Elias I Traboulsi

List of Publications by Year
in descending order

74	2,505	201674 27	197818 49
papers	citations	h-index	g-index

75	75	75	2453
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence and Importance of Pigmented Ocular Fundus Lesions in Gardner's Syndrome. New England Journal of Medicine, 1987, 316, 661-667.	27.0	254
2	Human microphthalmia associated with mutations in the retinal homeobox gene CHX10. Nature Genetics, 2000, 25, 397-401.	21.4	245
3	Comprehensive molecular diagnosis of 179 Leber congenital amaurosis and juvenile retinitis pigmentosa patients by targeted next generation sequencing. Journal of Medical Genetics, 2013, 50, 674-688.	3.2	139
4	Choroideremia: A review of general findings and pathogenesis. Ophthalmic Genetics, 2012, 33, 57-65.	1.2	105
5	Phenotypic Overlap Between Familial Exudative Vitreoretinopathy and Microcephaly, Lymphedema, and Chorioretinal Dysplasia Caused byKIF11Mutations. JAMA Ophthalmology, 2014, 132, 1393.	2.5	95
6	Update on the Morning Glory Disc Anomaly. Ophthalmic Genetics, 2008, 29, 47-52.	1.2	94
7	Update on the molecular genetics of retinitis pigmentosa. Ophthalmic Genetics, 2001, 22, 133-154.	1.2	91
8	Systemic and Ocular Findings in 100 Patients With Optic Nerve Hypoplasia. Journal of Child Neurology, 2006, 21, 949-956.	1.4	80
9	A Clinicopathologic Study of the Eyes in Familial Adenomatous Polyposis by Extracolonic Manifestations (Gardner's Syndrome). American Journal of Ophthalmology, 1990, 110, 550-561.	3.3	76
10	Congenital abnormalities of cranial nerve development: overview, molecular mechanisms, and further evidence of heterogeneity and complexity of syndromes with congenital limitation of eye movements. Transactions of the American Ophthalmological Society, 2004, 102, 373-89.	1.4	71
11	Pigmented Ocular Fundus Lesions in the Inherited Gastrointestinal Polyposis Syndromes and in Hereditary Nonpolyposis Colorectal Cancer. Ophthalmology, 1988, 95, 964-969.	5.2	68
12	Intracranial Vascular Anomalies in Patients With Morning Glory Disk Anomaly. American Journal of Ophthalmology, 2006, 142, 644-650.e2.	3.3	67
13	Congenital Hypertrophy of the Retinal Pigment Epithelium Predicts Colorectal Polyposis in Gardner's Syndrome. JAMA Ophthalmology, 1990, 108, 525.	2.4	62
14	The Value of Keratometry and Central Corneal Thickness Measurements in the Clinical Diagnosis of Marfan Syndrome. American Journal of Ophthalmology, 2008, 145, 997-1001.e1.	3.3	59
15	Lymphedema-distichiasis syndrome and FOXC2 gene mutation. American Journal of Ophthalmology, 2002, 134, 592-596.	3.3	57
16	A HISTOPATHOLOGIC STUDY OF THE PIGMENTED FUNDUS LESIONS IN FAMILIAL ADENOMATOUS POLYPOSIS. Retina, 1992, 12, 35-42.	1.7	51
17	Hepatoblastoma, pigmented ocular fundus lesions and jaw lesions in gardner syndrome. American Journal of Medical Genetics Part A, 1988, 29, 323-332.	2.4	50
18	Dominant Inheritance of Optic Pits. American Journal of Ophthalmology, 1997, 124, 112-113.	3.3	48

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19	Microfibril abnormalities of the lens capsule in patients with Marfan syndrome and ectopia lentis. Ophthalmic Genetics, 2000, 21, 9-15.	1.2	47
20	Choroideremia: Effect of age on visual acuity in patients and female carriers. Ophthalmic Genetics, 2012, 33, 66-73.	1.2	46
21	Strabismus in the Marfan Syndrome. American Journal of Ophthalmology, 1994, 117, 632-635.	3.3	45
22	Outcomes of Bilateral Cataracts Removed in Infants 1 to 7 Months of Age Using the Toddler Aphakia and Pseudophakia Treatment Study Registry. Ophthalmology, 2020, 127, 501-510.	5.2	40
23	Ophthalmological findings in 74 patients with mitochondrial disease. Ophthalmic Genetics, 2017, 38, 67-69.	1.2	36
24	Surgical management of lens subluxation in Marfan syndrome. Journal of AAPOS, 2014, 18, 140-146.	0.3	33
25	Pigmented ocular fundus lesions and APC mutations in familial adenomatous polyposis. Ophthalmic Genetics, 1996, 17, 167-174.	1.2	32
26	Ocular findings in ichthyosis follicularis-alopecia-photophobia (IFAP) syndrome. Ophthalmic Genetics, 2004, 25, 153-156.	1.2	29
27	Ocular Manifestations of Familial Adenomatous Polyposis (Gardner Syndrome). Ophthalmology Clinics of North America, 2005, 18, 163-166.	1.8	29
28	Congenital Fibrosis of the Extraocular Muscles: Report of 24 Cases Illustrating the Clinical Spectrum and Surgical Management. American Orthoptic Journal, 1993, 43, 45-53.	0.3	26
29	Predictors of visual acuity and genotype-phenotype correlates in a cohort of patients with Stargardt disease. British Journal of Ophthalmology, 2014, 98, 513-518.	3.9	26
30	Autosomal Dominant Retinitis Pigmentosa Secondary to Pre-mRNA Splicing-Factor GenePRPF31(RP11): Review of Disease Mechanism and Report of a Family with a Novel 3-Base Pair Insertion. Ophthalmic Genetics, 2013, 34, 183-188.	1.2	25
31	Ocular manifestations of the autoinflammatory syndromes. Ophthalmic Genetics, 2012, 33, 179-186.	1.2	24
32	The Use of a Mobile Van for School Vision Screening: Results of 63 841 Evaluations. American Journal of Ophthalmology, 2016, 163, 108-114.e1.	3.3	24
33	Stickler syndrome. Current Opinion in Ophthalmology, 2019, 30, 306-313.	2.9	24
34	Ophthalmologic Abnormalities in Mowat-Wilson Syndrome and a Mutation inZEB2. Ophthalmic Genetics, 2012, 33, 159-160.	1.2	21
35	Genome-Wide Association Study Identifies a Susceptibility Locus for Comitant Esotropia and Suggests a Parent-of-Origin Effect. , 2018, 59, 4054.		21
36	A novel dominant CRX mutation causes adult-onset macular dystrophy. Ophthalmic Genetics, 2018, 39, 120-124.	1.2	20

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37	Association of Optic Nerve Hypoplasia With Mitochondrial Cytopathies. Journal of Child Neurology, 2006, 21, 956-960.	1.4	19
38	Molecular biology and genetics of embryonic eyelid development. Ophthalmic Genetics, 2016, 37, 252-259.	1.2	19
39	Novel mutation in CTNNB1 causes familial exudative vitreoretinopathy (FEVR) and microcephaly: case report and review of the literature. Ophthalmic Genetics, 2020, 41, 63-68.	1.2	19
40	Outcomes of Bilateral Cataract Surgery in Infants 7 to 24 Months of Age Using the Toddler Aphakia and Pseudophakia Treatment Study Registry. Ophthalmology, 2021, 128, 302-308.	5.2	16
41	Anterior segment malformations in 18q-(de Grouchy) syndrome. Ophthalmic Paediatrics and Genetics, 1993, 14, 91-94.	0.4	13
42	Pigmented and depigmented lesions of the ocular fundus. Current Opinion in Ophthalmology, 2012, 23, 337-343.	2.9	13
43	Familial Wolf-Hirschhorn syndrome associated with Rieger anomaly of the eye. Ophthalmic Paediatrics and Genetics, 1990, 11, 23-30.	0.4	12
44	Presenting signs and clinical diagnosis in individuals referred to rule out Marfan syndrome. Ophthalmic Genetics, 2003, 24, 35-39.	1.2	12
45	Identification of a mutation in CNNM4 by whole exome sequencing in an Amish family and functional link between CNNM4 and IQCB1. Molecular Genetics and Genomics, 2018, 293, 699-710.	2.1	12
46	Vision First, a program to detect and treat eye diseases in young children: the first four years. Transactions of the American Ophthalmological Society, 2008, 106, 179-85; discussion 185-6.	1.4	12
47	Coats' disease and central nervous system venous malformation. Ophthalmic Genetics, 1996, 17, 215-218.	1.2	11
48	Evidence of retinal degeneration in Wolfram syndrome. Ophthalmic Genetics, 2019, 40, 34-38.	1.2	9
49	Congenital Abnormalities of the Optic Nerve: From Gene Mutation to Clinical Expression. Current Neurology and Neuroscience Reports, 2013, 13, 363.	4.2	8
50	Recurrent Rare Copy Number Variants Increase Risk for Esotropia. , 2020, 61, 22.		8
51	Cutis marmorata telangiectatica congenita: a focus on its diagnosis, ophthalmic anomalies, and possible etiologic factors. Ophthalmic Genetics, 2020, 41, 101-107.	1.2	8
52	Microcephaly and Congenital Grouped Pigmentation of the Retinal Pigment Epithelium Associated with Submicroscopic Deletions of 13q33.3-q34 and 11p15.4. Ophthalmic Genetics, 2009, 30, 136-141.	1.2	7
53	Outcomes of Strabismus Surgery with or without Trainee Participation as Surgeon. Ophthalmology, 2014, 121, 2066-2069.	5.2	6
54	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

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55	Mitochondrial DNA A3243G variant-associated retinopathy: a meta-analysis of the clinical course of visual acuity and correlation with systemic manifestations. Ophthalmic Genetics, 2021, 42, 420-430.	1.2	4
56	TULP1 related retinal dystrophy: report of rare and novel variants with a previously undescribed phenotype in two cases. Ophthalmic Genetics, 2022, 43, 277-281.	1.2	4
57	The Genetics of Strabismus. American Orthoptic Journal, 2001, 51, 67-74.	0.3	3
58	Hope and major strides for genetic diseases of the eye. Journal of Genetics, 2009, 88, 395-397.	0.7	3
59	The effects of surgical factors on postoperative astigmatism in patients enrolled in the Infant Aphakia Treatment Study (IATS). Journal of AAPOS, 2014, 18, 441-445.	0.3	3
60	Chronic Choroidal Neovascular Membrane in Choroideremia Treated With Intravitreal Bevacizumab. Ophthalmic Surgery Lasers and Imaging Retina, 2019, 50, e188-e192.	0.7	3
61	Observations on the Development and Progression of Unilateral High Myopia. American Orthoptic Journal, 2003, 53, 115-120.	0.3	2
62	Orbital rhabdomyosarcoma in a child with Leigh syndrome. Journal of AAPOS, 2018, 22, 150-152.e1.	0.3	2
63	Retinal dystrophy associated with a Kizuna (KIZ) mutation and a predominantly macular phenotype. Ophthalmic Genetics, 2019, 40, 455-460.	1.2	2
64	Hickamâ€™s Dictum: Pseudoxanthoma elasticum and Usher syndrome in a single patient. Ophthalmic Genetics, 2020, 41, 465-469.	1.2	2
65	Investigation of CEP290 genotype-phenotype correlations in a patient with retinitis pigmentosa, infertility, end-stage renal disease, and a novel mutation. Ophthalmic Genetics, 2020, 41, 171-174.	1.2	2
66	The landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying blue cone monochromacy. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	2
67	Rate of ocular trauma in children operated on for unilateral cataract in infancyâ€”data from the Infant Aphakia Treatment Study. Journal of AAPOS, 2020, 24, 301-303.	0.3	1
68	Novel clinical presentation of a CRX rod-cone dystrophy. BMJ Case Reports, 2021, 14, e233711.	0.5	1
69	Preventing Retinal Detachment in Patients with Stickler Syndrome: The Effects of Preemptive Laser Photocoagulation. Ophthalmology Retina, 2022, 6, 261-262.	2.4	1
70	Steering-wheel ocular injuries. Orbit, 1988, 7, 267-268.	0.8	0
71	Evaluation and Management of the Patient with Subluxated Lenses. , 2016, , 199-209.		0
72	The Phakomatoses. , 2016, , 359-378.		0

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73	Best Practices for Building and Supporting Effective ACGME-Mandated Program Evaluation Committees. MedEdPORTAL: the Journal of Teaching and Learning Resources, 2020, 16, 11039.	1.2	0
74	Severe retinal complications in Knobloch Syndrome - Three siblings without clinically apparent occipital defects and a review of the literature. Ophthalmic Genetics, 2022, , 1-9.	1.2	0