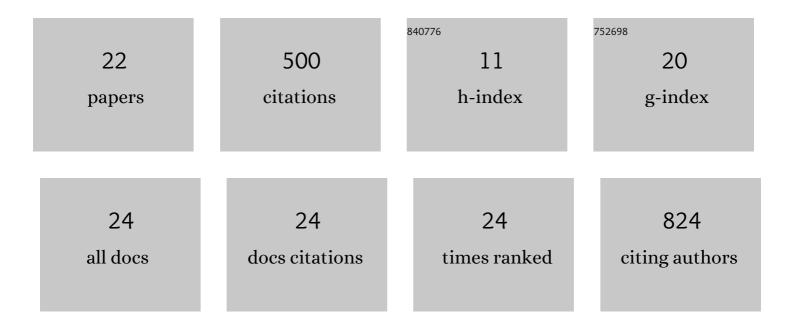
Delphine Blain

List of Publications by Year in descending order

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DEIDHINE RIAIN

#	Article	IF	CITATIONS
1	Uveal coloboma: clinical and basic science update. Current Opinion in Ophthalmology, 2006, 17, 447-470.	2.9	116
2	Compound heterozygosity for mutations in <i>PAX6</i> in a patient with complex brain anomaly, neonatal diabetes mellitus, and microophthalmia. American Journal of Medical Genetics, Part A, 2009, 149A, 2543-2546.	1.2	80
3	Prevalence of Mutations in eyeGENE Probands With a Diagnosis of Autosomal Dominant Retinitis Pigmentosa. , 2013, 54, 6255.		68
4	Molecular Testing for Hereditary Retinal Disease as Part of Clinical Care. JAMA Ophthalmology, 2007, 125, 252.	2.4	37
5	Ocular Manifestations of Trichothiodystrophy. Ophthalmology, 2011, 118, 2335-2342.	5.2	30
6	Genetic testing for inherited eye conditions in over 6,000 individuals through the <scp>eyeGENE</scp> network. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 828-837.	1.6	23
7	Increased Corneal Thickness In Patients With Ocular Coloboma. Journal of AAPOS, 2006, 10, 175-177.	0.3	19
8	Genomics in the Era of Molecular Ophthalmology. JAMA Ophthalmology, 2008, 126, 424.	2.4	18
9	Genotype–phenotype associations in a large <i>PRPH2</i> â€related retinopathy cohort. Human Mutation, 2020, 41, 1528-1539.	2.5	18
10	Molecular Diagnosis and Genetic Counseling in Ophthalmology. JAMA Ophthalmology, 2007, 125, 196.	2.4	16
11	Highâ€ŧhroughput custom capture sequencing identifies novel mutations in colobomaâ€associated genes: Mutation in DNAâ€binding domain of retinoic acid receptor beta affects nuclear localization causing ocular coloboma. Human Mutation, 2020, 41, 678-695.	2.5	13
12	Factor VII deficiency and developmental abnormalities in a patient with partial monosomy of 13q and trisomy of 16p: case report and review of the literature. BMC Medical Genetics, 2006, 7, 2.	2.1	11
13	Systemic Diagnostic Testing in Patients With Apparently Isolated Uveal Coloboma. American Journal of Ophthalmology, 2013, 156, 1159-1168.e4.	3.3	11
14	Ocular and Systemic Findings in Adults with Uveal Coloboma. Ophthalmology, 2020, 127, 1772-1774.	5.2	8
15	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
16	Motivations and Decision Making Processes of Men With X-linked Retinoschisis Considering Participation in an Ocular Gene Therapy Trial. American Journal of Ophthalmology, 2019, 204, 90-96.	3.3	7
17	Retinoschisis associated with Kearns-Sayre syndrome. Ophthalmic Genetics, 2020, 41, 497-500.	1.2	7
18	<i>De novo</i> frameshift mutation in <i>YAP1</i> associated with bilateral uveal coloboma and microphthalmia. Ophthalmic Genetics, 2022, 43, 513-517.	1.2	4

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#	Article	IF	CITATIONS
19	Clinical Phenotypes of CDHR1-Associated Retinal Dystrophies. Genes, 2022, 13, 925.	2.4	3
20	Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. Ophthalmic Genetics, 2021, 42, 320-325.	1.2	2
21	Review of evidence for environmental causes of uveal coloboma. Survey of Ophthalmology, 2021, , .	4.0	1
22	Response to Finsterer's "Exclude hereditary and acquired differential disorders before attributing retinoschisis to Kears-Sayre syndrome― Ophthalmic Genetics, 2021, 42, 100-100.	1.2	0