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	Association of HLA Class I and Class II Polymorphisms with Age-Related Macular Degeneration. , 2005, 46, 1726.		64
2	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
3	A mutation ofEPT1 (SELENOI)underlies a new disorder of Kennedy pathway phospholipid biosynthesis. Brain, 2017, 140, aww318.	3.7	58
4	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
5	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
6	A Review of the Molecular Genetics of Congenital Idiopathic Nystagmus (CIN). Ophthalmic Genetics, 2007, 28, 187-191.	0.5	39
7	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
8	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
9	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
10	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
11	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
12	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
13	Prevalence of myocilin gene mutations in a novel UK cohort of POAG patients. Eye, 2010, 24, 328-333.	1.1	18
14	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
15	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
16	The potential and value of objective eye tracking in the ophthalmology clinic. Eye, 2019, 33, 1200-1202.	1.1	16
17	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
18	The Molecular Genetics of Congenital Idiopathic Nystagmus. Seminars in Ophthalmology, 2006, 21, 87-90.	0.8	14

#	Article	IF	Citations
19	BBS5 and INPP5E mutations associated with ciliopathy disorders in families from Pakistan. Annals of Human Genetics, 2019, 83, 477-482.	0.3	10
20	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	0.7	10
21	Mutations in TYR and OCA2 associated with oculocutaneous albinism in Pakistani families. Meta Gene, 2018, 17, 48-55.	0.3	9
22	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
23	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
24	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
25	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
26	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
27	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
28	Fine mapping of the X-linked recessive congenital idiopathic nystagmus locus at Xq24-q26.3. Molecular Vision, 2006, 12, 1211-6.	1.1	6
29	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
30	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
31	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
32	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
33	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
34	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
35	Is This Actually CIN?. Pediatric Neurology, 2015, 53, e11.	1.0	1
36	Novel therapeutics in nystagmus: what has the genetics taught us so far?. Therapeutic Advances in Rare Disease, 2021, 2, 263300402199871.	0.3	1

#	Article	IF	Citations
37	Comparison of mouse and human retinal morphology and function in albinism: potential implications for therapeutic development. Lancet, The, 2017, 389, S59.	6.3	0
38	A novel method for examining corneal endothelial cell morphology in infants. Journal of AAPOS, 2017, 21, 241-243.e1.	0.2	0
39	Reply. Journal of AAPOS, 2019, 23, 124.	0.2	0
40	Infantile nystagmus. , 2022, , 403-406.		0