Dorothy A Thompson

List of Publications by Citations

83 papers	2,151 citations	24 h-index	44 g-index
94 ext. papers	2,532 ext. citations	4.2 avg, IF	4.23 L-index

#	Paper	IF	Citations
83	Epilepsy, ataxia, sensorineural deafness, tubulopathy, and KCNJ10 mutations. *New England Journal of Medicine*, **2009**, 360, 1960-70	59.2	421
82	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	11	235
81	Phenotypic variability in patients with retinal dystrophies due to mutations in CRB1. *British Journal of Ophthalmology*, **2011**, 95, 811-7	5.5	79
80	Phenotypic Characterization of Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. *Circulation*, **2017**, 136, 2022-2033	16.7	75
79	Monitoring visual function in children with syndromic craniosynostosis: a comparison of 3 methods. *JAMA Ophthalmology*, **2006**, 124, 1119-26		62
78	Altered electroretinograms in patients with KCNJ10 mutations and EAST syndrome. *Journal of Physiology*, **2011**, 589, 1681-9	3.9	54
77	Ophthalmological aspects of Pierson syndrome. *American Journal of Ophthalmology*, **2008**, 146, 602-611	4.9	53
76	Modulation of amblyopia therapy following early surgery for unilateral congenital cataracts. *British Journal of Ophthalmology*, **1995**, 79, 802-6	5.5	53
75	An assessment of the apex microarray technology in genotyping patients with Leber congenital amaurosis and early-onset severe retinal dystrophy. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 5684-9		50
74	ARNT2 mutation causes hypopituitarism, post-natal microcephaly, visual and renal anomalies. *Brain*, **2013**, 136, 3096-105	11.2	48
73	RDH12 retinopathy: novel mutations and phenotypic description. *Molecular Vision*, **2011**, 17, 2706-16	2.3	44
72	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, 6934-44		42
71	Phenotypic diversity and mutation spectrum in hypotrichosis with juvenile macular dystrophy. *Journal of Investigative Dermatology*, **2003**, 121, 1217-20	4.3	42
70	Prevalence of abnormal pattern reversal visual evoked potentials in craniosynostosis. *Plastic and Reconstructive Surgery*, **2006**, 118, 184-92	2.7	39
69	Childhood-onset autosomal recessive bestrophinopathy. *JAMA Ophthalmology*, **2011**, 129, 1088-93		36
68	Early VEP and ERG evidence of visual dysfunction in autosomal recessive osteopetrosis. *Neuropediatrics*, **1998**, 29, 137-44	1.6	36
67	Sustained raised intracranial pressure implicated only by pattern reversal visual evoked potentials after cranial vault expansion surgery. *Pediatric Neurosurgery*, **2003**, 39, 75-80	0.9	34

66	Screening of SPATA7 in patients with Leber congenital amaurosis and severe childhood-onset retinal dystrophy reveals disease-causing mutations **2011**, 52, 3032-8		31
65	The effect of stimulus contrast on the latency and amplitude of the pattern electroretinogram. *Vision Research*, **1989**, 29, 309-13	2.1	29
64	The achiasmia spectrum: congenitally reduced chiasmal decussation. *British Journal of Ophthalmology*, **2005**, 89, 1311-7	5.5	28
63	Visual outcomes following intraophthalmic artery melphalan for patients with refractory retinoblastoma and age appropriate vision. *British Journal of Ophthalmology*, **2013**, 97, 1464-70	5.5	27
62	Ocular and neurodevelopmental features of Duchenne muscular dystrophy: a signature of dystrophin function in the central nervous system. *European Journal of Human Genetics*, **2016**, 24, 562-8	5.3	26
61	Leber congenital amaurosis associated with AIPL1: challenges in ascribing disease causation, clinical findings, and implications for gene therapy. *PLoS ONE*, **2012**, 7, e32330	3.7	26
60	Pediatric clinical visual electrophysiology: a survey of actual practice. *Documenta Ophthalmologica*, **2006**, 113, 193-204	2.2	25
59	Visual-evoked potential evidence of chiasmal hypoplasia. *Ophthalmology*, **1999**, 106, 2354-61	7.3	24
58	Computation of the luminance and pattern components of the bar pattern electroretinogram. *Documenta Ophthalmologica*, **1987**, 66, 233-44	2.2	24
57	Joubert syndrome: long-term follow-up. *Developmental Medicine and Child Neurology*, **2004**, 46, 694-9	3.3	24
56	Full-field electroretinogram in autism spectrum disorder. *Documenta Ophthalmologica*, **2016**, 132, 83-99	2.2	23
55	Vertical or asymmetric nystagmus need not imply neurological disease. *British Journal of Ophthalmology*, **2000**, 84, 175-80	5.5	23
54	Foveal cavitation as an optical coherence tomography finding in central cone dysfunction. *Retina*, **2012**, 32, 1411-9	3.6	22
53	Unilateral BEST1-Associated Retinopathy. *American Journal of Ophthalmology*, **2016**, 169, 24-32	4.9	21
52	VEP estimation of visual acuity: a systematic review. *Documenta Ophthalmologica*, **2021**, 142, 25-74	2.2	21
51	The optokinetic response differences between congenital profound and nonprofound unilateral visual deprivation. *Ophthalmology*, **1995**, 102, 1615-22	7.3	20
50	ISCEV extended protocol for the dark-adapted red flash ERG. *Documenta Ophthalmologica*, **2018**, 136, 191-197	2.2	20
49	Retinal nerve fibre layer thinning is associated with worse visual outcome after optic neuritis in children with a relapsing demyelinating syndrome. *Developmental Medicine and Child Neurology*, **2018**, 60, 1244-1250	3.3	19

48	The effects of image degradation on retinal illuminance and pattern responses to checkerboard stimuli. *Documenta Ophthalmologica*, **1987**, 66, 267-75	2.2	19
47	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
46	Clinical Characterization of CNGB1-Related Autosomal Recessive Retinitis Pigmentosa. *JAMA Ophthalmology*, **2017**, 135, 137-144	3.9	17
45	ISCEV extended protocol for VEP methods of estimation of visual acuity. *Documenta Ophthalmologica*, **2021**, 142, 17-24	2.2	17
44	THE PHYSIOLOGY OF THE RETINAL PIGMENT EPITHELIUM IN DANON DISEASE. *Retina*, **2016**, 36, 629-38	3.6	16
43	Visual field loss in children with craniosynostosis. *Childs Nervous System*, **2011**, 27, 1289-96	1.7	16
42	Juvenile xanthogranuloma with presumed involvement of the optic disc and retina. *JAMA Ophthalmology*, **2004**, 122, 1551-5		15
41	Infantile hyperinsulinism associated with enteropathy, deafness and renal tubulopathy: clinical manifestations of a syndrome caused by a contiguous gene deletion located on chromosome 11p. *Journal of Pediatric Endocrinology and Metabolism*, **2004**, 17, 1613-21	1.6	15
40	Electrophysiological and Eye-Movement Abnormalities in Children With the Bardet-Biedl Syndrome. *Journal of Pediatric Ophthalmology and Strabismus*, **1995**, 32, 364-367	0.9	15
39	When do asymmetrical full-field pattern reversal visual evoked potentials indicate visual pathway dysfunction in children?. *Documenta Ophthalmologica*, **2011**, 122, 9-18	2.2	14
38	Retinal characteristics of the congenital disorder of glycosylation PMM2-CDG. *Journal of Inherited Metabolic Disease*, **2013**, 36, 1039-47	5.4	11
37	Motion and pattern cortical potentials in adults with high-functioning autism spectrum disorder. *Documenta Ophthalmologica*, **2012**, 125, 219-27	2.2	11
36	Retinal on-pathway deficit in congenital disorder of glycosylation due to phosphomannomutase deficiency. *JAMA Ophthalmology*, **2012**, 130, 712-9		11
35	Multi-centre variability of ISCEV standard ERGs in two normal adults. *Documenta Ophthalmologica*, **2015**, 130, 83-101	2.2	10
34	The reproducibility of binocular pattern reversal visual evoked potentials: a single subject design. *Documenta Ophthalmologica*, **2011**, 122, 133-9	2.2	10
33	The origins of luminance and pattern responses of the pattern electroretinogram. *International Journal of Psychophysiology*, **1994**, 16, 219-27	2.9	10
32	Expansion of ocular phenotypic features associated with mutations in ADAMTS18. *JAMA Ophthalmology*, **2014**, 132, 996-1001	3.9	8
31	The changing shape of the ISCEV standard pattern onset VEP. *Documenta Ophthalmologica*, **2017**, 135, 69-76	2.2	7

30	An alternative electroretinography protocol for children: a study of diagnostic agreement and accuracy relative to ISCEV standard electroretinograms. *Acta Ophthalmologica*, **2021**,	3.7	7
29	Review of cases presenting with microcephaly and bilateral congenital cataract in a paediatric cataract clinic. *Eye*, **2008**, 22, 273-81	4.4	6
28	Saccadic instabilities in albinism without nystagmus. *Experimental Brain Research*, **2006**, 175, 45-9	2.3	6
27	A meta-analysis of clinical electro-oculography values. *Documenta Ophthalmologica*, **2017**, 135, 219-232	2.2	5
26	Visual impairment, severe visual impairment, and blindness in children in Britain (BCVIS2): a national observational study. *The Lancet Child and Adolescent Health*, **2021**, 5, 190-200	14.5	5
25	Light-Adapted Electroretinogram Differences in Autism Spectrum Disorder. *Journal of Autism and Developmental Disorders*, **2020**, 50, 2874-2885	4.6	4
24	Colour, contrast and the visual evoked potential. *Ophthalmic and Physiological Optics*, **1992**, 12, 225-8	4.1	4
23	The effect of 0.5% thymoxamine on the pattern-onset electroretinogram. *Documenta Ophthalmologica*, **1989**, 72, 47-54	2.2	4
22	Cataract in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD). *Ophthalmic Genetics* , **2003**, 24, 49-57	1.2	3
21	An optical stimulator for studying the topography of electrical and magnetic visual evoked responses. *Documenta Ophthalmologica*, **1992**, 81, 219-25	2.2	3
20	An overview of bifocal contact lenses. *Journal of the British Contact Lens Association*, **1991**, 14, 71-74		3
19	Visual outcomes in children with syndromic craniosynostosis: a review of 165 cases. *Eye*, **2021**,	4.4	3
18	Pattern-onset and OFFset visual evoked potentials in the diagnosis of hemianopic field defects. *Documenta Ophthalmologica*, **2021**, 142, 165-176	2.2	3
17	Congenital high myopia and central macular atrophy: a report of 3 families. *Eye*, **2015**, 29, 936-42	4.4	2
16	Distortion of spatial selectivity by pattern onset stimulation. *Documenta Ophthalmologica*, **1989**, 72, 1-8	2.2	2
15	The photopic negative response in autism spectrum disorder. *Australasian journal of optometry, The* , **2021**, 104, 841-847	2.7	2
14	An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. *Eye*, **2021**, 35, 2438-2448	4.4	2
13	Pattern Onset ERGs and VEPs Produced by Patterns Arising From Light Increment and Decrement **2018**, 59, 94-99		2

12	ERGs on the brain: the benefits of simultaneous flash retinal and cortical responses in paediatric cerebral visual impairment. *Documenta Ophthalmologica*, **2018**, 136, 223-227	2.2	1
11	Variability of flashes and background luminances of clinical electroretinography stimuli across 14 UK centres. *Journal of Modern Optics*, **2013**, 60, 1209-1216	1.1	1
10	Hereditary primary lateral sclerosis with cone dysfunction. *Ophthalmic Genetics*, **2010**, 31, 221-6	1.2	1
9	Visual electrophysiology **2013**, 55-62		1
8	Misaligned foveal morphology and sector retinal dysfunction in AKT1-mosaic Proteus syndrome. *Documenta Ophthalmologica*, **2021**, 142, 119-126	2.2	1
7	Case report: Unilateral optic nerve aplasia and developmental hemi-chiasmal dysplasia with VEP misrouting. *Documenta Ophthalmologica*, **2021**, 142, 247-255	2.2	1
6	The electroretinogram b-wave amplitude: a differential physiological measure for Attention Deficit Hyperactivity Disorder and Autism Spectrum Disorder.. *Journal of Neurodevelopmental Disorders*, **2022**, 14, 30	4.6	1
5	Diffuse bear-track retina: profound, bilateral, grouped congenital pigmentation of the retinal pigment epithelium in an infant. *Journal of AAPOS*, **2020**, 24, 384-386	1.3	0
4	Giant pattern VEPs in children. *European Journal of Paediatric Neurology*, **2021**, 34, 33-42	3.8	0
3	Pediatric Visual Electrophysiology **2006**, 1-74		
2	Pediatric Visual Electrophysiology **2003**, 90-121		
1	Clinicians in PPE: A Visually Impaired Patient‘s View. *Journal of Pediatric Ophthalmology and Strabismus*, **2021**, 58, 407	0.9	