

# Dorothy A Thompson

## List of Publications by Year in descending order

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93  
papers

2,885  
citations

185998

28  
h-index

189595

50  
g-index

94  
all docs

94  
docs citations

94  
times ranked

3961  
citing authors

#	ARTICLE	IF	CITATIONS
1	Epilepsy, Ataxia, Sensorineural Deafness, Tubulopathy, and <i>KCNJ10</i> Mutations. <i>New England Journal of Medicine</i> , 2009, 360, 1960-1970.	13.9	518
2	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	2.6	343
3	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
4	Phenotypic variability in patients with retinal dystrophies due to mutations in <i>CRB1</i> . <i>British Journal of Ophthalmology</i> , 2011, 95, 811-817.	2.1	95
5	Monitoring Visual Function in Children With Syndromic Craniosynostosis. <i>JAMA Ophthalmology</i> , 2006, 124, 1119.	2.6	71
6	Ophthalmological Aspects of Pierson Syndrome. <i>American Journal of Ophthalmology</i> , 2008, 146, 602-611.e1.	1.7	66
7	Altered electroretinograms in patients with <i>KCNJ10</i> mutations and EAST syndrome. <i>Journal of Physiology</i> , 2011, 589, 1681-1689.	1.3	66
8	<i>ARNT2</i> mutation causes hypopituitarism, post-natal microcephaly, visual and renal anomalies. <i>Brain</i> , 2013, 136, 3096-3105.	3.7	66
9	Modulation of amblyopia therapy following early surgery for unilateral congenital cataracts.. <i>British Journal of Ophthalmology</i> , 1995, 79, 802-806.	2.1	62
10	The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in <i>CRX</i> , With Report of a Novel Macular Dystrophy Phenotype. <i>Investigative Ophthalmology and Visual Science</i> , 2014, 55, 6934-6944.	3.3	59
11	VEP estimation of visual acuity: a systematic review. <i>Documenta Ophthalmologica</i> , 2021, 142, 25-74.	1.0	57
12	An Assessment of the Apex Microarray Technology in Genotyping Patients with Leber Congenital Amaurosis and Early-Onset Severe Retinal Dystrophy. , 2007, 48, 5684.		56
13	<i>RDH12</i> retinopathy: novel mutations and phenotypic description. <i>Molecular Vision</i> , 2011, 17, 2706-16.	1.1	47
14	Early VEP and ERG Evidence of Visual Dysfunction in Autosomal Recessive Osteopetrosis. <i>Neuropediatrics</i> , 1998, 29, 137-144.	0.3	46
15	Phenotypic Diversity and Mutation Spectrum in Hypotrichosis with Juvenile Macular Dystrophy. <i>Journal of Investigative Dermatology</i> , 2003, 121, 1217-1220.	0.3	46
16	Prevalence of Abnormal Pattern Reversal Visual Evoked Potentials in Craniosynostosis. <i>Plastic and Reconstructive Surgery</i> , 2006, 118, 184-192.	0.7	46
17	Childhood-Onset Autosomal Recessive Bestrophinopathy. <i>JAMA Ophthalmology</i> , 2011, 129, 1088.	2.6	46
18	Sustained Raised Intracranial Pressure Implicated Only by Pattern Reversal Visual Evoked Potentials after Cranial Vault Expansion Surgery. <i>Pediatric Neurosurgery</i> , 2003, 39, 75-80.	0.4	39

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19	Full-field electroretinogram in autism spectrum disorder. <i>Documenta Ophthalmologica</i> , 2016, 132, 83-99.	1.0	38
20	Retinal nerve fibre layer thinning is associated with worse visual outcome after optic neuritis in children with a relapsing demyelinating syndrome. <i>Developmental Medicine and Child Neurology</i> , 2018, 60, 1244-1250.	1.1	38
21	Visual impairment, severe visual impairment, and blindness in children in Britain (BCVIS2): a national observational study. <i>The Lancet Child and Adolescent Health</i> , 2021, 5, 190-200.	2.7	37
22	The effect of stimulus contrast on the latency and amplitude of the pattern electroretinogram. <i>Vision Research</i> , 1989, 29, 309-313.	0.7	36
23	Ocular and neurodevelopmental features of Duchenne muscular dystrophy: a signature of dystrophin function in the central nervous system. <i>European Journal of Human Genetics</i> , 2016, 24, 562-568.	1.4	36
24	ISCEV extended protocol for the dark-adapted red flash ERG. <i>Documenta Ophthalmologica</i> , 2018, 136, 191-197.	1.0	36
25	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36
26	Screening of <i>SPATA7</i> in Patients with Leber Congenital Amaurosis and Severe Childhood-Onset Retinal Dystrophy Reveals Disease-Causing Mutations. , 2011, 52, 3032.		34
27	ISCEV extended protocol for VEP methods of estimation of visual acuity. <i>Documenta Ophthalmologica</i> , 2021, 142, 17-24.	1.0	33
28	The achiasmia spectrum: congenitally reduced chiasmal decussation. <i>British Journal of Ophthalmology</i> , 2005, 89, 1311-1317.	2.1	31
29	Joubert syndrome: long-term follow-up. <i>Developmental Medicine and Child Neurology</i> , 2004, 46, 694-9.	1.1	31
30	Visual outcomes following intraocular melphalan for patients with refractory retinoblastoma and age appropriate vision. <i>British Journal of Ophthalmology</i> , 2013, 97, 1464-1470.	2.1	29
31	Pediatric clinical visual electrophysiology: a survey of actual practice. <i>Documenta Ophthalmologica</i> , 2006, 113, 193-204.	1.0	28
32	Leber Congenital Amaurosis Associated with AIPL1: Challenges in Ascribing Disease Causation, Clinical Findings, and Implications for Gene Therapy. <i>PLoS ONE</i> , 2012, 7, e32330.	1.1	28
33	Computation of the luminance and pattern components of the bar pattern electroretinogram. <i>Documenta Ophthalmologica</i> , 1987, 66, 233-244.	1.0	27
34	FOVEAL CAVITATION AS AN OPTICAL COHERENCE TOMOGRAPHY FINDING IN CENTRAL CONE DYSFUNCTION. <i>Retina</i> , 2012, 32, 1411-1419.	1.0	27
35	Visual-evoked potential evidence of chiasmal hypoplasia <sup>11</sup> The views expressed in this publication are those of the authors and not necessarily those of the NHS Executive.. <i>Ophthalmology</i> , 1999, 106, 2354-2361.	2.5	26
36	Vertical or asymmetric nystagmus need not imply neurological disease. <i>British Journal of Ophthalmology</i> , 2000, 84, 175-180.	2.1	26

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37	Unilateral BEST1 -Associated Retinopathy. American Journal of Ophthalmology, 2016, 169, 24-32.	1.7	26
38	The Optokinetic Response Differences between Congenital Profound and Nonprofound Unilateral Visual Deprivation. Ophthalmology, 1995, 102, 1615-1622.	2.5	23
39	Clinical Characterization of <i>CNGB1</i> -Related Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2017, 135, 137.	1.4	23
40	THE PHYSIOLOGY OF THE RETINAL PIGMENT EPITHELIUM IN DANON DISEASE. Retina, 2016, 36, 629-638.	1.0	21
41	Electrophysiological and Eye-Movement Abnormalities in Children With the Bardet-Biedl Syndrome. Journal of Pediatric Ophthalmology and Strabismus, 1995, 32, 364-367.	0.3	21
42	The effects of image degradation on retinal illuminance and pattern responses to checkerboard stimuli. Documenta Ophthalmologica, 1987, 66, 267-275.	1.0	20
43	Light-Adapted Electroretinogram Differences in Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2020, 50, 2874-2885.	1.7	20
44	Visual field loss in children with craniosynostosis. Child's Nervous System, 2011, 27, 1289-1296.	0.6	19
45	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.	0.4	18
46	Motion and pattern cortical potentials in adults with high-functioning autism spectrum disorder. Documenta Ophthalmologica, 2012, 125, 219-227.	1.0	17
47	Juvenile Xanthogranuloma With Presumed Involvement of the Optic Disc and Retina. JAMA Ophthalmology, 2004, 122, 1551.	2.6	16
48	Retinal characteristics of the congenital disorder of glycosylation PMM2-CDG. Journal of Inherited Metabolic Disease, 2013, 36, 1039-1047.	1.7	16
49	Expansion of Ocular Phenotypic Features Associated With Mutations in <i>ADAMTS18</i> . JAMA Ophthalmology, 2014, 132, 996.	1.4	15
50	When do asymmetrical full-field pattern reversal visual evoked potentials indicate visual pathway dysfunction in children?. Documenta Ophthalmologica, 2011, 122, 9-18.	1.0	14
51	Retinal On-Pathway Deficit in Congenital Disorder of Glycosylation Due to Phosphomannomutase Deficiency. JAMA Ophthalmology, 2012, 130, 712-9.	2.6	14
52	An alternative electroretinography protocol for children: a study of diagnostic agreement and accuracy relative to ISCEV standard electroretinograms. Acta Ophthalmologica, 2022, 100, 322-330.	0.6	14
53	Multi-centre variability of ISCEV standard ERGs in two normal adults. Documenta Ophthalmologica, 2015, 130, 83-101.	1.0	13
54	The changing shape of the ISCEV standard pattern onset VEP. Documenta Ophthalmologica, 2017, 135, 69-76.	1.0	13

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55	The electroretinogram b-wave amplitude: a differential physiological measure for Attention Deficit Hyperactivity Disorder and Autism Spectrum Disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 30.	1.5	13
56	The reproducibility of binocular pattern reversal visual evoked potentials: a single subject design. <i>Documenta Ophthalmologica</i> , 2011, 122, 133-139.	1.0	12
57	The photopic negative response in autism spectrum disorder. <i>Australasian journal of optometry</i> , The, 2021, 104, 841-847.	0.6	12
58	Discrete Wavelet Transform Analysis of the Electroretinogram in Autism Spectrum Disorder and Attention Deficit Hyperactivity Disorder. <i>Frontiers in Neuroscience</i> , 2022, 16, .	1.4	12
59	The origins of luminance and pattern responses of the pattern electroretinogram. <i>International Journal of Psychophysiology</i> , 1994, 16, 219-227.	0.5	11
60	Visual outcomes in children with syndromic craniosynostosis: a review of 165 cases. <i>Eye</i> , 2022, 36, 1005-1011.	1.1	10
61	The effect of 0.5% thymoxamine on the pattern-onset electroretinogram. <i>Documenta Ophthalmologica</i> , 1989, 72, 47-54.	1.0	6
62	Saccadic instabilities in albinism without nystagmus. <i>Experimental Brain Research</i> , 2006, 175, 45-49.	0.7	6
63	Review of cases presenting with microcephaly and bilateral congenital cataract in a paediatric cataract clinic. <i>Eye</i> , 2008, 22, 273-281.	1.1	6
64	An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. <i>Eye</i> , 2021, 35, 2438-2448.	1.1	6
65	Colour, contrast and the visual evoked potential. <i>Ophthalmic and Physiological Optics</i> , 1992, 12, 225-228.	1.0	5
66	A meta-analysis of clinical electro-oculography values. <i>Documenta Ophthalmologica</i> , 2017, 135, 219-232.	1.0	5
67	An optical stimulator for studying the topography of electrical and magnetic visual evoked responses. <i>Documenta Ophthalmologica</i> , 1992, 81, 219-225.	1.0	4
68	An overview of bifocal contact lenses. <i>Journal of the British Contact Lens Association</i> , 1991, 14, 71-74.	0.2	3
69	Cataract in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD). <i>Ophthalmic Genetics</i> , 2003, 24, 49-57.	0.5	3
70	Pattern Onset ERGs and VEPs Produced by Patterns Arising From Light Increment and Decrement. , 2018, 59, 94.		3
71	Pattern-onset and OFFset visual evoked potentials in the diagnosis of hemianopic field defects. <i>Documenta Ophthalmologica</i> , 2021, 142, 165-176.	1.0	3
72	Giant pattern VEPs in children. <i>European Journal of Paediatric Neurology</i> , 2021, 34, 33-42.	0.7	3

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73	Distortion of spatial selectivity by pattern onset stimulation. Documenta Ophthalmologica, 1989, 72, 1-8.	1.0	2
74	Hereditary primary lateral sclerosis with cone dysfunction. Ophthalmic Genetics, 2010, 31, 221-226.	0.5	2
75	Congenital high myopia and central macular atrophy: a report of 3 families. Eye, 2015, 29, 936-942.	1.1	2
76	ERGs on the brain: the benefits of simultaneous flash retinal and cortical responses in paediatric cerebral visual impairment. Documenta Ophthalmologica, 2018, 136, 223-227.	1.0	2
77	Case report: Unilateral optic nerve aplasia and developmental hemi-chiasmal dysplasia with VEP misrouting. Documenta Ophthalmologica, 2021, 142, 247-255.	1.0	2
78	Introduction to the special section on Visual electrophysiology 2021: a coming of age. Eye, 2021, 35, 2339-2340.	1.1	2
79	Prognostic value of transient pattern visual evoked potentials in children with cerebral visual impairment. Developmental Medicine and Child Neurology, 2022, 64, 618-624.	1.1	2
80	Electrophysiological and fundoscopic detection of intracranial hypertension in craniosynostosis. Eye, 2023, 37, 139-145.	1.1	2
81	A clinical classification system for paravenous pigmentary chorioretinal atrophy (PPCRA) in childhood. Journal of AAPOS, 2011, 15, e21.	0.2	1
82	Variability of flashes and background luminances of clinical electroretinography stimuli across 14 UK centres. Journal of Modern Optics, 2013, 60, 1209-1216.	0.6	1
83	Misaligned foveal morphology and sector retinal dysfunction in AKT1-mosaic Proteus syndrome. Documenta Ophthalmologica, 2021, 142, 119-126.	1.0	1
84	Visual electrophysiology. , 2013, , 55-62.		1
85	Diffuse bear-track retina: profound, bilateral, grouped congenital pigmentation of the retinal pigment epithelium in an infant. Journal of AAPOS, 2020, 24, 384-386.	0.2	1
86	Pediatric Visual Electrophysiology. , 2003, , 90-121.		0
87	Pediatric Visual Electrophysiology. , 2006, , 1-74.		0
88	Congenital optic nerve aplasia. Journal of AAPOS, 2012, 16, e14.	0.2	0
89	The 50th ISCEV international symposium abstract issue. Documenta Ophthalmologica, 2012, 124, 1-2.	1.0	0
90	Exploring the microvascular abnormalities in a cohort of paediatric patients with spinal muscular atrophy. Neuromuscular Disorders, 2016, 26, S105.	0.3	0

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91	Clinicians in PPE: A Visually Impaired Patient's View. Journal of Pediatric Ophthalmology and Strabismus, 2021, 58, 407-407.	0.3	0
92	The effect of spatio-temporal chromatic and achromatic contrast on VEPs. Ophthalmic and Physiological Optics, 1992, 12, 97.	1.0	0
93	The 59th annual ISCEV symposium Liverpool, United Kingdom, 3rd-6th August 2022. Documenta Ophthalmologica, 0, , .	1.0	0