

# Isabelle Meunier

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. <i>Brain</i> , 2017, 140, 2586-2596.	7.6	100
2	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. <i>American Journal of Human Genetics</i> , 2013, 93, 571-578.	6.2	71
3	Frequency and Clinical Pattern of Vitelliform Macular Dystrophy Caused by Mutations of Interphotoreceptor Matrix IMPG1 and IMPG2 Genes. <i>Ophthalmology</i> , 2014, 121, 2406-2414.	5.2	66
4	Systematic Screening of BEST1 and PRPH2 in Juvenile and Adult Vitelliform Macular Dystrophies: A Rationale for Molecular Analysis. <i>Ophthalmology</i> , 2011, 118, 1130-1136.	5.2	62
5	Genome Editing in Patient iPSCs Corrects the Most Prevalent USH2A Mutations and Reveals Intriguing Mutant mRNA Expression Profiles. <i>Molecular Therapy - Methods and Clinical Development</i> , 2020, 17, 156-173.	4.1	56
6	WFS1 in Optic Neuropathies: Mutation Findings in Nonsyndromic Optic Atrophy and Assessment of Clinical Severity. <i>Ophthalmology</i> , 2016, 123, 1989-1998.	5.2	46
7	Extensive Macular Atrophy with Pseudodrusen-like Appearance: A New Clinical Entity. <i>American Journal of Ophthalmology</i> , 2009, 147, 609-620.	3.3	44
8	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. <i>Ophthalmic Epidemiology</i> , 2013, 20, 13-25.	1.7	44
9	Genome Editing as a Treatment for the Most Prevalent Causative Genes of Autosomal Dominant Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , 2019, 20, 2542.	4.1	40
10	Early-Onset Foveal Involvement in Retinitis Punctata Albescens With Mutations in <i>RLBP1</i> . <i>JAMA Ophthalmology</i> , 2013, 131, 1314.	2.5	33
11	The effect of PTC124 on choroideremia fibroblasts and iPSC-derived RPE raises considerations for therapy. <i>Scientific Reports</i> , 2018, 8, 8234.	3.3	30
12	High Prevalence of PRPH2 in Autosomal Dominant Retinitis Pigmentosa in France and Characterization of Biochemical and Clinical Features. <i>American Journal of Ophthalmology</i> , 2015, 159, 302-314.	3.3	29
13	Clinical Evaluation and Cone Alterations in Choroideremia. <i>Ophthalmology</i> , 2016, 123, 1830-1832.	5.2	24
14	A novel duplication of PRMD13 causes North Carolina macular dystrophy: overexpression of PRDM13 orthologue in drosophila eye reproduces the human phenotype. <i>Human Molecular Genetics</i> , 2017, 26, 4367-4374.	2.9	24
15	Allele-Specific Knockout by CRISPR/Cas to Treat Autosomal Dominant Retinitis Pigmentosa Caused by the G56R Mutation in NR2E3. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2607.	4.1	19
16	A new autosomal dominant eye and lung syndrome linked to mutations in TIMP3 gene. <i>Scientific Reports</i> , 2016, 6, 32544.	3.3	17
17	Dominant <i>ACO2</i> mutations are a frequent cause of isolated optic atrophy. <i>Brain Communications</i> , 2021, 3, fcab063.	3.3	16
18	Clinical Characteristics and Risk Factors of Extensive Macular Atrophy with Pseudodrusen. <i>Ophthalmology</i> , 2016, 123, 1865-1873.	5.2	13

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19	A dominant mutation in <i>MAPKAPK3</i> , an actor of p38 signaling pathway, causes a new retinal dystrophy involving Bruch's membrane and retinal pigment epithelium. <i>Human Molecular Genetics</i> , 2016, 25, 916-926.	2.9	13
20	Dietary, environmental, and genetic risk factors of Extensive Macular Atrophy with Pseudodrusen, a severe bilateral macular atrophy of middle-aged patients. <i>Scientific Reports</i> , 2018, 8, 6840.	3.3	12
21	Deciphering the natural history of SCA7 in children. <i>European Journal of Neurology</i> , 2020, 27, 2267-2276.	3.3	12
22	CRB1-Related Retinal Dystrophies in a Cohort of 50 Patients: A Reappraisal in the Light of Specific Müller Cell and Photoreceptor CRB1 Isoforms. <i>International Journal of Molecular Sciences</i> , 2021, 22, 12642.	4.1	11
23	Pathogenic variants in <i>IMPG1</i> cause autosomal dominant and autosomal recessive retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2021, 58, 570-578.	3.2	10
24	Melanoma-associated retinopathy during pembrolizumab treatment probably controlled by intravitreal injections of dexamethasone. <i>Documenta Ophthalmologica</i> , 2021, 142, 257-263.	2.2	10
25	Novel TLL5 Variants Associated with Cone-Rod Dystrophy and Early-Onset Severe Retinal Dystrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6410.	4.1	9
26	Pathogenicity of novel atypical variants leading to choroideremia as determined by functional analyses. <i>Human Mutation</i> , 2019, 40, 31-35.	2.5	8
27	<i>CHM</i> mutation spectrum and disease: An update at the time of human therapeutic trials. <i>Human Mutation</i> , 2021, 42, 323-341.	2.5	8
28	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. <i>Human Mutation</i> , 2022, 43, 832-858.	2.5	8
29	Generation of an iPSC line, INMi001-A, carrying the two most common USH2A mutations from a compound heterozygote with non-syndromic retinitis pigmentosa. <i>Stem Cell Research</i> , 2018, 33, 228-232.	0.7	7
30	Generation of a human iPSC line, INMi002-A, carrying the most prevalent USH2A variant associated with Usher syndrome type 2. <i>Stem Cell Research</i> , 2018, 33, 247-250.	0.7	6
31	ALPK1 Gene Mutations Drive Autoinflammation with Ectodermal Dysplasia and Progressive Vision Loss. <i>Journal of Clinical Immunology</i> , 2021, 41, 1671-1673.	3.8	6
32	Characterization of SSBP1-related optic atrophy and foveopathy. <i>Scientific Reports</i> , 2021, 11, 18703.	3.3	6
33	Generation of a human iPSC line, INMi004-A, with a point mutation in CRX associated with autosomal dominant Leber congenital amaurosis. <i>Stem Cell Research</i> , 2019, 38, 101476.	0.7	5
34	Novel roles for voltage-gated type Ca <sup>2+</sup> and Cl <sup>-</sup> channels in phagocytosis and angiogenic factor balance identified in human iPSC-derived RPE. <i>FASEB Journal</i> , 2021, 35, e21406.	0.5	5
35	Martinique Crinkled Retinal Pigment Epitheliopathy. <i>Ophthalmology</i> , 2016, 123, 2196-2204.	5.2	4
36	A Novel Chromosomal Translocation Identified due to Complex Genetic Instability in iPSC Generated for Choroideremia. <i>Cells</i> , 2019, 8, 1068.	4.1	4

#	ARTICLE	IF	CITATIONS
37	Generation of a human iPSC line, INMi003-A, with a missense mutation in CRX associated with autosomal dominant cone-rod dystrophy. <i>Stem Cell Research</i> , 2019, 38, 101478.	0.7	4
38	Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. <i>Mitochondrion</i> , 2021, 59, 169-174.	3.4	3
39	Contribution of Whole-Genome Sequencing and Transcript Analysis to Decipher Retinal Diseases Associated with MFSD8 Variants. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4294.	4.1	3
40	The landscape of submicroscopic structural variants at the <i>OPN1LW/OPN1MW</i> gene cluster on Xq28 underlying blue cone monochromacy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	7.1	2
41	Retinitis Punctata Albescens and RLBP1-Allied Phenotypes. <i>Ophthalmology Science</i> , 2021, 1, 100052.	2.5	1
42	Molecular Therapy for Choroideremia: Pre-clinical and Clinical Progress to Date. <i>Molecular Diagnosis and Therapy</i> , 2021, 25, 661-675.	3.8	1
43	Macular Dystrophies. , 2021, , 1-29.		0
44	Quantification of the early pupillary dilation kinetic to assess rod and cone activity. <i>Scientific Reports</i> , 2021, 11, 9549.	3.3	0
45	Macular Dystrophies. , 2022, , 3967-3995.		0
46	Next-Generation Sequencing Identifies Novel PMPCA Variants in Patients with Late-Onset Dominant Optic Atrophy. <i>Genes</i> , 2022, 13, 1202.	2.4	0