Isabelle Audo

List of Publications by Year
in descending order

104 papers	3,352 citations	218677 26 h-index	182427 51 g-index
107 all docs	107 docs citations	107 times ranked	3346 citing authors

#	Article	IF	Citations
1	Tissueâ€specific genotypeâ€“phenotype correlations among USH2Aâ€related disorders in the RUSH2A study. Human Mutation, 2022, 43, 613-624.	2.5	10
2	Threeâ€Year Safety Results of SAR422459 (EIAVâ€ABCA4) Gene Therapy in Patients With ABCA4â€Associated Stargardt Disease: An Openâ€Label Doseâ€Escalation Phase I/IIa Clinical Trial, Cohorts 1â€5. American Journal of Ophthalmology, 2022, 240, 285-301.	3.3	24
3	The RUSH2A Study: Dark-Adapted Visual Fields in Patients With Retinal Degeneration Associated With Biallelic Variants in the USH2A Gene. , 2022, 63, 17.		7
4	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
5	The research output of rod-cone dystrophy genetics. Orphanet Journal of Rare Diseases, 2022, 17, 175.	2.7	2
6	Macular Dystrophies. , 2022, , 3967-3995.		0
7	A Rare Case of Didanosine-Induced Mid-Peripheral Chorioretinal Atrophy Identified Incidentally 11 Years after the Drug Cessation. Medicina (Lithuania), 2022, 58, 735.	2.0	2
8	Large Benefit from Simple Things: High-Dose Vitamin A Improves RBP4-Related Retinal Dystrophy. International Journal of Molecular Sciences, 2022, 23, 6590.	4.1	4
9	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
10	Retrospective Natural History Study of RPGR-Related Cone- and Cone-Rod Dystrophies While Expanding the Mutation Spectrum of the Disease. International Journal of Molecular Sciences, 2022, 23, 7189.	4.1	7
11	<scp>WDR34</scp>, a candidate gene for nonâ€syndromic rodâ€cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
12	The genetics of rod-cone dystrophy in Arab countries: a systematic review. European Journal of Human Genetics, 2021, 29, 897-910.	2.8	10
13	Congenital Stationary Night Blindness (CSNB): An Inherited Retinal Disorder Where Clear Correlations Can Be Made. Essentials in Ophthalmology, 2021, , 139-152.	0.1	1
14	Macular Dystrophies. , 2021, , 1-29.		0
15	Optic Atrophy and Inner Retinal Thinning in CACNA1F-Related Congenital Stationary Night Blindness. Genes, 2021, 12, 330.	2.4	6
16	CHM mutation spectrum and disease: An update at the time of human therapeutic trials. Human Mutation, 2021, 42, 323-341.	2.5	8
17	Of fluid and tubes. Journal Francais D'Ophtalmologie, 2021, 44, 277-278.	0.4	0
18	Restoration of mGluR6 Localization Following AAV-Mediated Delivery in a Mouse Model of Congenital Stationary Night Blindness. , 2021, 62, 24.		10

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19	A New Mouse Model for Complete Congenital Stationary Night Blindness Due to Gpr179 Deficiency. International Journal of Molecular Sciences, 2021, 22, 4424.	4.1	3
20	DEEP PHENOTYPING AND FURTHER INSIGHTS INTO ITM2B-RELATED RETINAL DYSTROPHY. Retina, 2021, 41, 872-881.	1.7	2
21	Management of a case of Enhanced S-cone syndrome with massive foveoschisis treated with pars plana vitrectomy with silicone oil tamponade. Ophthalmic Genetics, 2021, 42, 615-618.	1.2	3
22	CNGB1 â€related rodâ€cone dystrophy: A mutation review and update. Human Mutation, 2021, 42, 641-666.	2.5	16
23	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
24	Didanosine-induced Retinopathy: New Insights with Long-term Follow-up. Ocular Immunology and Inflammation, 2021, , 1-8.	1.8	3
25	Auditory and olfactory findings in patients with USH2A â€related retinal degenerationâ€"Findings at baseline from the rate of progression in USH2A â€related retinal degeneration natural history study () Tj ETQq1 1 0.784314 rgBT /Ove	1.2	8
26	Mutated CCDC51 Coding for a Mitochondrial Protein, MITOK Is a Candidate Gene Defect for Autosomal Recessive Rod-Cone Dystrophy. International Journal of Molecular Sciences, 2021, 22, 7875.	4.1	3
27	Retinitis Punctata Albescens and RLBP1-Allied Phenotypes. Ophthalmology Science, 2021, 1, 100052.	2.5	1
28	Characteristics of Retinitis Pigmentosa Associated with ADGRV1 and Comparison with USH2A in Patients from a Multicentric Usher Syndrome Study Treatrush. International Journal of Molecular Sciences, 2021, 22, 10352.	4.1	3
29	Assessing Photoreceptor Status in Retinal Dystrophies: From High-Resolution Imaging to Functional Vision. American Journal of Ophthalmology, 2021, 230, 12-47.	3.3	19
30	Deepâ€intronic variants in CNGB3 cause achromatopsia by pseudoexon activation. Human Mutation, 2020, 41, 255-264.	2.5	26
31	PHENOTYPIC CHARACTERISTICS OF RODâ€"CONE DYSTROPHY ASSOCIATED WITH MYO7A MUTATIONS IN A LARGE FRENCH COHORT. Retina, 2020, 40, 1603-1615.	1.7	16
32	Phenotype Driven Analysis of Whole Genome Sequencing Identifies Deep Intronic Variants that Cause Retinal Dystrophies by Aberrant Exonization. , 2020, 61, 36.		17
33	The RUSH2A Study: Best-Corrected Visual Acuity, Full-Field Electroretinography Amplitudes, and Full-Field Stimulus Thresholds at Baseline. Translational Vision Science and Technology, 2020, 9, 9.	2.2	31
34	Impact of the COVID-19 lockdown on basic science research in ophthalmology: the experience of a highly specialized research facility in France. Eye, 2020, 34, 1187-1188.	2.1	15
35	Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276.	3.3	12
36	Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with Neurodevelopmental and Pancreatic Involvement. American Journal of Human Genetics, 2020, 106, 859-871.	6.2	22

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37	Identification and characterization of novel TRPM1 autoantibodies from serum of patients with melanoma-associated retinopathy. PLoS ONE, 2020, 15, e0231750.	2.5	12
38	Baseline Visual Field Findings in the RUSH2A Study: Associated Factors and Correlation With Other Measures of Disease Severity. American Journal of Ophthalmology, 2020, 219, 87-100.	3.3	22
39	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17
40	Multiexon deletion alleles of ATF6 linked to achromatopsia. JCI Insight, 2020, 5, .	5.0	13
41	Prevalence of ABCA4 Deep-Intronic Variants and Related Phenotype in An Unsolved â€œOne-Hitâ€•Cohort with Stargardt Disease. International Journal of Molecular Sciences, 2019, 20, 5053.	4.1	26
42	Outer Retinal Alterations Associated With Visual Outcomes in Best Vitelliform Macular Dystrophy. American Journal of Ophthalmology, 2019, 208, 429-437.	3.3	14
43	An Ashkenazi Jewish founder mutation in CACNA1F causes retinal phenotype in both hemizygous males and heterozygous female carriers. Ophthalmic Genetics, 2019, 40, 443-448.	1.2	8
44	Phenotype Analysis of Retinal Dystrophies in Light of the Underlying Genetic Defects: Application to Cone and Cone-Rod Dystrophies. International Journal of Molecular Sciences, 2019, 20, 4854.	4.1	20
45	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the GNAT2 gene. Human Mutation, 2019, 40, 1145-1155.	2.5	15
46	Neoplasia and intraocular inflammation: From masquerade syndromes to immunotherapy-induced uveitis. Progress in Retinal and Eye Research, 2019, 72, 100761.	15.5	37
47	Longitudinal Clinical Follow-up and Genetic Spectrum of Patients With Rod-Cone Dystrophy Associated With Mutations inPDE6AandPDE6B. JAMA Ophthalmology, 2019, 137, 669.	2.5	32
48	Where are the missing gene defects in inherited retinal disorders? Intronic and synonymous variants contribute at least to 4% of CACNA1F â€•mediated inherited retinal disorders. Human Mutation, 2019, 40, 765-787.	2.5	24
49	Novel Missense Mutations in BEST1 Are Associated with Bestrophinopathies in Lebanese Patients. Genes, 2019, 10, 151.	2.4	7
50	Next Generation Sequencing Identifies Five Novel Mutations in Lebanese Patients with Bardetâ€“Biedl and Usher Syndromes. Genes, 2019, 10, 1047.	2.4	6
51	Peripapillary Sparing With Near Infrared Autofluorescence Correlates With Electroretinographic Findings in Patients With Stargardt Disease. , 2019, 60, 4951.		4
52	Whole exome sequencing resolves complex phenotype and identifies CC2D2A mutations underlying nonâ€•syndromic rodâ€•cone dystrophy. Clinical Genetics, 2019, 95, 329-333.	2.0	19
53	A novel nonsense variant in REEP6 is involved in a sporadic rodâ€•cone dystrophy case. Clinical Genetics, 2018, 93, 707-711.	2.0	7
54	MERTK mutation update in inherited retinal diseases. Human Mutation, 2018, 39, 887-913.	2.5	41

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55	A Novel Heterozygous Missense Mutation inGNAT1Leads to Autosomal Dominant Riggs Type of Congenital Stationary Night Blindness. BioMed Research International, 2018, 2018, 1-10.	1.9	8
56	Expanding the Mutation Spectrum in ABCA4: Sixty Novel Disease Causing Variants and Their Associated Phenotype in a Large French Stargardt Cohort. International Journal of Molecular Sciences, 2018, 19, 2196.	4.1	22
57	Mutations in the gene PDE6C encoding the catalytic subunit of the cone photoreceptor phosphodiesterase in patients with achromatopsia. Human Mutation, 2018, 39, 1366-1371.	2.5	18
58	Phenotypic Characteristics of a French Cohort of Patients with X-Linked Retinoschisis. Ophthalmology, 2018, 125, 1587-1596.	5.2	25
59	Impact of Retinitis Pigmentosa on Quality of Life, Mental Health, and Employment Among Young Adults. American Journal of Ophthalmology, 2017, 177, 169-174.	3.3	50
60	ARL2BP mutations account for 0.1% of autosomal recessive rodâ€cone dystrophies with the report of a novel splice variant. Clinical Genetics, 2017, 92, 109-111.	2.0	7
61	Electrophysiological features and multimodal imaging in ritonavir-related maculopathy. Documenta Ophthalmologica, 2017, 135, 241-248.	2.2	14
62	Further Insights into the Ciliary Gene and Protein KIZ and Its Murine Ortholog PLK1S1 Mutated in Rod-Cone Dystrophy. Genes, 2017, 8, 277.	2.4	7
63	LRIT3 Differentially Affects Connectivity and Synaptic Transmission of Cones to ON- and OFF-Bipolar Cells. , 2017, 58, 1768.		25
64	Testâ€“Retest Variability of Functional and Structural Parameters in Patients with Stargardt Disease Participating in the SAR422459 Gene Therapy Trial. Translational Vision Science and Technology, 2016, 5, 10.	2.2	37
65	Nextâ€generation sequencing confirms the implication of <scp>SLC24A1</scp> in autosomalâ€recessive congenital stationary night blindness. Clinical Genetics, 2016, 89, 690-699.	2.0	16
66	Biallelic Mutations in GNB3 Cause a Unique Form of Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2016, 98, 1011-1019.	6.2	49
67	An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. European Journal of Human Genetics, 2016, 24, 1730-1738.	2.8	77
68	Clinical Characteristics and Risk Factors of Extensive Macular Atrophy with Pseudodrusen. Ophthalmology, 2016, 123, 1865-1873.	5.2	13
69	Martinique Crinkled Retinal Pigment Epitheliopathy. Ophthalmology, 2016, 123, 2196-2204.	5.2	4
70	A new autosomal dominant eye and lung syndrome linked to mutations in TIMP3 gene. Scientific Reports, 2016, 6, 32544.	3.3	17
71	Run of homozygosity analysis reveals a novel nonsense variant of the CNGB1 gene involved in retinitis pigmentosa 45. Ophthalmic Genetics, 2016, 37, 357-359.	1.2	7
72	Identification of a Novel Homozygous Nonsense Mutation Confirms the Implication of GNAT1 in Rod-Cone Dystrophy. PLoS ONE, 2016, 11, e0168271.	2.5	15

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73	Next-generation sequencing applied to a large French cone and cone-rod dystrophy cohort: mutation spectrum and new genotype-phenotype correlation. Orphanet Journal of Rare Diseases, 2015, 10, 85.	2.7	79
74	<scp>LRIT</scp>3 is essential to localize <scp>TRPM</scp>1 to the dendritic tips of depolarizing bipolar cells and may play a role in cone synapse formation. European Journal of Neuroscience, 2015, 42, 1966-1975.	2.6	48
75	Targeted Next Generation Sequencing Identifies Novel Mutations inRP1as a Relatively Common Cause of Autosomal Recessive Rod-Cone Dystrophy. BioMed Research International, 2015, 2015, 1-11.	1.9	25
76	Cone Dystrophy in Patient with HomozygousRP1L1Mutation. BioMed Research International, 2015, 2015, 1-13.	1.9	16
77	Threshold Levels of Visual Field and Acuity Loss Related to Significant Decreases in the Quality of Life and Emotional States of Patients with Retinitis Pigmentosa. Ophthalmic Research, 2015, 54, 78-84.	1.9	22
78	Congenital stationary night blindness: An analysis and update of genotypeâ€“phenotype correlations and pathogenic mechanisms. Progress in Retinal and Eye Research, 2015, 45, 58-110.	15.5	269
79	Spectral-Domain Optical Coherence Tomography in Wagner Syndrome: Characterization of Vitreoretinal Interface and Foveal Changes. American Journal of Ophthalmology, 2015, 160, 1065-1072.e1.	3.3	7
80	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
81	Clinical Characteristics and Current Therapies for Inherited Retinal Degenerations. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017111-a017111.	6.2	171
82	Genotypic and Phenotypic Characterization of P23H Line 1 Rat Model. PLoS ONE, 2015, 10, e0127319.	2.5	51
83	The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. Human Molecular Genetics, 2014, 23, 491-501.	2.9	29
84	Whole-Exome Sequencing Identifies KIZ as a Ciliary Gene Associated with Autosomal-Recessive Rod-Cone Dystrophy. American Journal of Human Genetics, 2014, 94, 625-633.	6.2	52
85	Spectrum of Cav1.4 dysfunction in congenital stationary night blindness type 2. Biochimica Et Biophysica Acta - Biomembranes, 2014, 1838, 2053-2065.	2.6	26
86	Lrit3 Deficient Mouse (nob6): A Novel Model of Complete Congenital Stationary Night Blindness (cCSNB). PLoS ONE, 2014, 9, e90342.	2.5	50
87	Molecular profiling of complete congenital stationary night blindness: a pilot study on an Indian cohort. Molecular Vision, 2014, 20, 341-51.	1.1	14
88	Whole-Exome Sequencing Identifies LRIT3 Mutations as a Cause of Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2013, 92, 67-75.	6.2	120
89	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
90	Further Insights Into GPR179: Expression, Localization, and Associated Pathogenic Mechanisms Leading to Complete Congenital Stationary Night Blindness. , 2013, 54, 8041.		20

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91	Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. Orphanet Journal of Rare Diseases, 2012, 7, 8.	2.7	144
92	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2012, 90, 321-330.	6.2	121
93	RP1 and autosomal dominant rod-cone dystrophy: Novel mutations, a review of published variants, and genotype-phenotype correlation. Human Mutation, 2012, 33, 73-80.	2.5	33
94	Foveal Damage in Habitual Poppers Users. JAMA Ophthalmology, 2011, 129, 703.	2.4	67
95	Novel C2orf71 mutations account for â¼1% of cases in a large French arRP cohort. Human Mutation, 2011, 32, E2091-103.	2.5	29
96	Autofluorescence Imaging in a Case of Benign Familial Fleck Retina. JAMA Ophthalmology, 2011, 125, 714.	2.4	12
97	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
98	EYS is a major gene for rod-cone dystrophies in France. Human Mutation, 2010, 31, E1406-E1435.	2.5	86
99	Spectrum of Rhodopsin Mutations in French Autosomal Dominant Rodâ€“Cone Dystrophy Patients. , 2010, 51, 3687.		45
100	An Unusual Retinal Phenotype Associated With a Novel Mutation in RHO. JAMA Ophthalmology, 2010, 128, 1036.	2.4	24
101	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
102	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2009, 85, 720-729.	6.2	207
103	The Negative ERG: Clinical Phenotypes and Disease Mechanisms of Inner Retinal Dysfunction. Survey of Ophthalmology, 2008, 53, 16-40.	4.0	137
104	Phenotypic Variation in Enhanced S-cone Syndrome. , 2008, 49, 2082.		107