0173682.	2.5	11
42	Biallelic Mutations in MITF Cause Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, and Deafness. American Journal of Human Genetics, 2016, 99, 1388-1394.	6.2	74
43	Cystic cerebellar dysplasia and biallelicLAMA1mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. Journal of Medical Genetics, 2016, 53, 318-329.	3.2	25
44	Ophthalmic Manifestations and Long-Term Visual Outcomes in Patients with Cobalamin C Deficiency. Ophthalmology, 2016, 123, 571-582.	5.2	34
45	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391.	2.9	40
46	nlz1 is required for cilia formation in zebrafish embryogenesis. Developmental Biology, 2015, 406, 203-211.	2.0	8
47	aldh7a1 Regulates Eye and Limb Development in Zebrafish. PLoS ONE, 2014, 9, e101782.	2.5	20
48	Multiple A2E treatments lead to melanization of rod outer segment-challenged ARPE-19 cells. Molecular Vision, 2014, 20, 285-300.	1.1	24
49	Systemic Diagnostic Testing in Patients With Apparently Isolated Uveal Coloboma. American Journal of Ophthalmology, 2013, 156, 1159-1168.e4.	3.3	11
50	A broad range of ophthalmologic anomalies is part of the holoprosencephaly spectrum. American Journal of Medical Genetics, Part A, 2011, 155, 2713-2720.	1.2	35
51	Nitisinone improves eye and skin pigmentation defects in a mouse model of oculocutaneous albinism. Journal of Clinical Investigation, 2011, 121, 3914-3923.	8.2	45
52	Papillorenal Syndrome-Causing Missense Mutations in PAX2/Pax2 Result in Hypomorphic Alleles in Mouse and Human. PLoS Genetics, 2010, 6, e1000870.	3.5	21
53	Expression profiling during ocular development identifies 2 Nlz genes with a critical role in optic fissure closure. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 1462-1467.	7.1	67
54	Incomplete penetrance and phenotypic variability characterize Gdf6-attributable oculo-skeletal phenotypes. Human Molecular Genetics, 2009, 18, 1110-1121.	2.9	92

#	Article	IF	Citations
55	Uveal coloboma: clinical and basic science update. Current Opinion in Ophthalmology, 2006, 17, 447-470.	2.9	116