Delphine Blain

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6833799/publications.pdf

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22	500	840776	752698
papers	citations	11	20
		h-index	g-index

24	24	24	824
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
2	De novo frameshift mutation in YAP1 associated with bilateral uveal coloboma and microphthalmia. Ophthalmic Genetics, 2022, 43, 513-517.	1.2	4
3	Clinical Phenotypes of CDHR1-Associated Retinal Dystrophies. Genes, 2022, 13, 925.	2.4	3
4	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
5	Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. Ophthalmic Genetics, 2021, 42, 320-325.	1.2	2
6	Review of evidence for environmental causes of uveal coloboma. Survey of Ophthalmology, 2021, , .	4.0	1
7	High-throughput custom capture sequencing identifies novel mutations in coloboma-associated genes: Mutation in DNA-binding domain of retinoic acid receptor beta affects nuclear localization causing ocular coloboma. Human Mutation, 2020, 41, 678-695.	2.5	13
8	Retinoschisis associated with Kearns-Sayre syndrome. Ophthalmic Genetics, 2020, 41, 497-500.	1.2	7
9	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
10	Ocular and Systemic Findings in Adults with Uveal Coloboma. Ophthalmology, 2020, 127, 1772-1774.	5.2	8
11	Genotype–phenotype associations in a large PRPH2 -related retinopathy cohort. Human Mutation, 2020, 41, 1528-1539.	2.5	18
12	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
13	Systemic Diagnostic Testing in Patients With Apparently Isolated Uveal Coloboma. American Journal of Ophthalmology, 2013, 156, 1159-1168.e4.	3.3	11
14	Prevalence of Mutations in eyeGENE Probands With a Diagnosis of Autosomal Dominant Retinitis Pigmentosa. , 2013, 54, 6255.		68
15	Ocular Manifestations of Trichothiodystrophy. Ophthalmology, 2011, 118, 2335-2342.	5.2	30
16	Compound heterozygosity for mutations in PAX6 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
17	Genomics in the Era of Molecular Ophthalmology. JAMA Ophthalmology, 2008, 126, 424.	2.4	18
18	Molecular Testing for Hereditary Retinal Disease as Part of Clinical Care. JAMA Ophthalmology, 2007, 125, 252.	2.4	37

#	Article	IF	Citations
19	Molecular Diagnosis and Genetic Counseling in Ophthalmology. JAMA Ophthalmology, 2007, 125, 196.	2.4	16
20	Increased Corneal Thickness In Patients With Ocular Coloboma. Journal of AAPOS, 2006, 10, 175-177.	0.3	19
21	Uveal coloboma: clinical and basic science update. Current Opinion in Ophthalmology, 2006, 17, 447-470.	2.9	116
22	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