

# Christina Zeitz

## List of Publications by Year in descending order

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136  
papers

4,993  
citations

109321

35  
h-index

118850

62  
g-index

142  
all docs

142  
docs citations

142  
times ranked

4449  
citing authors

#	ARTICLE	IF	CITATIONS
1	Congenital stationary night blindness: An analysis and update of genotype-phenotype correlations and pathogenic mechanisms. <i>Progress in Retinal and Eye Research</i> , 2015, 45, 58-110.	15.5	269
2	The complete form of X-linked congenital stationary night blindness is caused by mutations in a gene encoding a leucine-rich repeat protein. <i>Nature Genetics</i> , 2000, 26, 324-327.	21.4	231
3	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2009, 85, 720-729.	6.2	207
4	NMNAT1 mutations cause Leber congenital amaurosis. <i>Nature Genetics</i> , 2012, 44, 1040-1045.	21.4	171
5	Mutations in CABP4, the Gene Encoding the Ca <sup>2+</sup> -Binding Protein 4, Cause Autosomal Recessive Night Blindness. <i>American Journal of Human Genetics</i> , 2006, 79, 657-667.	6.2	153
6	<i>CRB1</i> mutations in inherited retinal dystrophies. <i>Human Mutation</i> , 2012, 33, 306-315.	2.5	153
7	Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 8.	2.7	144
8	Mutations in GRM6 Cause Autosomal Recessive Congenital Stationary Night Blindness with a Distinctive Scotopic 15-Hz Flicker Electroretinogram. , 2005, 46, 4328.		136
9	Mutations in IFT172 cause isolated retinal degeneration and Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 230-242.	2.9	136
10	Mutation in the Auxiliary Calcium-Channel Subunit CACNA2D4 Causes Autosomal Recessive Cone Dystrophy. <i>American Journal of Human Genetics</i> , 2006, 79, 973-977.	6.2	135
11	The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Studies. <i>Ophthalmology</i> , 2016, 123, 817-828.	5.2	126
12	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2012, 90, 321-330.	6.2	121
13	Whole-Exome Sequencing Identifies LRIT3 Mutations as a Cause of Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2013, 92, 67-75.	6.2	120
14	EYS is a major gene for rod-cone dystrophies in France. <i>Human Mutation</i> , 2010, 31, E1406-E1435.	2.5	86
15	Next-generation sequencing applied to a large French cone and cone-rod dystrophy cohort: mutation spectrum and new genotype-phenotype correlation. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 85.	2.7	79
16	A nationwide genetic analysis of inherited retinal diseases in Israel as assessed by the Israeli inherited retinal disease consortium (IIRDC). <i>Human Mutation</i> , 2020, 41, 140-149.	2.5	75
17	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. <i>American Journal of Human Genetics</i> , 2013, 93, 571-578.	6.2	71
18	Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. <i>Human Mutation</i> , 2010, 31, E1772-E1800.	2.5	69

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19	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
20	Mutation of Solute Carrier SLC16A12 Associates with a Syndrome Combining Juvenile Cataract with Microcornea and Renal Glucosuria. American Journal of Human Genetics, 2008, 82, 772-779.	6.2	66
21	Night blindness-associated mutations in the ligand-binding, cysteine-rich, and intracellular domains of the metabotropic glutamate receptor 6 abolish protein trafficking. Human Mutation, 2007, 28, 771-780.	2.5	60
22	Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). Ophthalmology, 2016, 123, 1887-1897.	5.2	59
23	Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotype-Phenotype Correlations, and Inheritance Models. Genes, 2018, 9, 215.	2.4	58
24	Progression of Stargardt Disease as Determined by Fundus Autofluorescence Over a 12-Month Period. JAMA Ophthalmology, 2019, 137, 1134.	2.5	57
25	Identification and Functional Characterization of a Novel Rhodopsin Mutation Associated with Autosomal Dominant CSNB. , 2008, 49, 4105.		52
26	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
27	Genotypic and Phenotypic Characterization of P23H Line 1 Rat Model. PLoS ONE, 2015, 10, e0127319.	2.5	51
28	Novel mutations in the folliculin gene associated with spontaneous pneumothorax. European Respiratory Journal, 2008, 32, 1316-1320.	6.7	50
29	Lrit3 Deficient Mouse (nob6): A Novel Model of Complete Congenital Stationary Night Blindness (cCSNB). PLoS ONE, 2014, 9, e90342.	2.5	50
30	Prevalence and novelty of PRPF31 mutations in French autosomal dominant rod-cone dystrophy patients and a review of published reports. BMC Medical Genetics, 2010, 11, 145.	2.1	49
31	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
32	<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
33	Identification and characterization of a novel RPCR isoform in human retina. Human Mutation, 2007, 28, 797-807.	2.5	47
34	cDNA subtraction cloning reveals novel genes whose temporal and spatial expression indicates association with trophoblast invasion. Developmental Biology, 2000, 222, 158-169.	2.0	46
35	Spectrum of Rhodopsin Mutations in French Autosomal Dominant Rod-Cone Dystrophy Patients. , 2010, 51, 3687.		45
36	Detailed genetic characteristics of an international large cohort of patients with Stargardt disease: ProgStar study report 8. British Journal of Ophthalmology, 2019, 103, 390-397.	3.9	45

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37	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
38	Molecular genetics and protein function involved in nocturnal vision. <i>Expert Review of Ophthalmology</i> , 2007, 2, 467-485.	0.6	42
39	Molecular Epidemiology in 591 Italian Probands With Nonsyndromic Retinitis Pigmentosa and Usher Syndrome. , 2021, 62, 13.		42
40	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
41	<i>MERTK</i> mutation update in inherited retinal diseases. <i>Human Mutation</i> , 2018, 39, 887-913.	2.5	41
42	Copy-Number Variations in <i>EYS</i> : A Significant Event in the Appearance of arRP. , 2011, 52, 5625.		40
43	Mosaic synaptopathy and functional defects in Cav1.4 heterozygous mice and human carriers of CSNB2. <i>Human Molecular Genetics</i> , 2014, 23, 1538-1550.	2.9	38
44	RP1 and autosomal dominant rod-cone dystrophy: Novel mutations, a review of published variants, and genotype-phenotype correlation. <i>Human Mutation</i> , 2012, 33, 73-80.	2.5	33
45	Longitudinal Clinical Follow-up and Genetic Spectrum of Patients With Rod-Cone Dystrophy Associated With Mutations in <i>PDE6A</i> and <i>PDE6B</i> . <i>JAMA Ophthalmology</i> , 2019, 137, 669.	2.5	32
46	NYX(Nyctalopin on Chromosome X), the Gene Mutated in Congenital Stationary Night Blindness, Encodes a Cell Surface Protein. , 2003, 44, 4184.		31
47	Detailed investigations of proximal tubular function in Imerslund-Gräsbeck syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 111.	2.1	31
48	Novel <i>C2orf71</i> mutations account for ~1% of cases in a large French arRP cohort. <i>Human Mutation</i> , 2011, 32, E2091-103.	2.5	29
49	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. <i>Human Molecular Genetics</i> , 2014, 23, 491-501.	2.9	29
50	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
51	Nyctalopin is essential for synaptic transmission in the cone dominated zebrafish retina. <i>European Journal of Neuroscience</i> , 2006, 24, 1664-1674.	2.6	27
52	A Workshop on Measuring the Progression of Atrophy Secondary to Stargardt Disease in the ProgStar Studies: Findings and Lessons Learned. <i>Translational Vision Science and Technology</i> , 2019, 8, 16.	2.2	27
53	Isolation of the Mouse Nyctalopin GeneNyxand Expression Studies in Mouse and Rat Retina. , 2003, 44, 2260.		26
54	Spectrum of Cav1.4 dysfunction in congenital stationary night blindness type 2. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2014, 1838, 2053-2065.	2.6	26

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55	Prevalence of ABCA4 Deep-Intronic Variants and Related Phenotype in An Unsolved "One-Hit" Cohort with Stargardt Disease. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5053.	4.1	26
56	Novel mutations in CACNA1F and NYX in Dutch families with X-linked congenital stationary night blindness. <i>Molecular Vision</i> , 2005, 11, 179-83.	1.1	26
57	Targeted Next Generation Sequencing Identifies Novel Mutations in <i>RP1</i> as a Relatively Common Cause of Autosomal Recessive Rod-Cone Dystrophy. <i>BioMed Research International</i> , 2015, 2015, 1-11.	1.9	25
58	LRIT3 Differentially Affects Connectivity and Synaptic Transmission of Cones to ON- and OFF-Bipolar Cells. , 2017, 58, 1768.		25
59	Phenotypic Characteristics of a French Cohort of Patients with X-Linked Retinoschisis. <i>Ophthalmology</i> , 2018, 125, 1587-1596.	5.2	25
60	The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 142.	2.7	25
61	An Unusual Retinal Phenotype Associated With a Novel Mutation in RHO. <i>JAMA Ophthalmology</i> , 2010, 128, 1036.	2.4	24
62	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
63	Longitudinal Changes of Fixation Location and Stability Within 12 Months in Stargardt Disease: ProgStar Report No. 12. <i>American Journal of Ophthalmology</i> , 2018, 193, 54-61.	3.3	24
64	Where are the missing gene defects in inherited retinal disorders? Intronic and synonymous variants contribute at least to 4% of <i>CACNA1F</i> -mediated inherited retinal disorders. <i>Human Mutation</i> , 2019, 40, 765-787.	2.5	24
65	Expanding the Mutation Spectrum in ABCA4: Sixty Novel Disease Causing Variants and Their Associated Phenotype in a Large French Stargardt Cohort. <i>International Journal of Molecular Sciences</i> , 2018, 19, 2196.	4.1	22
66	Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with Neurodevelopmental and Pancreatic Involvement. <i>American Journal of Human Genetics</i> , 2020, 106, 859-871.	6.2	22
67	Retinal Phenotype of Patients With Isolated Retinal Degeneration Due to <i>CLN3</i> Pathogenic Variants in a French Retinitis Pigmentosa Cohort. <i>JAMA Ophthalmology</i> , 2021, 139, 278.	2.5	21
68	Further Insights Into GPR179: Expression, Localization, and Associated Pathogenic Mechanisms Leading to Complete Congenital Stationary Night Blindness. , 2013, 54, 8041.		20
69	Phenotype Analysis of Retinal Dystrophies in Light of the Underlying Genetic Defects: Application to Cone and Cone-Rod Dystrophies. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4854.	4.1	20
70	Interplay between cell-adhesion molecules governs synaptic wiring of cone photoreceptors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 23914-23924.	7.1	20
71	Faster Sensitivity Loss around Dense Scotomas than for Overall Macular Sensitivity in Stargardt Disease: ProgStar Report No. 14. <i>American Journal of Ophthalmology</i> , 2020, 216, 219-225.	3.3	20
72	Aripiprazole-induced chorioretinopathy: multimodal imaging and electrophysiological features. <i>Documenta Ophthalmologica</i> , 2015, 131, 35-41.	2.2	19

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73	Whole exome sequencing resolves complex phenotype and identifies <i>CC2D2A</i> mutations underlying non-syndromic rod-cone dystrophy. <i>Clinical Genetics</i> , 2019, 95, 329-333.	2.0	19
74	Assessing Photoreceptor Status in Retinal Dystrophies: From High-Resolution Imaging to Functional Vision. <i>American Journal of Ophthalmology</i> , 2021, 230, 12-47.	3.3	19
75	A common NYX mutation in Flemish patients with X linked CSNB. <i>British Journal of Ophthalmology</i> , 2009, 93, 692-696.	3.9	18
76	A new autosomal dominant eye and lung syndrome linked to mutations in TIMP3 gene. <i>Scientific Reports</i> , 2016, 6, 32544.	3.3	17
77	Phenotype Driven Analysis of Whole Genome Sequencing Identifies Deep Intronic Variants that Cause Retinal Dystrophies by Aberrant Exonization. , 2020, 61, 36.		17
78	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to <i>RPGR</i> Mutations. , 2020, 61, 36.		17
79	A novel DFNB31 mutation associated with Usher type 2 syndrome showing variable degrees of auditory loss in a consanguineous Portuguese family. <i>Molecular Vision</i> , 2011, 17, 1598-606.	1.1	17
80	DNA sequence comparison of human and mouse retinitis pigmentosa GTPase regulator ( <i>RPGR</i> ) identifies tissue-specific exons and putative regulatory elements. <i>Human Genetics</i> , 2001, 109, 271-278.	3.8	16
81	Cone Dystrophy in Patient with Homozygous <i>RP1L1</i> Mutation. <i>BioMed Research International</i> , 2015, 2015, 1-13.	1.9	16
82	Next-generation sequencing confirms the implication of <i>SLC24A1</i> in autosomal recessive congenital stationary night blindness. <i>Clinical Genetics</i> , 2016, 89, 690-699.	2.0	16
83	PHENOTYPIC CHARACTERISTICS OF ROD-CONE DYSTROPHY ASSOCIATED WITH MYO7A MUTATIONS IN A LARGE FRENCH COHORT. <i>Retina</i> , 2020, 40, 1603-1615.	1.7	16
84	<i>CNGB1</i> -related rod-cone dystrophy: A mutation review and update. <i>Human Mutation</i> , 2021, 42, 641-666.	2.5	16
85	Impact of the COVID-19 lockdown on basic science research in ophthalmology: the experience of a highly specialized research facility in France. <i>Eye</i> , 2020, 34, 1187-1188.	2.1	15
86	Identification of a Novel Homozygous Nonsense Mutation Confirms the Implication of GNAT1 in Rod-Cone Dystrophy. <i>PLoS ONE</i> , 2016, 11, e0168271.	2.5	15
87	Disease-Causing Mutations in BEST1 Gene Are Associated with Altered Sorting of Bestrophin-1 Protein. <i>International Journal of Molecular Sciences</i> , 2013, 14, 15121-15140.	4.1	14
88	Outer Retinal Alterations Associated With Visual Outcomes in Best Vitelliform Macular Dystrophy. <i>American Journal of Ophthalmology</i> , 2019, 208, 429-437.	3.3	14
89	TRPM1 Mutations are the Most Common Cause of Autosomal Recessive Congenital Stationary Night Blindness (CSNB) in the Palestinian and Israeli Populations. <i>Scientific Reports</i> , 2019, 9, 12047.	3.3	14
90	Molecular profiling of complete congenital stationary night blindness: a pilot study on an Indian cohort. <i>Molecular Vision</i> , 2014, 20, 341-51.	1.1	14

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91	Identification and characterization of novel TRPM1 autoantibodies from serum of patients with melanoma-associated retinopathy. PLoS ONE, 2020, 15, e0231750.	2.5	12
92	Riggs-type dominant congenital stationary night blindness: ERG findings, a new GNAT1 mutation and a systemic association. Documenta Ophthalmologica, 2018, 137, 57-62.	2.2	11
93	The genetics of rod-cone dystrophy in Arab countries: a systematic review. European Journal of Human Genetics, 2021, 29, 897-910.	2.8	10
94	Restoration of mGluR6 Localization Following AAV-Mediated Delivery in a Mouse Model of Congenital Stationary Night Blindness. , 2021, 62, 24.		10
95	Substantial restoration of night vision in adult mice with congenital stationary night blindness. Molecular Therapy - Methods and Clinical Development, 2021, 22, 15-25.	4.1	10
96	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
97	Novel splice-site mutation in TTLL5 causes cone dystrophy in a consanguineous family. Molecular Vision, 2017, 23, 131-139.	1.1	9
98	A Novel Heterozygous Missense Mutation in <i>GNAT1</i> Leads to Autosomal Dominant Riggs Type of Congenital Stationary Night Blindness. BioMed Research International, 2018, 2018, 1-10.	1.9	8
99	An Ashkenazi Jewish founder mutation in <i>CACNA1F</i> causes retinal phenotype in both hemizygous males and heterozygous female carriers. Ophthalmic Genetics, 2019, 40, 443-448.	1.2	8
100	<i>CHM</i> mutation spectrum and disease: An update at the time of human therapeutic trials. Human Mutation, 2021, 42, 323-341.	2.5	8
101	<i>ARL2BP</i> mutations account for 0.1% of autosomal recessive rod-cone dystrophies with the report of a novel splice variant. Clinical Genetics, 2017, 92, 109-111.	2.0	7
102	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
103	A novel nonsense variant in <i>REEP6</i> is involved in a sporadic rod-cone dystrophy case. Clinical Genetics, 2018, 93, 707-711.	2.0	7
104	Novel Missense Mutations in BEST1 Are Associated with Bestrophinopathies in Lebanese Patients. Genes, 2019, 10, 151.	2.4	7
105	A novel missense mutation of <i>GJA8</i> causes congenital cataract in a large Mauritanian family. European Journal of Ophthalmology, 2019, 29, 621-628.	1.3	7
106	<i>WDR34</i> , a candidate gene for non-syndromic rod-cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
107	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
108	Near-infrared fundus autofluorescence alterations correlate with swept-source optical coherence tomography angiography findings in patients with retinitis pigmentosa. Scientific Reports, 2021, 11, 3180.	3.3	7

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109	Cystoid maculopathy is a frequent feature of Cohen syndrome-associated retinopathy. <i>Scientific Reports</i> , 2021, 11, 16412.	3.3	7
110	Retrospective Natural History Study of RPGR-Related Cone- and Cone-Rod Dystrophies While Expanding the Mutation Spectrum of the Disease. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7189.	4.1	7
111	Next Generation Sequencing Identifies Five Novel Mutations in Lebanese Patients with Bardet-Biedl and Usher Syndromes. <i>Genes</i> , 2019, 10, 1047.	2.4	6
112	AUTOSOMAL DOMINANT VITREORETINOCHOROIODOPATHY. <i>Retina</i> , 2019, 39, 867-878.	1.7	6
113	Optic Atrophy and Inner Retinal Thinning in CACNA1F-Related Congenital Stationary Night Blindness. <i>Genes</i> , 2021, 12, 330.	2.4	6
114	Generation of an induced pluripotent stem cell (iPSC) line from a patient with autosomal dominant retinitis pigmentosa due to a mutation in the NR2E3 gene. <i>Stem Cell Research</i> , 2017, 24, 1-4.	0.7	5
115	Arrayed Primer Extension Microarray for the Analysis of Genes Associated with Congenital Stationary Night Blindness. <i>Methods in Molecular Biology</i> , 2013, 963, 319-326.	0.9	4
116	Retinal findings in a patient of French ancestry with CABP4-related retinal disease. <i>Documenta Ophthalmologica</i> , 2018, 136, 135-143.	2.2	4
117	Generation of human induced pluripotent stem cell lines from a patient with ITM2B-related retinal dystrophy and a non mutated brother. <i>Stem Cell Research</i> , 2019, 41, 101625.	0.7	4
118	Peripapillary Sparing With Near Infrared Autofluorescence Correlates With Electroretinographic Findings in Patients With Stargardt Disease. , 2019, 60, 4951.		4
119	Large Benefit from Simple Things: High-Dose Vitamin A Improves RBP4-Related Retinal Dystrophy. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6590.	4.1	4
120	A New Mouse Model for Complete Congenital Stationary Night Blindness Due to Gpr179 Deficiency. <i>International Journal of Molecular Sciences</i> , 2021, 22, 4424.	4.1	3
121	Management of a case of Enhanced S-cone syndrome with massive foveoschisis treated with pars plana vitrectomy with silicone oil tamponade. <i>Ophthalmic Genetics</i> , 2021, 42, 615-618.	1.2	3
122	Mutated CCDC51 Coding for a Mitochondrial Protein, MITOK Is a Candidate Gene Defect for Autosomal Recessive Rod-Cone Dystrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7875.	4.1	3
123	First identification of ITM2B interactome in the human retina. <i>Scientific Reports</i> , 2021, 11, 17210.	3.3	3
124	Mutation profile of glaucoma candidate genes in Mauritanian families with primary congenital glaucoma. <i>Molecular Vision</i> , 2019, 25, 373-381.	1.1	3
125	Macular sensitivity in patients with congenital stationary night-blindness. <i>British Journal of Ophthalmology</i> , 2019, 103, 1507-1510.	3.9	2
126	Identification of a novel GRM6 mutation in a previously described consanguineous family with complete congenital stationary night blindness. <i>Ophthalmic Genetics</i> , 2019, 40, 182-184.	1.2	2



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127	Disinhibition of intrinsic photosensitive retinal ganglion cells in patients with X-linked congenital stationary night blindness. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2019, 257, 1207-1215.	1.9	2
128	DEEP PHENOTYPING AND FURTHER INSIGHTS INTO ITM2B-RELATED RETINAL DYSTROPHY. <i>Retina</i> , 2021, 41, 872-881.	1.7	2
129	The research output of rod-cone dystrophy genetics. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 175.	2.7	2
130	Congenital Stationary Night Blindness (CSNB): An Inherited Retinal Disorder Where Clear Correlations Can Be Made. <i>Essentials in Ophthalmology</i> , 2021, , 139-152.	0.1	1
131	Congenital stationary night blindness in a patient with mild learning disability due to a compound heterozygous microdeletion of 15q13 and a missense mutation in <i>TRPM1</i> . <i>Ophthalmic Genetics</i> , 2021, 42, 296-299.	1.2	1
132	Challenges of Phenotype-Genotype Correlations in Rare Diseases—Reply. <i>JAMA Ophthalmology</i> , 2021, 139, 1323.	2.5	1
133	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2012, 91, 209.	6.2	0
134	A cone-rod dystrophy patient with a homozygous RP1L1 mutation. <i>Acta Ophthalmologica</i> , 2013, 91, 0-0.	1.1	0
135	Gene therapy for Stargardt disease. <i>Acta Ophthalmologica</i> , 2014, 92, 0-0.	1.1	0
136	Retinal Phenotype of Patients with <i>CLRN1</i> -Associated Usher 3A Syndrome in French Light4Deaf Cohort. , 2022, 63, 25.		0