# Delphine Blain

List of Publications by Year
in descending order

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

Version: 2024-02-01

| 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 | Uveal coloboma: clinical and basic science update. Current Opinion in Ophthalmology, 2006, 17, 447-470. | 2.9 | 116 |
| 2 | 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 |
| 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 | Genotypeâ€“phenotype associations in a large PRPH2 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€throughput 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 CNGA3 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 | De novo frameshift mutation in YAP1 associated with bilateral uveal coloboma and microphthalmia. Ophthalmic Genetics, 2022, 43, 513-517. | 1.2 | 4 |

| # | 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 |