Isabelle Meunier

List of Publications by Year
in descending order

46 papers	909 citations	516710 16 h-index	501196 28 g-index
50 all docs	50 docs citations	50 times ranked	1330 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	Contribution of Whole-Genome Sequencing and Transcript Analysis to Decipher Retinal Diseases Associated with MFSD8 Variants. International Journal of Molecular Sciences, 2022, 23, 4294.	4.1	3
3	Macular Dystrophies. , 2022, , 3967-3995.		0
4	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
5	Next-Generation Sequencing Identifies Novel PMPCA Variants in Patients with Late-Onset Dominant Optic Atrophy. Genes, 2022, 13, 1202.	2.4	0
6	Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2021, 58, 570-578.	3.2	10
7	Melanoma-associated retinopathy during pembrolizumab treatment probably controlled by intravitreal injections of dexamethasone. Documenta Ophthalmologica, 2021, 142, 257-263.	2.2	10
8	Macular Dystrophies. , 2021, , 1-29.		0
9	CHM mutation spectrum and disease: An update at the time of human therapeutic trials. Human Mutation, 2021, 42, 323-341.	2.5	8
10	Novel roles for voltageâ€gated Tâ€type Ca ²⁺ and ClCâ€2 channels in phagocytosis and angiogenic factor balance identified in human iPSCâ€derived RPE. FASEB Journal, 2021, 35, e21406.	0.5	5
11	Allele-Specific Knockout by CRISPR/Cas to Treat Autosomal Dominant Retinitis Pigmentosa Caused by the G56R Mutation in NR2E3. International Journal of Molecular Sciences, 2021, 22, 2607.	4.1	19
12	DominantACO2mutations are a frequent cause of isolated optic atrophy. Brain Communications, 2021, 3, fcab063.	3.3	16
13	Quantification of the early pupillary dilation kinetic to assess rod and cone activity. Scientific Reports, 2021, 11, 9549.	3.3	0
14	Novel TTLL5 Variants Associated with Cone-Rod Dystrophy and Early-Onset Severe Retinal Dystrophy. International Journal of Molecular Sciences, 2021, 22, 6410.	4.1	9
15	ALPK1 Gene Mutations Drive Autoinflammation with Ectodermal Dysplasia and Progressive Vision Loss. Journal of Clinical Immunology, 2021, 41, 1671-1673.	3.8	6
16	Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. Mitochondrion, 2021, 59, 169-174.	3.4	3
17	Characterization of SSBP1-related optic atrophy and foveopathy. Scientific Reports, 2021, 11, 18703.	3.3	6
18	Retinitis Punctata Albescens and RLBP1-Allied Phenotypes. Ophthalmology Science, 2021, 1, 100052.	2.5	1

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19	Molecular Therapy for Choroideremia: Pre-clinical and Clinical Progress to Date. Molecular Diagnosis and Therapy, 2021, 25, 661-675.	3.8	1
20	CRB1-Related Retinal Dystrophies in a Cohort of 50 Patients: A Reappraisal in the Light of Specific MÃ¼ller Cell and Photoreceptor CRB1 Isoforms. International Journal of Molecular Sciences, 2021, 22, 12642.	4.1	11
21	Genome Editing in Patient iPSCs Corrects the Most Prevalent USH2A Mutations and Reveals Intriguing Mutant mRNA Expression Profiles. Molecular Therapy - Methods and Clinical Development, 2020, 17, 156-173.	4.1	56
22	Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276.	3.3	12
23	A Novel Chromosomal Translocation Identified due to Complex Genetic Instability in iPSC Generated for Choroideremia. Cells, 2019, 8, 1068.	4.1	4
24	Generation of a human iPSC line, INMi003-A, with a missense mutation in CRX associated with autosomal dominant cone-rod dystrophy. Stem Cell Research, 2019, 38, 101478.	0.7	4
25	Generation of a human iPSC line, INMi004-A, with a point mutation in CRX associated with autosomal dominant Leber congenital amaurosis. Stem Cell Research, 2019, 38, 101476.	0.7	5
26	Genome Editing as a Treatment for the Most Prevalent Causative Genes of Autosomal Dominant Retinitis Pigmentosa. International Journal of Molecular Sciences, 2019, 20, 2542.	4.1	40
27	Pathogenicity of novel atypical variants leading to choroideremia as determined by functional analyses. Human Mutation, 2019, 40, 31-35.	2.5	8
28	Dietary, environmental, and genetic risk factors of Extensive Macular Atrophy with Pseudodrusen, a severe bilateral macular atrophy of middle-aged patients. Scientific Reports, 2018, 8, 6840.	3.3	12
29	Generation of a human iPSC line, INMi002-A, carrying the most prevalent USH2A variant associated with Usher syndrome type 2. Stem Cell Research, 2018, 33, 247-250.	0.7	6
30	Generation of an iPSC line, INMi001-A, carrying the two most common USH2A mutations from a compound heterozygote with non-syndromic retinitis pigmentosa. Stem Cell Research, 2018, 33, 228-232.	0.7	7
31	The effect of PTC124 on choroideremia fibroblasts and iPSC-derived RPE raises considerations for therapy. Scientific Reports, 2018, 8, 8234.	3.3	30
32	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. Brain, 2017, 140, 2586-2596.	7.6	100
33	A novel duplication of PRMD13 causes North Carolina macular dystrophy: overexpression of PRDM13 orthologue in drosophila eye reproduces the human phenotype. Human Molecular Genetics, 2017, 26, 4367-4374.	2.9	24
34	WFS1 in Optic Neuropathies: Mutation Findings in Nonsyndromic Optic Atrophy and Assessment of Clinical Severity. Ophthalmology, 2016, 123, 1989-1998.	5.2	46
35	Clinical Evaluation and Cone Alterations in Choroideremia. Ophthalmology, 2016, 123, 1830-1832.	5.2	24
36	Clinical Characteristics and Risk Factors of Extensive Macular Atrophy with Pseudodrusen. Ophthalmology, 2016, 123, 1865-1873.	5.2	13

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37	Martinique Crinkled Retinal Pigment Epitheliopathy. Ophthalmology, 2016, 123, 2196-2204.	5.2	4
38	A new autosomal dominant eye and lung syndrome linked to mutations in TIMP3 gene. Scientific Reports, 2016, 6, 32544.	3.3	17
39	A dominant mutation inMAPKAPK3, an actor of p38 signaling pathway, causes a new retinal dystrophy involving Bruch's membrane and retinal pigment epithelium. Human Molecular Genetics, 2016, 25, 916-926.	2.9	13
40	High Prevalence of PRPH2 in Autosomal Dominant Retinitis Pigmentosa in France and Characterization of Biochemical and Clinical Features. American Journal of Ophthalmology, 2015, 159, 302-314.	3.3	29
41	Frequency and Clinical Pattern of Vitelliform Macular Dystrophy Caused by Mutations of Interphotoreceptor Matrix IMPG1 and IMPG2 Genes. Ophthalmology, 2014, 121, 2406-2414.	5.2	66
42	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	6.2	71
43	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. Ophthalmic Epidemiology, 2013, 20, 13-25.	1.7	44
44	Early-Onset Foveal Involvement in Retinitis Punctata Albescens With Mutations inRLBP1. JAMA Ophthalmology, 2013, 131, 1314.	2.5	33
45	Systematic Screening of BEST1 and PRPH2 in Juvenile and Adult Vitelliform Macular Dystrophies: A Rationale for Molecular Analysis. Ophthalmology, 2011, 118, 1130-1136.	5.2	62
46	Extensive Macular Atrophy with Pseudodrusen-like Appearance: A New Clinical Entity. American Journal of Ophthalmology, 2009, 147, 609-620.	3.3	44