

Christina Zeitz

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4076174/publications.pdf>

Version: 2024-02-01

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 | The research output of rod-cone dystrophy genetics. Orphanet Journal of Rare Diseases, 2022, 17, 175. | 2.7 | 2 |
| 2 | Retinal Phenotype of Patients with <i>CLRN1</i> -Associated Usher 3A Syndrome in French Light4Deaf Cohort. , 2022, 63, 25. | | 0 |
| 3 | 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 |
| 4 | 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 |
| 5 | <i>WDR34</i> , a candidate gene for non-syndromic rod-cone dystrophy. Clinical Genetics, 2021, 99, 298-302. | 2.0 | 7 |
| 6 | The genetics of rod-cone dystrophy in Arab countries: a systematic review. European Journal of Human Genetics, 2021, 29, 897-910. | 2.8 | 10 |
| 7 | New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176. | 2.5 | 7 |
| 8 | Congenital Stationary Night Blindness (CSNB): An Inherited Retinal Disorder Where Clear Correlations Can Be Made. Essentials in Ophthalmology, 2021, , 139-152. | 0.1 | 1 |
| 9 | 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 |
| 10 | Optic Atrophy and Inner Retinal Thinning in CACNA1F-Related Congenital Stationary Night Blindness. Genes, 2021, 12, 330. | 2.4 | 6 |
| 11 | <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 |
| 12 | Molecular Epidemiology in 591 Italian Probands With Nonsyndromic Retinitis Pigmentosa and Usher Syndrome. , 2021, 62, 13. | | 42 |
| 13 | Retinal Phenotype of Patients With Isolated Retinal Degeneration Due to <i>CLN3</i> Pathogenic Variants in a French Retinitis Pigmentosa Cohort. JAMA Ophthalmology, 2021, 139, 278. | 2.5 | 21 |
| 14 | The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet Journal of Rare Diseases, 2021, 16, 142. | 2.7 | 25 |
| 15 | 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> . Ophthalmic Genetics, 2021, 42, 296-299. | 1.2 | 1 |
| 16 | Restoration of mGluR6 Localization Following AAV-Mediated Delivery in a Mouse Model of Congenital Stationary Night Blindness. , 2021, 62, 24. | | 10 |
| 17 | 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 |
| 18 | DEEP PHENOTYPING AND FURTHER INSIGHTS INTO ITM2B-RELATED RETINAL DYSTROPHY. Retina, 2021, 41, 872-881. | 1.7 | 2 |

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 19 | 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 |
| 20 | <i>CNGB1</i> -related rod-cone dystrophy: A mutation review and update. <i>Human Mutation</i> , 2021, 42, 641-666. | 2.5 | 16 |
| 21 | Novel <i>TLL5</i> 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 |
| 22 | Mutated <i>CCDC51</i> Coding for a Mitochondrial Protein, <i>MITOK</i> Is a Candidate Gene Defect for Autosomal Recessive Rod-Cone Dystrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7875. | 4.1 | 3 |
| 23 | Cystoid maculopathy is a frequent feature of Cohen syndrome-associated retinopathy. <i>Scientific Reports</i> , 2021, 11, 16412. | 3.3 | 7 |
| 24 | First identification of <i>ITM2B</i> interactome in the human retina. <i>Scientific Reports</i> , 2021, 11, 17210. | 3.3 | 3 |
| 25 | Substantial restoration of night vision in adult mice with congenital stationary night blindness. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 22, 15-25. | 4.1 | 10 |
| 26 | 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 |
| 27 | Challenges of Phenotype-Genotype Correlations in Rare Diseases—Reply. <i>JAMA Ophthalmology</i> , 2021, 139, 1323. | 2.5 | 1 |
| 28 | 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 |
| 29 | PHENOTYPIC CHARACTERISTICS OF ROD-CONE DYSTROPHY ASSOCIATED WITH <i>MYO7A</i> MUTATIONS IN A LARGE FRENCH COHORT. <i>Retina</i> , 2020, 40, 1603-1615. | 1.7 | 16 |
| 30 | 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 |
| 31 | Phenotype Driven Analysis of Whole Genome Sequencing Identifies Deep Intronic Variants that Cause Retinal Dystrophies by Aberrant Exonization. , 2020, 61, 36. | | 17 |
| 32 | 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 |
| 33 | Loss of Function of <i>RIMS2</i> 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 |
| 34 | 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 |
| 35 | Identification and characterization of novel <i>TRPM1</i> autoantibodies from serum of patients with melanoma-associated retinopathy. <i>PLoS ONE</i> , 2020, 15, e0231750. | 2.5 | 12 |
| 36 | Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to <i>RPCR</i> Mutations. , 2020, 61, 36. | | 17 |

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 37 | Detailed genetic characteristics of an international large cohort of patients with Stargardt disease: ProgStar study report 8. <i>British Journal of Ophthalmology</i> , 2019, 103, 390-397. | 3.9 | 45 |
| 38 | Progression of Stargardt Disease as Determined by Fundus Autofluorescence Over a 12-Month Period. <i>JAMA Ophthalmology</i> , 2019, 137, 1134. | 2.5 | 57 |
| 39 | Macular sensitivity in patients with congenital stationary night-blindness. <i>British Journal of Ophthalmology</i> , 2019, 103, 1507-1510. | 3.9 | 2 |
| 40 | 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 |
| 41 | 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 |
| 42 | An Ashkenazi Jewish founder mutation in <i>CACNA1F</i> causes retinal phenotype in both hemizygous males and heterozygous female carriers. <i>Ophthalmic Genetics</i> , 2019, 40, 443-448. | 1.2 | 8 |
| 43 | 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 |
| 44 | 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 |
| 45 | Identification of a novel <i>GRM6</i> mutation in a previously described consanguineous family with complete congenital stationary night blindness. <i>Ophthalmic Genetics</i> , 2019, 40, 182-184. | 1.2 | 2 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | 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 |
| 50 | Novel Missense Mutations in BEST1 Are Associated with Bestrophinopathies in Lebanese Patients. <i>Genes</i> , 2019, 10, 151. | 2.4 | 7 |
| 51 | 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 |
| 52 | 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 |
| 53 | Peripapillary Sparing With Near Infrared Autofluorescence Correlates With Electroretinographic Findings in Patients With Stargardt Disease. , 2019, 60, 4951. | | 4 |
| 54 | A novel missense mutation of <i>GJA8</i> causes congenital cataract in a large Mauritanian family. <i>European Journal of Ophthalmology</i> , 2019, 29, 621-628. | 1.3 | 7 |

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 55 | 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 |
| 56 | AUTOSOMAL DOMINANT VITREORETINOCHOROIDOPATHY. <i>Retina</i> , 2019, 39, 867-878. | 1.7 | 6 |
| 57 | Mutation profile of glaucoma candidate genes in Mauritanian families with primary congenital glaucoma. <i>Molecular Vision</i> , 2019, 25, 373-381. | 1.1 | 3 |
| 58 | A novel nonsense variant in <i>REEP6</i> is involved in a sporadic rod-cone dystrophy case. <i>Clinical Genetics</i> , 2018, 93, 707-711. | 2.0 | 7 |
| 59 | <i>MERTK</i> mutation update in inherited retinal diseases. <i>Human Mutation</i> , 2018, 39, 887-913. | 2.5 | 41 |
| 60 | Retinal findings in a patient of French ancestry with CABP4-related retinal disease. <i>Documenta Ophthalmologica</i> , 2018, 136, 135-143. | 2.2 | 4 |
| 61 | A Novel Heterozygous Missense Mutation in <i>GNAT1</i> Leads to Autosomal Dominant Riggs Type of Congenital Stationary Night Blindness. <i>BioMed Research International</i> , 2018, 2018, 1-10. | 1.9 | 8 |
| 62 | 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 |
| 63 | Riggs-type dominant congenital stationary night blindness: ERG findings, a new GNAT1 mutation and a systemic association. <i>Documenta Ophthalmologica</i> , 2018, 137, 57-62. | 2.2 | 11 |
| 64 | Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotype-Phenotype Correlations, and Inheritance Models. <i>Genes</i> , 2018, 9, 215. | 2.4 | 58 |
| 65 | Phenotypic Characteristics of a French Cohort of Patients with X-Linked Retinoschisis. <i>Ophthalmology</i> , 2018, 125, 1587-1596. | 5.2 | 25 |
| 66 | 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 |
| 67 | <i>ARL2BP</i> mutations account for 0.1% of autosomal recessive rod-cone dystrophies with the report of a novel splice variant. <i>Clinical Genetics</i> , 2017, 92, 109-111. | 2.0 | 7 |
| 68 | 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 |
| 69 | 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 |
| 70 | Further Insights into the Ciliary Gene and Protein KIZ and Its Murine Ortholog PLK1S1 Mutated in Rod-Cone Dystrophy. <i>Genes</i> , 2017, 8, 277. | 2.4 | 7 |
| 71 | LRIT3 Differentially Affects Connectivity and Synaptic Transmission of Cones to ON- and OFF-Bipolar Cells. , 2017, 58, 1768. | | 25 |
| 72 | Novel splice-site mutation in TLL5 causes cone dystrophy in a consanguineous family. <i>Molecular Vision</i> , 2017, 23, 131-139. | 1.1 | 9 |

| # | ARTICLE | IF | CITATIONS |
|----|--|------|-----------|
| 73 | 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 |
| 74 | Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease Study (ProgStar Report No. 2). <i>Ophthalmology</i> , 2016, 123, 1887-1897. | 5.2 | 59 |
| 75 | Biallelic Mutations in <i>GNB3</i> Cause a Unique Form of Autosomal-Recessive Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2016, 98, 1011-1019. | 6.2 | 49 |
| 76 | A new autosomal dominant eye and lung syndrome linked to mutations in <i>TIMP3</i> gene. <i>Scientific Reports</i> , 2016, 6, 32544. | 3.3 | 17 |
| 77 | The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Studies. <i>Ophthalmology</i> , 2016, 123, 817-828. | 5.2 | 126 |
| 78 | Identification of a Novel Homozygous Nonsense Mutation Confirms the Implication of <i>GNAT1</i> in Rod-Cone Dystrophy. <i>PLoS ONE</i> , 2016, 11, e0168271. | 2.5 | 15 |
| 79 | 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 |
| 80 | <i>LRIT3</i> is essential to localize <i>TRPM1</i> to the dendritic tips of depolarizing bipolar cells and may play a role in cone synapse formation. <i>European Journal of Neuroscience</i> , 2015, 42, 1966-1975. | 2.6 | 48 |
| 81 | 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 |
| 82 | Cone Dystrophy in Patient with Homozygous <i>RP1L1</i> Mutation. <i>BioMed Research International</i> , 2015, 2015, 1-13. | 1.9 | 16 |
| 83 | High Prevalence of <i>PRPH2</i> 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 |
| 84 | 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 |
| 85 | Aripiprazole-induced chorioretinopathy: multimodal imaging and electrophysiological features. <i>Documenta Ophthalmologica</i> , 2015, 131, 35-41. | 2.2 | 19 |
| 86 | Mutations in <i>IFT172</i> cause isolated retinal degeneration and Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 230-242. | 2.9 | 136 |
| 87 | Genotypic and Phenotypic Characterization of P23H Line 1 Rat Model. <i>PLoS ONE</i> , 2015, 10, e0127319. | 2.5 | 51 |
| 88 | Mosaic synaptopathy and functional defects in <i>Cav1.4</i> heterozygous mice and human carriers of <i>CSNB2</i> . <i>Human Molecular Genetics</i> , 2014, 23, 1538-1550. | 2.9 | 38 |
| 89 | The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies <i>ITM2B</i> 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 |
| 90 | Whole-Exome Sequencing Identifies <i>KIZ</i> as a Ciliary Gene Associated with Autosomal-Recessive Rod-Cone Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 625-633. | 6.2 | 52 |

| # | ARTICLE | IF | CITATIONS |
|-----|---|------|-----------|
| 91 | 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 |
| 92 | Lrit3 Deficient Mouse (nob6): A Novel Model of Complete Congenital Stationary Night Blindness (cCSNB). <i>PLoS ONE</i> , 2014, 9, e90342. | 2.5 | 50 |
| 93 | Gene therapy for Stargardt disease. <i>Acta Ophthalmologica</i> , 2014, 92, 0-0. | 1.1 | 0 |
| 94 | 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 |
| 95 | 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 |
| 96 | Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. <i>American Journal of Human Genetics</i> , 2013, 93, 571-578. | 6.2 | 71 |
| 97 | 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 |
| 98 | 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 |
| 99 | Further Insights Into GPR179: Expression, Localization, and Associated Pathogenic Mechanisms Leading to Complete Congenital Stationary Night Blindness. , 2013, 54, 8041. | | 20 |
| 100 | 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 |
| 101 | Detailed investigations of proximal tubular function in Imerslund-Gräsbeck syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 111. | 2.1 | 31 |
| 102 | A cone-rod dystrophy patient with a homozygous RP1L1 mutation. <i>Acta Ophthalmologica</i> , 2013, 91, 0-0. | 1.1 | 0 |
| 103 | NMNAT1 mutations cause Leber congenital amaurosis. <i>Nature Genetics</i> , 2012, 44, 1040-1045. | 21.4 | 171 |
| 104 | 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 |
| 105 | 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 |
| 106 | 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 |
| 107 | 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 |
| 108 | <i>CRB1</i> mutations in inherited retinal dystrophies. <i>Human Mutation</i> , 2012, 33, 306-315. | 2.5 | 153 |

| # | ARTICLE | IF | CITATIONS |
|-----|---|-----|-----------|
| 109 | Novel <i>C2orf71</i> mutations account for ¼1% of cases in a large French arRP cohort. <i>Human Mutation</i> , 2011, 32, E2091-103. | 2.5 | 29 |
| 110 | Copy-Number Variations in <i>EYS</i> : A Significant Event in the Appearance of arRP. , 2011, 52, 5625. | | 40 |
| 111 | A novel <i>DFNB31</i> 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 |
| 112 | A Mutation in <i>SLC24A1</i> Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2010, 87, 523-531. | 6.2 | 67 |
| 113 | <i>EYS</i> is a major gene for rod-cone dystrophies in France. <i>Human Mutation</i> , 2010, 31, E1406-E1435. | 2.5 | 86 |
| 114 | Mutation spectrum of <i>EYS</i> in Spanish patients with autosomal recessive retinitis pigmentosa. <i>Human Mutation</i> , 2010, 31, E1772-E1800. | 2.5 | 69 |
| 115 | Prevalence and novelty of <i>PRPF31</i> mutations in French autosomal dominant rod-cone dystrophy patients and a review of published reports. <i>BMC Medical Genetics</i> , 2010, 11, 145. | 2.1 | 49 |
| 116 | Spectrum of Rhodopsin Mutations in French Autosomal Dominant Rod-Cone Dystrophy Patients. , 2010, 51, 3687. | | 45 |
| 117 | An Unusual Retinal Phenotype Associated With a Novel Mutation in <i>RHO</i> . <i>JAMA Ophthalmology</i> , 2010, 128, 1036. | 2.4 | 24 |
| 118 | Genotyping Microarray for <i>CSNB</i> -Associated Genes. , 2009, 50, 5919. | | 41 |
| 119 | <i>TRPM1</i> 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 |
| 120 | A common <i>NYX</i> mutation in Flemish patients with X linked <i>CSNB</i> . <i>British Journal of Ophthalmology</i> , 2009, 93, 692-696. | 3.9 | 18 |
| 121 | Mutation of Solute Carrier <i>SLC16A12</i> Associates with a Syndrome Combining Juvenile Cataract with Microcornea and Renal Glucosuria. <i>American Journal of Human Genetics</i> , 2008, 82, 772-779. | 6.2 | 66 |
| 122 | Novel mutations in the folliculin gene associated with spontaneous pneumothorax. <i>European Respiratory Journal</i> , 2008, 32, 1316-1320. | 6.7 | 50 |
| 123 | Identification and Functional Characterization of a Novel Rhodopsin Mutation Associated with Autosomal Dominant <i>CSNB</i> . , 2008, 49, 4105. | | 52 |
| 124 | Molecular genetics and protein function involved in nocturnal vision. <i>Expert Review of Ophthalmology</i> , 2007, 2, 467-485. | 0.6 | 42 |
| 125 | Night blindness-associated mutations in the ligand-binding, cysteine-rich, and intracellular domains of the metabotropic glutamate receptor 6 abolish protein trafficking. <i>Human Mutation</i> , 2007, 28, 771-780. | 2.5 | 60 |
| 126 | Identification and characterization of a novel <i>RPGR</i> isoform in human retina. <i>Human Mutation</i> , 2007, 28, 797-807. | 2.5 | 47 |

| # | ARTICLE | IF | CITATIONS |
|-----|--|------|-----------|
| 127 | Mutations in CABP4, the Gene Encoding the Ca ²⁺ -Binding Protein 4, Cause Autosomal Recessive Night Blindness. <i>American Journal of Human Genetics</i> , 2006, 79, 657-667. | 6.2 | 153 |
| 128 | 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 |
| 129 | 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 |
| 130 | Mutations in GRM6 Cause Autosomal Recessive Congenital Stationary Night Blindness with a Distinctive Scotopic 15-Hz Flicker Electroretinogram. , 2005, 46, 4328. | | 136 |
| 131 | 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 |
| 132 | NYX(Nyctalopin on Chromosome X), the Gene Mutated in Congenital Stationary Night Blindness, Encodes a Cell Surface Protein. , 2003, 44, 4184. | | 31 |
| 133 | Isolation of the Mouse Nyctalopin Gene Nyx and Expression Studies in Mouse and Rat Retina. , 2003, 44, 2260. | | 26 |
| 134 | DNA sequence comparison of human and mouse retinitis pigmentosa GTPase regulator (RPGR) identifies tissue-specific exons and putative regulatory elements. <i>Human Genetics</i> , 2001, 109, 271-278. | 3.8 | 16 |
| 135 | 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 |
| 136 | cDNA subtraction cloning reveals novel genes whose temporal and spatial expression indicates association with trophoblast invasion. <i>Developmental Biology</i> , 2000, 222, 158-169. | 2.0 | 46 |