Jay E Self

List of Publications by Year
in descending order

40	678	623188	642321
papers	citations	14	23
		h-index	g-index

43	43	43	1182
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A severe case of Boschâ€“<scp>Boonstraâ€“Schaaf</scp> optic atrophy syndrome with a novel description of coloboma and septoâ€optic dysplasia, owing to a start codon variant in the <scp>NR2F1</scp> gene. American Journal of Medical Genetics, Part A, 2022, 188, 900-906.	0.7	5
2	Evidence that the Ser192Tyr/Arg402Gln in cis Tyrosinase gene haplotype is a disease-causing allele in oculocutaneous albinism type 1B (OCA1B). Npj Genomic Medicine, 2022, 7, 2.	1.7	9
3	Infantile nystagmus. , 2022, , 403-406.		0
4	Comparison of the handheld RETeval ERG system with a routine ERG system in healthy adults and in paediatric patients. Eye, 2021, 35, 2180-2189.	1.1	8
5	Disorders of vision in neonatal hypoxic-ischaemic encephalopathy: a systematic review. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2021, 106, 357-362.	1.4	6
6	Beyond Visual Acuity: Development of a Simple Test of the Slow-To-See Phenomenon in Children with Infantile Nystagmus Syndrome. Current Eye Research, 2021, 46, 263-270.	0.7	2
7	Novel therapeutics in nystagmus: what has the genetics taught us so far?. Therapeutic Advances in Rare Disease, 2021, 2, 263300402199871.	0.3	1
8	Cerebral visual impairmentâ€related vision problems in primary school children: a crossâ€sectional survey. Developmental Medicine and Child Neurology, 2021, 63, 683-689.	1.1	37
9	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
10	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609.	1.1	43
11	Characterization of the Frmd7 Knock-Out Mice Generated by the EUCOMM/COMP Repository as a Model for Idiopathic Infantile Nystagmus (IIN). Genes, 2020, 11, 1157.	1.0	4
12	Cataract management in children: a review of the literature and current practice across five large UK centres. Eye, 2020, 34, 2197-2218.	1.1	33
13	Management of nystagmus in children: a review of the literature and current practice in UK specialist services. Eye, 2020, 34, 1515-1534.	1.1	23
14	Evaluating the impact of information and support for people with nystagmus in the digital age: A patient and carer questionnaire study. Current Eye Research, 2020, 45, 713-717.	0.7	4
15	The potential and value of objective eye tracking in the ophthalmology clinic. Eye, 2019, 33, 1200-1202.	1.1	16
16	A small gene sequencing panel realises a high diagnostic rate in patients with congenital nystagmus following basic phenotyping. Scientific Reports, 2019, 9, 13229.	1.6	9
17	BBS5 and INPP5E mutations associated with ciliopathy disorders in families from Pakistan. Annals of Human Genetics, 2019, 83, 477-482.	0.3	10
18	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	0.7	10

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19	Tyrosinase (TYR) gene sequencing and literature review reveals recurrent mutations and multiple population founder gene mutations as causative of oculocutaneous albinism (OCA) in Pakistani families. Eye, 2019, 33, 1339-1346.	1.1	5
20	Oral levodopa rescues retinal morphology and visual function in a murine model of human albinism. Pigment Cell and Melanoma Research, 2019, 32, 657-671.	1.5	23
21	Reply. Journal of AAPOS, 2019, 23, 124.	0.2	0
22	Comprehensive sequencing of the myocilin gene in a selected cohort of severe primary open-angle glaucoma patients. Scientific Reports, 2019, 9, 3100.	1.6	8
23	Supranuclear eye movements and nystagmus in children: A review of the literature and guide to clinical examination, interpretation of findings and age-appropriate norms. Eye, 2019, 33, 261-273.	1.1	15
24	Mutations in TYR and OCA2 associated with oculocutaneous albinism in Pakistani families. Meta Gene, 2018, 17, 48-55.	0.3	9
25	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	1.5	59
26	Atropine Penalization Versus Occlusion Therapies for Unilateral Amblyopia after the Critical Period of Visual Development: A Systematic Review. Ophthalmology and Therapy, 2018, 7, 323-332.	1.0	9
27	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
28	Identification and functional analysis of a novel oculocerebrorenal syndrome of Lowe () gene variant in two pedigrees with varying phenotypes including isolated congenital cataract. Molecular Vision, 2018, 24, 847-852.	1.1	3
29	Comparison of mouse and human retinal morphology and function in albinism: potential implications for therapeutic development. Lancet, The, 2017, 389, S59.	6.3	0
30	A novel method for examining corneal endothelial cell morphology in infants. Journal of AAPOS, 2017, 21, 241-243.e1.	0.2	0
31	Identification of a functionally significant tri-allelic genotype in the Tyrosinase gene (TYR) causing hypomorphic oculocutaneous albinism (OCA1B). Scientific Reports, 2017, 7, 4415.	1.6	47
32	A mutation ofEPT1 (SELENOI)underlies a new disorder of Kennedy pathway phospholipid biosynthesis. Brain, 2017, 140, aww318.	3.7	58
33	Is an iris claw IOL a good option for correcting surgically induced aphakia in children? A review of the literature and illustrative case study. Eye, 2016, 30, 1155-1159.	1.1	17
34	Is This Actually CIN?. Pediatric Neurology, 2015, 53, e11.	1.0	1
35	Prevalence of myocilin gene mutations in a novel UK cohort of POAG patients. Eye, 2010, 24, 328-333.	1.1	18
36	A Review of the Molecular Genetics of Congenital Idiopathic Nystagmus (CIN). Ophthalmic Genetics, 2007, 28, 187-191.	0.5	39

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37	The Molecular Genetics of Congenital Idiopathic Nystagmus. Seminars in Ophthalmology, 2006, 21, 87-90.	0.8	14
38	Fine mapping of the X-linked recessive congenital idiopathic nystagmus locus at Xq24-q26.3. Molecular Vision, 2006, 12, 1211-6.	1.1	6
39	Association of HLA Class I and Class II Polymorphisms with Age-Related Macular Degeneration. , 2005, 46, 1726.		64
40	Reducing conditions significantly attenuate the neuroprotective efficacy of competitive, but not other NMDA receptor antagonists in vitro. European Journal of Neuroscience, 2000, 12, 3833-3842.	1.2	19