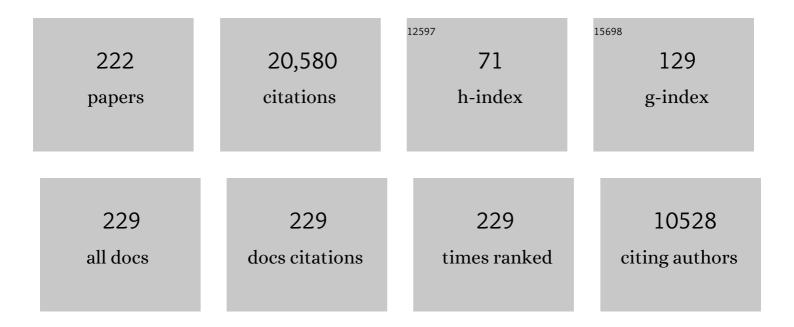
## Artur V Cideciyan

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3361883/publications.pdf Version: 2024-02-01



ADTILD V CIDECIVAN

| #  | Article   | IF   | CITATIONS |
|----|---|------|-----------|
| 1  | Longitudinal Changes of Fixation Stability and Location Within 24 Months in Stargardt Disease:<br>ProgStar Report No. 16. American Journal of Ophthalmology, 2022, 233, 78-89.  | 1.7  | 5         |
| 2  | Full-field stimulus testing: Role in the clinic and as an outcome measure in clinical trials of severe childhood retinal disease. Progress in Retinal and Eye Research, 2022, 87, 101000.   | 7.3  | 28        |
| 3  | Longitudinal Changes in Scotopic and Mesopic Macular Function as Assessed with Microperimetry in<br>Patients With Stargardt Disease: SMART Study Report No. 2. American Journal of Ophthalmology, 2022,<br>236, 32-44.                | 1.7  | 2         |
| 4  | Intravitreal antisense oligonucleotide sepofarsen in Leber congenital amaurosis type 10: a phase 1b/2<br>trial. Nature Medicine, 2022, 28, 1014-1021.   | 15.2 | 46        |
| 5  | Restoration of Cone Sensitivity to Individuals with Congenital Photoreceptor Blindness within the Phase 1/2 Sepofarsen Trial. Ophthalmology Science, 2022, 2, 100133.   | 1.0  | 5         |
| 6  | Mobility test to assess functional vision in dark-adapted patients with Leber congenital amaurosis.<br>BMC Ophthalmology, 2022, 22, .   | 0.6  | 4         |
| 7  | The landscape of submicroscopic structural variants at the <i>OPN1LW/OPN1MW</i> gene cluster on Xq28 underlying blue cone monochromacy. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, . | 3.3  | 2         |
| 8  | Leber Congenital Amaurosis Due to GUCY2D Mutations: Longitudinal Analysis of Retinal Structure and<br>Visual Function. International Journal of Molecular Sciences, 2021, 22, 2031.   | 1.8  | 7         |
| 9  | Durable vision improvement after a single treatment with antisense oligonucleotide sepofarsen: a<br>case report. Nature Medicine, 2021, 27, 785-789.  | 15.2 | 41        |
| 10 | The Progression of Stargardt Disease Using Volumetric Hill of Vision Analyses Over 24 Months:<br>ProgStar Report No.15. American Journal of Ophthalmology, 2021, 230, 123-133.  | 1.7  | 10        |
| 11 | Safety and improved efficacy signals following gene therapy in childhood blindness caused by GUCY2D mutations. IScience, 2021, 24, 102409.  | 1.9  | 22        |
| 12 | A Novel ARL3 Gene Mutation Associated With Autosomal Dominant Retinal Degeneration. Frontiers in<br>Cell and Developmental Biology, 2021, 9, 720782.  | 1.8  | 13        |
| 13 | Gene therapy reforms photoreceptor structure and restores vision in NPHP5-associated Leber congenital amaurosis. Molecular Therapy, 2021, 29, 2456-2468.  | 3.7  | 18        |
| 14 | Macular Rod Function in Retinitis Pigmentosa Measured With Scotopic Microperimetry. Translational<br>Vision Science and Technology, 2021, 10, 3.  | 1.1  | 5         |
| 15 | Measures of Function and Structure to Determine Phenotypic Features, Natural History, and<br>Treatment Outcomes in Inherited Retinal Diseases. Annual Review of Vision Science, 2021, 7, 747-772.                                     | 2.3  | 14        |
| 16 | Childhood-onset genetic cone-rod photoreceptor diseases and underlying pathobiology.<br>EBioMedicine, 2021, 63, 103200.   | 2.7  | 4         |
| 17 | RPGR isoform imbalance causes ciliary defects due to exon ORF15 mutations in X-linked retinitis pigmentosa (XLRP). Human Molecular Genetics, 2021, 29, 3706-3716.   | 1.4  | 16        |
| 18 | Long-Term Structural Outcomes of Late-Stage RPE65 Gene Therapy. Molecular Therapy, 2020, 28,<br>266-278.  | 3.7  | 56        |

| #  | Article  | IF   | CITATIONS |
|----|--|------|-----------|
| 19 | Toxicity and Efficacy Evaluation of an Adeno-Associated Virus Vector Expressing Codon-Optimized<br><i>RPGR</i> Delivered by Subretinal Injection in a Canine Model of X-linked Retinitis Pigmentosa.<br>Human Gene Therapy, 2020, 31, 253-267. | 1.4  | 22        |
| 20 | Progress in treating inherited retinal diseases: Early subretinal gene therapy clinical trials and candidates for future initiatives. Progress in Retinal and Eye Research, 2020, 77, 100827.  | 7.3  | 133       |
| 21 | Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second<br>Monaciano Symposium. Translational Vision Science and Technology, 2020, 9, 2.   | 1.1  | 56        |
| 22 | Rod function deficit in retained photoreceptors of patients with class B Rhodopsin mutations.<br>Scientific Reports, 2020, 10, 12552.  | 1.6  | 10        |
| 23 | Foveal Therapy in Blue Cone Monochromacy: Predictions of Visual Potential From Artificial<br>Intelligence. Frontiers in Neuroscience, 2020, 14, 800.   | 1.4  | 7         |
| 24 | Dose Range Finding Studies with Two RPGR Transgenes in a Canine Model of X-Linked Retinitis<br>Pigmentosa Treated with Subretinal Gene Therapy. Human Gene Therapy, 2020, 31, 743-755.   | 1.4  | 15        |
| 25 | The Effect of Attention on Fixation Stability During Dynamic Fixation Testing in Stargardt Disease.<br>American Journal of Ophthalmology, 2020, 217, 305-316.  | 1.7  | 6         |
| 26 | Faster Sensitivity Loss around Dense Scotomas than for Overall Macular Sensitivity in Stargardt<br>Disease: ProgStar Report No. 14. American Journal of Ophthalmology, 2020, 216, 219-225.   | 1.7  | 20        |
| 27 | Transient pupillary light reflex in CEP290- or NPHP5-associated Leber congenital amaurosis: Latency as a potential outcome measure of cone function. Vision Research, 2020, 168, 53-63.  | 0.7  | 14        |
| 28 | Reading Performance in Blue Cone Monochromacy: Defining an Outcome Measure for a Clinical Trial.<br>Translational Vision Science and Technology, 2020, 9, 13.  | 1.1  | 5         |
| 29 | Detailed genetic characteristics of an international large cohort of patients with Stargardt disease:<br>ProgStar study report 8. British Journal of Ophthalmology, 2019, 103, 390-397.  | 2.1  | 45        |
| 30 | Progression of Stargardt Disease as Determined by Fundus Autofluorescence Over a 12-Month Period.<br>JAMA Ophthalmology, 2019, 137, 1134.  | 1.4  | 57        |
| 31 | Treatment Potential for Macular Cone Vision in Leber Congenital Amaurosis Due<br>to <i>CEP290</i> or <i>NPHP5</i> Mutations: Predictions From Artificial Intelligence. , 2019, 60, 2551.   |      | 27        |
| 32 | Short-Wavelength Sensitive Cone (S-cone) Testing as an Outcome Measure for NR2E3 Clinical Treatment Trials. International Journal of Molecular Sciences, 2019, 20, 2497.   | 1.8  | 13        |
| 33 | Leber Congenital Amaurosis (LCA): Potential for Improvement of Vision. , 2019, 60, 1680.   |      | 50        |
| 34 | Autosomal Dominant Retinitis Pigmentosa Due to Class B Rhodopsin Mutations: An Objective Outcome<br>for Future Treatment Trials. International Journal of Molecular Sciences, 2019, 20, 5344.  | 1.8  | 11        |
| 35 | Effect of an intravitreal antisense oligonucleotide on vision in Leber congenital amaurosis due to a photoreceptor cilium defect. Nature Medicine, 2019, 25, 225-228.  | 15.2 | 177       |
| 36 | A G86R mutation in the calcium-sensor protein GCAP1 alters regulation of retinal guanylyl cyclase and causes dominant cone-rod degeneration. Journal of Biological Chemistry, 2019, 294, 3476-3488.  | 1.6  | 29        |

| #  | Article  | IF  | CITATIONS |
|----|--|-----|-----------|
| 37 | <i>BEST1</i> gene therapy corrects a diffuse retina-wide microdetachment modulated by light<br>exposure. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115,<br>E2839-E2848. | 3.3 | 62        |
| 38 | Expanded Retinal Disease Spectrum Associated With Autosomal Recessive Mutations in GUCY2D.<br>American Journal of Ophthalmology, 2018, 190, 58-68.   | 1.7 | 20        |
| 39 | Blue Cone Monochromacy Caused by the C203R Missense Mutation or Large Deletion Mutations. , 2018, 59, 5762.  |     | 21        |
| 40 | Translational Retinal Research and Therapies. Translational Vision Science and Technology, 2018, 7, 8.   | 1.1 | 11        |
| 41 | Progression in X-linked Retinitis Pigmentosa Due to <i>ORF15-RPGR</i> Mutations: Assessment of Localized Vision Changes Over 2 Years. , 2018, 59, 4558.  |     | 17        |
| 42 | Efficacy Outcome Measures for Clinical Trials of USH2A Caused by the Common c.2299delG Mutation.<br>American Journal of Ophthalmology, 2018, 193, 114-129.   | 1.7 | 19        |
| 43 | Cone Vision Changes in the Enhanced S-Cone Syndrome Caused by <i>NR2E3</i> Gene Mutations. , 2018, 59, 3209.   |     | 19        |
| 44 | Mutation-independent rhodopsin gene therapy by knockdown and replacement with a single AAV<br>vector. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115,<br>E8547-E8556.    | 3.3 | 114       |
| 45 | Longitudinal Changes of Fixation Location and Stability Within 12 Months in Stargardt Disease:<br>ProgStar Report No. 12. American Journal of Ophthalmology, 2018, 193, 54-61.                                       | 1.7 | 24        |
| 46 | Visual Acuity Change Over 24 Months and Its Association With Foveal Phenotype and Genotype in<br>Individuals With Stargardt Disease. JAMA Ophthalmology, 2018, 136, 920.   | 1.4 | 44        |
| 47 | Human Melanopic Circuit in Isolation from Photoreceptor Input:Light Sensitivity and Temporal Profile. Journal of Vision, 2018, 18, 1347.   | 0.1 | 0         |
| 48 | Fixation Location and Stability Using the MP-1 Microperimeter in Stargardt Disease. Ophthalmology Retina, 2017, 1, 68-76.  | 1.2 | 37        |
| 49 | Defining Outcomes for Clinical Trials of Leber Congenital Amaurosis Caused by GUCY2D Mutations.<br>American Journal of Ophthalmology, 2017, 177, 44-57.  | 1.7 | 29        |
| 50 | Macular Sensitivity Measured With Microperimetry in Stargardt Disease in the Progression of<br>Atrophy Secondary to Stargardt Disease (ProgStar) Study. JAMA Ophthalmology, 2017, 135, 696.                          | 1.4 | 60        |
| 51 | Optimization of Retinal Gene Therapy for X-Linked Retinitis Pigmentosa Due to RPGR Mutations.<br>Molecular Therapy, 2017, 25, 1866-1880.   | 3.7 | 60        |
| 52 | Visual Acuity Change over 12 Months in the Prospective Progression of Atrophy Secondary to<br>Stargardt Disease (ProgStar) Study. Ophthalmology, 2017, 124, 1640-1651.   | 2.5 | 43        |
| 53 | Incidence of Atrophic Lesions in Stargardt Disease in the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Study. JAMA Ophthalmology, 2017, 135, 687.  | 1.4 | 47        |
| 54 | REEP6 mediates trafficking of a subset of Clathrin-coated vesicles and is critical for rod photoreceptor function and survival. Human Molecular Genetics, 2017, 26, 2218-2230.                                       | 1.4 | 23        |

| #  | Article  | IF  | CITATIONS |
|----|--|-----|-----------|
| 55 | Novel pathogenic mutations in C1QTNF5 support a dominant negative disease mechanism in late-onset retinal degeneration. Scientific Reports, 2017, 7, 12147.  | 1.6 | 30        |
| 56 | Progression of Stargardt Disease as Determined by Fundus Autofluorescence in the Retrospective<br>Progression of Stargardt Disease Study (ProgStar Report No. 9). JAMA Ophthalmology, 2017, 135, 1232. | 1.4 | 77        |
| 57 | Progression of Visual Acuity and Fundus Autofluorescence in Recent-Onset Stargardt Disease:<br>ProgStar Study Report #4. Ophthalmology Retina, 2017, 1, 514-523.                                       | 1.2 | 28        |
| 58 | InÂVitro Modeling Using Ciliopathy-Patient-Derived Cells Reveals Distinct Cilia Dysfunctions Caused by CEP290 Mutations. Cell Reports, 2017, 20, 384-396.  | 2.9 | 120       |
| 59 | EYS Mutations Causing Autosomal Recessive Retinitis Pigmentosa: Changes of Retinal Structure and Function with Disease Progression. Genes, 2017, 8, 178.   | 1.0 | 35        |
| 60 | Towards Treatment of Stargardt Disease: Workshop Organized and Sponsored by the Foundation<br>Fighting Blindness. Translational Vision Science and Technology, 2017, 6, 6.                             | 1.1 | 44        |
| 61 | Outcome Measures for Clinical Trials of Leber Congenital Amaurosis Caused by the Intronic Mutation in the <i>CEP290</i> Gene. , 2017, 58, 2609.  |     | 46        |
| 62 | Pupillary Light Reflexes in Severe Photoreceptor Blindness Isolate the Melanopic Component of Intrinsically Photosensitive Retinal Ganglion Cells. , 2017, 58, 3215.                                   |     | 13        |
| 63 | Imaging Lenticular Autofluorescence in Older Subjects. , 2017, 58, 4940.   |     | 9         |
| 64 | Postretinal Structure and Function in Severe Congenital Photoreceptor Blindness Caused by<br>Mutations in the GUCY2D Gene. , 2017, 58, 959.  |     | 16        |
| 65 | Complexity of the Class B Phenotype in Autosomal Dominant Retinitis Pigmentosa Due to Rhodopsin<br>Mutations. , 2016, 57, 4847.  |     | 30        |
| 66 | Visual Function and Central Retinal Structure in Choroideremia. , 2016, 57, OCT377.  |     | 65        |
| 67 | Developing an Outcome Measure With High Luminance for Optogenetics Treatment of Severe Retinal Degenerations and for Gene Therapy of Cone Diseases. , 2016, 57, 3211.                                  |     | 18        |
| 68 | Outer Retinal Changes Including the Ellipsoid Zone Band in Usher Syndrome 1B due toMYO7AMutations. , 2016, 57, OCT253.   |     | 26        |
| 69 | Retinal Structure Measurements as Inclusion Criteria for Stem Cell–Based Therapies of Retinal Degenerations. , 2016, 57, ORSFn1.   |     | 13        |
| 70 | Automated Light- and Dark-Adapted Perimetry for Evaluating Retinitis Pigmentosa: Filling a Need to<br>Accommodate Multicenter Clinical Trials. , 2016, 57, 3118.                                       |     | 40        |
| 71 | Patterns of Individual Variation in Visual Pathway Structure and Function in the Sighted and Blind.<br>PLoS ONE, 2016, 11, e0164677.   | 1.1 | 38        |
| 72 | Comparison of Short-Wavelength Reduced-Illuminance and Conventional Autofluorescence Imaging<br>in Stargardt Macular Dystrophy. American Journal of Ophthalmology, 2016, 168, 269-278.                 | 1.7 | 29        |

| #  | Article   | IF   | CITATIONS |
|----|---|------|-----------|
| 73 | Visual Acuity Loss and Associated Risk Factors in the Retrospective Progression of Stargardt Disease<br>Study (ProgStar Report No. 2). Ophthalmology, 2016, 123, 1887-1897.   | 2.5  | 59        |
| 74 | Overlap of abnormal photoreceptor development and progressive degeneration in Leber congenital amaurosis caused by <i>NPHP5</i> mutation. Human Molecular Genetics, 2016, 25, 4211-4226.  | 1.4  | 35        |
| 75 | Five-Year Safety and Performance Results from the Argus II Retinal Prosthesis System Clinical Trial.<br>Ophthalmology, 2016, 123, 2248-2254.  | 2.5  | 281       |
| 76 | Variegated yet non-random rod and cone photoreceptor disease patterns<br>in <i>RPGR-ORF15</i> -associated retinal degeneration. Human Molecular Genetics, 2016, 25, 5444-5459.  | 1.4  | 35        |
| 77 | <i>SPATA7</i> : Evolving phenotype from cone-rod dystrophy to retinitis pigmentosa. Ophthalmic Genetics, 2016, 37, 333-338.   | 0.5  | 17        |
| 78 | The Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar)<br>Studies. Ophthalmology, 2016, 123, 817-828.  | 2.5  | 126       |
| 79 | Leber Congenital Amaurosis: Genotypes and Retinal Structure Phenotypes. Advances in Experimental<br>Medicine and Biology, 2016, 854, 169-175.   | 0.8  | 27        |
| 80 | Autofluorescence Imaging With Near-Infrared Excitation: Normalization by Reflectance to Reduce<br>Signal From Choroidal Fluorophores. , 2015, 56, 3393.   |      | 48        |
| 81 | Molecular Heterogeneity Within the Clinical Diagnosis of Pericentral Retinal Degeneration. , 2015, 56, 6007.  |      | 20        |
| 82 | Predicting Progression of <i>ABCA4</i> -Associated Retinal Degenerations Based on Longitudinal<br>Measurements of the Leading Disease Front. , 2015, 56, 5946.  |      | 36        |
| 83 | Pseudo-Fovea Formation After Gene Therapy for RPE65-LCA. Investigative Ophthalmology and Visual Science, 2015, 56, 526-537.   | 3.3  | 39        |
| 84 | Genetics and Disease Expression in the CNGA3 Form of Achromatopsia. Ophthalmology, 2015, 122, 997-1007.   | 2.5  | 61        |
| 85 | Protein misfolding and the pathogenesis of ABCA4-associated retinal degenerations. Human<br>Molecular Genetics, 2015, 24, 3220-3237.  | 1.4  | 69        |
| 86 | Improvement in vision: a new goal for treatment of hereditary retinal degenerations. Expert Opinion on Orphan Drugs, 2015, 3, 563-575.  | 0.5  | 23        |
| 87 | Long-Term Results from an Epiretinal Prosthesis to Restore Sight to the Blind. Ophthalmology, 2015, 122, 1547-1554.   | 2.5  | 224       |
| 88 | Improvement and Decline in Vision with Gene Therapy in Childhood Blindness. New England Journal of<br>Medicine, 2015, 372, 1920-1926.   | 13.9 | 333       |
| 89 | Outcome measure for the treatment of cone photoreceptor diseases: orientation to a scene with cone-only contrast. BMC Ophthalmology, 2015, 15, 98.  | 0.6  | 4         |
| 90 | Successful arrest of photoreceptor and vision loss expands the therapeutic window of retinal gene<br>therapy to later stages of disease. Proceedings of the National Academy of Sciences of the United<br>States of America, 2015, 112, E5844-53. | 3.3  | 75        |

| #   | Article   | IF  | CITATIONS |
|-----|---|-----|-----------|
| 91  | Gene Augmentation for X-Linked Retinitis Pigmentosa Caused by Mutations in RPGR. Cold Spring Harbor<br>Perspectives in Medicine, 2015, 5, a017392-a017392.  | 2.9 | 19        |
| 92  | Drusen and Photoreceptor Abnormalities in African-Americans with Intermediate Non-neovascular<br>Age-related Macular Degeneration. Current Eye Research, 2015, 40, 398-406.   | 0.7 | 15        |
| 93  | Blue Cone Monochromacy: Visual Function and Efficacy Outcome Measures for Clinical Trials. PLoS<br>ONE, 2015, 10, e0125700.   | 1.1 | 29        |
| 94  | Canine Retina Has a Primate Fovea-Like Bouquet of Cone Photoreceptors Which Is Affected by Inherited<br>Macular Degenerations. PLoS ONE, 2014, 9, e90390.   | 1.1 | 100       |
| 95  | Natural History of Cone Disease in the Murine Model of Leber Congenital Amaurosis Due to CEP290<br>Mutation: Determining the Timing and Expectation of Therapy. PLoS ONE, 2014, 9, e92928.  | 1.1 | 23        |
| 96  | <i>TULP1</i> Mutations Causing Early-Onset Retinal Degeneration: Preserved but Insensitive Macular<br>Cones. , 2014, 55, 5354.  |     | 47        |
| 97  | Late-Onset Retinal Degeneration Caused byC1QTNF5Mutation. JAMA Ophthalmology, 2014, 132, 1252.  | 1.4 | 21        |
| 98  | Inner and Outer Retinal Changes in Retinal Degenerations Associated With <i>ABCA4</i> Mutations. , 2014, 55, 1810.  |     | 48        |
| 99  | Human retinal gene therapy for Leber congenital amaurosis shows advancing retinal degeneration<br>despite enduring visual improvement. Proceedings of the National Academy of Sciences of the United<br>States of America, 2013, 110, E517-25.                                    | 3.3 | 401       |
| 100 | Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human<br>Molecular Genetics, 2013, 22, 5136-5145.  | 1.4 | 159       |
| 101 | Reply to Townes-Anderson: <i>RPE65</i> gene therapy does not alter the natural history of retinal degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E1706.  | 3.3 | 4         |
| 102 | Determining consequences of retinal membrane guanylyl cyclase (RetGC1) deficiency in human Leber congenital amaurosis en route to therapy: residual cone-photoreceptor vision correlates with biochemical properties of the mutants. Human Molecular Genetics, 2013, 22, 168-183. | 1.4 | 89        |
| 103 | Human Cone Visual Pigment Deletions Spare Sufficient Photoreceptors to Warrant Gene Therapy.<br>Human Gene Therapy, 2013, 24, 993-1006.   | 1.4 | 97        |
| 104 | Retinal optogenetic therapies: clinical criteria for candidacy. Clinical Genetics, 2013, 84, 175-182.   | 1.0 | 40        |
| 105 | Intervisit Variability of Visual Parameters in Leber Congenital Amaurosis Caused<br>by <i>RPE65</i> Mutations. , 2013, 54, 1378.  |     | 24        |
| 106 | Abnormal Thickening as well as Thinning of the Photoreceptor Layer in Intermediate Age-Related<br>Macular Degeneration. , 2013, 54, 1603.   |     | 77        |
| 107 | Gene Therapy for Retinitis Pigmentosa Caused by <i>MFRP</i> Mutations: Human Phenotype and Preliminary Proof of Concept. Human Gene Therapy, 2012, 23, 367-376.   | 1.4 | 35        |
| 108 | Gene therapy rescues photoreceptor blindness in dogs and paves the way for treating human X-linked retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 2132-2137.   | 3.3 | 237       |

| #   | Article  | IF   | CITATIONS |
|-----|--|------|-----------|
| 109 | Gene Therapy for Leber Congenital Amaurosis Caused by RPE65 Mutations. JAMA Ophthalmology, 2012, 130, 9.   | 2.6  | 580       |
| 110 | Interim Results from the International Trial of Second Sight's Visual Prosthesis. Ophthalmology, 2012, 119, 779-788.   | 2.5  | 675       |
| 111 | Macular Function in Macular Degenerations: Repeatability of Microperimetry as a Potential Outcome<br>Measure for <i>ABCA4</i> -Associated Retinopathy Trials. , 2012, 53, 841.   |      | 105       |
| 112 | <i>RPGR-</i> Associated Retinal Degeneration in Human X-Linked RP and a Murine Model. , 2012, 53, 5594.  |      | 56        |
| 113 | Retinal Disease Course in Usher Syndrome 1B Due to <i>MYO7A</i> Mutations. , 2011, 52, 7924.   |      | 68        |
| 114 | Autosomal Recessive Retinitis Pigmentosa Caused by Mutations in the <i>MAK</i> Gene. , 2011, 52, 9665.   |      | 39        |
| 115 | Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken<br>Syndrome. JAMA Ophthalmology, 2011, 129, 81.  | 2.6  | 62        |
| 116 | A Missense Mutation in DHDDS, Encoding Dehydrodolichyl Diphosphate Synthase, Is Associated with<br>Autosomal-Recessive Retinitis Pigmentosa in Ashkenazi Jews. American Journal of Human Genetics, 2011,<br>88, 207-215.                         | 2.6  | 120       |
| 117 | Human Retinal Disease from <i>AIPL1</i> Gene Mutations: Foveal Cone Loss with Minimal Macular<br>Photoreceptors and Rod Function Remaining. , 2011, 52, 70.  |      | 59        |
| 118 | Human <i>CRB1</i> -Associated Retinal Degeneration: Comparison with the <i>rd8 Crb1</i> -Mutant<br>Mouse Model. , 2011, 52, 6898.  |      | 98        |
| 119 | Probing Mechanisms of Photoreceptor Degeneration in a New Mouse Model of the Common Form of<br>Autosomal Dominant Retinitis Pigmentosa due to P23H Opsin Mutations. Journal of Biological<br>Chemistry, 2011, 286, 10551-10567.                  | 1.6  | 221       |
| 120 | Defective photoreceptor phagocytosis in a mouse model of enhanced S one syndrome causes progressive retinal degeneration. FASEB Journal, 2011, 25, 3157-3176.  | 0.2  | 76        |
| 121 | Cone photoreceptors are the main targets for gene therapy of NPHP5 (IQCB1) or NPHP6 (CEP290) blindness: generation of an all-cone Nphp6 hypomorph mouse that mimics the human retinal ciliopathy. Human Molecular Genetics, 2011, 20, 1411-1423. | 1.4  | 115       |
| 122 | Leber congenital amaurosis due to RPE65 mutations and its treatment with gene therapy. Progress in<br>Retinal and Eye Research, 2010, 29, 398-427.   | 7.3  | 219       |
| 123 | Normal Central Retinal Function and Structure Preserved in Retinitis Pigmentosa. , 2010, 51, 1079.   |      | 81        |
| 124 | Retinal Disease in Rpe65-Deficient Mice: Comparison to Human Leber Congenital Amaurosis Due<br>to <i>RPE65</i> Mutations. , 2010, 51, 5304.  |      | 27        |
| 125 | Molecular Anthropology Meets Genetic Medicine to Treat Blindness in the North African Jewish<br>Population: Human Gene Therapy Initiated in Israel. Human Gene Therapy, 2010, 21, 1749-1757.   | 1.4  | 65        |
| 126 | Treatment Possibilities for Retinitis Pigmentosa. New England Journal of Medicine, 2010, 363, 1669-1671.   | 13.9 | 71        |

| #   | ARTICLE   | IF   | CITATIONS |
|-----|---|------|-----------|
| 127 | Defining the Residual Vision in Leber Congenital Amaurosis Caused by <i>RPE65</i> Mutations. , 2009, 50, 2368.  |      | 109       |
| 128 | Disease Boundaries in the Retina of Patients with Usher Syndrome Caused by <i>MYO7A</i> Gene Mutations. , 2009, 50, 1886.   |      | 83        |
| 129 | Retinal Pigment Epithelium Defects in Humans and Mice with Mutations in <i>MYO7A</i> : Imaging Melanosome-Specific Autofluorescence. , 2009, 50, 4386.  |      | 75        |
| 130 | Loss of cone photoreceptors caused by chromophore depletion is partially prevented by the artificial chromophore pro-drug, 9-cis-retinyl acetate. Human Molecular Genetics, 2009, 18, 2277-2287.                                | 1.4  | 77        |
| 131 | ABCA4 disease progression and a proposed strategy for gene therapy. Human Molecular Genetics, 2009, 18, 931-941.  | 1.4  | 163       |
| 132 | CERKLMutations Cause an Autosomal Recessive Cone-Rod Dystrophy with Inner Retinopathy. , 2009, 50, 5944.  |      | 83        |
| 133 | Genetic heterogeneity in autosomal dominant retinitis pigmentosa with low-frequency damped electroretinographic wavelets. Eye, 2009, 23, 230-233.   | 1.1  | 15        |
| 134 | Vision 1 Year after Gene Therapy for Leber's Congenital Amaurosis. New England Journal of Medicine, 2009, 361, 725-727.   | 13.9 | 197       |
| 135 | Human <i>RPE65</i> Gene Therapy for Leber Congenital Amaurosis: Persistence of Early Visual<br>Improvements and Safety at 1 Year. Human Gene Therapy, 2009, 20, 999-1004.   | 1.4  | 305       |
| 136 | Leber congenital amaurosis caused by Lebercilin (LCA5) mutation: retained photoreceptors adjacent to retinal disorganization. Molecular Vision, 2009, 15, 1098-106.   | 1.1  | 26        |
| 137 | ABCA4 gene analysis in patients with autosomal recessive cone and cone rod dystrophies. European<br>Journal of Human Genetics, 2008, 16, 812-819.   | 1.4  | 54        |
| 138 | Usher syndromes due to MYO7A, PCDH15, USH2A or GPR98 mutations share retinal disease mechanism.<br>Human Molecular Genetics, 2008, 17, 2405-2415.   | 1.4  | 106       |
| 139 | Human gene therapy for RPE65 isomerase deficiency activates the retinoid cycle of vision but with slow rod kinetics. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 15112-15117.   | 3.3  | 639       |
| 140 | Treatment of Leber Congenital Amaurosis Due to <i>RPE65</i> Mutations by Ocular Subretinal Injection<br>of Adeno-Associated Virus Gene Vector: Short-Term Results of a Phase I Trial. Human Gene Therapy,<br>2008, 19, 979-990. | 1.4  | 880       |
| 141 | Retinal Laminar Architecture in Human Retinitis Pigmentosa Caused by <i>Rhodopsin</i> Gene<br>Mutations. , 2008, 49, 1580.  |      | 118       |
| 142 | Retinal Disease in Usher Syndrome III Caused by Mutations in the Clarin-1 Gene. , 2008, 49, 2651.   |      | 75        |
| 143 | Photoreceptor Layer Topography in Children with Leber Congenital Amaurosis Caused by <i>RPE65</i> Mutations. , 2008, 49, 4573.  |      | 86        |
| 144 | Phase I Trial of Leber Congenital Amaurosis due to RPE65 Mutations by Ocular Subretinal Injection of<br>Adeno-Associated Virus Gene Vector: Short-Term Results. Human Gene Therapy, 2008, .                                     | 1.4  | 13        |

| #   | Article   | IF  | CITATIONS |
|-----|---|-----|-----------|
| 145 | Human cone photoreceptor dependence on RPE65 isomerase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15123-15128.  | 3.3 | 135       |
| 146 | Evidence for retinal remodelling in retinitis pigmentosa caused by PDE6B mutation. British Journal of Ophthalmology, 2007, 91, 699-701.   | 2.1 | 35        |
| 147 | Full-field stimulus testing (FST) to quantify visual perception in severely blind candidates for treatment trials. Physiological Measurement, 2007, 28, N51-N56.  | 1.2 | 92        |
| 148 | Leber Congenital Amaurosis Caused by an RPGRIP1 Mutation Shows Treatment Potential.<br>Ophthalmology, 2007, 114, 895-898.   | 2.5 | 44        |
| 149 | Reduced-illuminance autofluorescence imaging in ABCA4-associated retinal degenerations. Journal of the Optical Society of America A: Optics and Image Science, and Vision, 2007, 24, 1457.                                  | 0.8 | 131       |
| 150 | Canine and Human Visual Cortex Intact and Responsive Despite Early Retinal Blindness from RPE65<br>Mutation. PLoS Medicine, 2007, 4, e230.  | 3.9 | 107       |
| 151 | RDH12andRPE65, Visual Cycle Genes Causing Leber Congenital Amaurosis, Differ in Disease Expression. , 2007, 48, 332.  |     | 66        |
| 152 | Macular Pigment and Lutein Supplementation inABCA4-Associated Retinal Degenerations. , 2007, 48, 1319.  |     | 63        |
| 153 | Inner Retinal Abnormalities in X-linked Retinitis Pigmentosa with <i>RPGR</i> Mutations. , 2007, 48, 4759.  |     | 107       |
| 154 | Centrosomal-ciliary geneCEP290/NPHP6 mutations result in blindness with unexpected sparing of photoreceptors and visual brain: implications for therapy of Leber congenital amaurosis. Human Mutation, 2007, 28, 1074-1083. | 1.1 | 148       |
| 155 | Electroretinographic analyses of Rpe65-mutant rd12 mice: developing an in vivo bioassay for human gene therapy trials of Leber congenital amaurosis. Molecular Vision, 2007, 13, 1701-10.                                   | 1.1 | 28        |
| 156 | Lentiviral Expression of Retinal Guanylate Cyclase-1 (RetGC1) Restores Vision in an Avian Model of<br>Childhood Blindness. PLoS Medicine, 2006, 3, e201.  | 3.9 | 80        |
| 157 | Safety in Nonhuman Primates of Ocular AAV2-RPE65, a Candidate Treatment for Blindness in Leber<br>Congenital Amaurosis. Human Gene Therapy, 2006, 17, 845-858.  | 1.4 | 142       |
| 158 | Retinal Disease Expression in Bardet-Biedl Syndrome-1 (BBS1) Is a Spectrum from Maculopathy to Retina-Wide Degeneration. , 2006, 47, 5004.  |     | 83        |
| 159 | In vivo function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. Human Molecular Genetics, 2006, 15, 2588-2602.   | 1.4 | 113       |
| 160 | Remodeling of the Human Retina in Choroideremia: Rab Escort Protein 1 (REP-1) Mutations. , 2006, 47, 4113.  |     | 156       |
| 161 | Safety of Recombinant Adeno-Associated Virus Type 2–RPE65 Vector Delivered by Ocular Subretinal<br>Injection. Molecular Therapy, 2006, 13, 1074-1084.   | 3.7 | 196       |
| 162 | 881. Safety, Efficacy and Biodistribution of Recombinant AAV2-RPE65 Vector Delivered by Ocular<br>Subretinal Injection. Molecular Therapy, 2006, 13, S339.  | 3.7 | 0         |

| #   | Article   | IF  | CITATIONS |
|-----|---|-----|-----------|
| 163 | Biochemical Characterisation of the C1QTNF5 Gene Associated with Late-Onset Retinal Degeneration. , 2006, 572, 41-48.   |     | 10        |
| 164 | Safety in Nonhuman Primates of Ocular AAV2-RPE65, a Candidate Treatment for Blindness in Leber<br>Congenital Amaurosis. Human Gene Therapy, 2006, .   | 1.4 | 0         |
| 165 | OUTCOME MEASURES AND THEIR APPLICATION IN CLINICAL TRIALS FOR RETINAL DEGENERATIVE DISEASES.<br>Retina, 2005, 25, 772-777.  | 1.0 | 37        |
| 166 | In vivo dynamics of retinal injury and repair in the rhodopsin mutant dog model of human retinitis<br>pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2005,<br>102, 5233-5238. | 3.3 | 183       |
| 167 | ABCA4-Associated Retinal Degenerations Spare Structure and Function of the Human Parapapillary Retina. , 2005, 46, 4739.  |     | 117       |
| 168 | Long-Term Restoration of Rod and Cone Vision by Single Dose rAAV-Mediated Gene Transfer to the Retina in a Canine Model of Childhood Blindness. Molecular Therapy, 2005, 12, 1072-1082.                                     | 3.7 | 421       |
| 169 | Nonhuman Primate Models for Diabetic Ocular Neovascularization Using AAV2-Mediated<br>Overexpression of Vascular Endothelial Growth Factor. Diabetes, 2005, 54, 1141-1149.  | 0.3 | 64        |
| 170 | Disease Expression in Usher Syndrome Caused byVLGR1Gene Mutation (USH2C) and Comparison withUSH2APhenotype. , 2005, 46, 734.  |     | 60        |
| 171 | Identifying photoreceptors in blind eyes caused by RPE65 mutations: Prerequisite for human gene therapy success. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 6177-6182.     | 3.3 | 249       |
| 172 | Quantifying rod photoreceptor-mediated vision in retinal degenerations: dark-adapted thresholds as outcome measures. Experimental Eye Research, 2005, 80, 259-272.  | 1.2 | 145       |
| 173 | Mutations in ABCA4 result in accumulation of lipofuscin before slowing of the retinoid cycle: a reappraisal of the human disease sequence. Human Molecular Genetics, 2004, 13, 525-534.                                     | 1.4 | 231       |
| 174 | Impairment of the Transient Pupillary Light Reflex inRpe65â^'/â^'Mice and Humans with Leber Congenital<br>Amaurosis. , 2004, 45, 1259.  |     | 92        |
| 175 | Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. Human Molecular Genetics, 2004, 13, 1893-1902.  | 1.4 | 94        |
| 176 | Lifespan and mitochondrial control of neurodegeneration. Nature Genetics, 2004, 36, 1153-1158.  | 9.4 | 106       |
| 177 | Mutation analysis ofNR2E3 andNRL genes in Enhanced S Cone Syndrome. Human Mutation, 2004, 24, 439-439.  | 1.1 | 92        |
| 178 | In Utero Gene Therapy Rescues Vision in a Murine Model of Congenital Blindness. Molecular Therapy,<br>2004, 9, 182-188.   | 3.7 | 191       |
| 179 | Crumbs homolog 1 (CRB1) mutations result in a thick human retina with abnormal lamination. Human<br>Molecular Genetics, 2003, 12, 1073-1078.  | 1.4 | 205       |
| 180 | In vivo micropathology of Best macular dystrophy with optical coherence tomography. Experimental<br>Eye Research, 2003, 76, 203-211.  | 1.2 | 68        |

| #   | Article  | IF  | CITATIONS |
|-----|--|-----|-----------|
| 181 | Clinicopathologic effects of mutant GUCY2D in Leber congenital amaurosis. Ophthalmology, 2003, 110, 549-558.   | 2.5 | 50        |
| 182 | Mutation in a short-chain collagen gene, CTRP5 , results in extracellular deposit formation in<br>late-onset retinal degeneration: a genetic model for age-related macular degeneration. Human<br>Molecular Genetics, 2003, 12, 2657-2667.               | 1.4 | 172       |
| 183 | Cone Deactivation Kinetics and GRK1/GRK7 Expression in Enhanced S Cone Syndrome Caused by Mutations inNR2E3. , 2003, 44, 1268.   |     | 37        |
| 184 | De Novo Mutation in theRP1Gene (Arg677ter) Associated with Retinitis Pigmentosa. , 2003, 44, 3593.   |     | 32        |
| 185 | Naturally occurring rhodopsin mutation in the dog causes retinal dysfunction and degeneration<br>mimicking human dominant retinitis pigmentosa. Proceedings of the National Academy of Sciences of<br>the United States of America, 2002, 99, 6328-6333. | 3.3 | 150       |
| 186 | The nuclear receptor NR2E3 plays a role in human retinal photoreceptor differentiation and degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 473-478.   | 3.3 | 218       |
| 187 | Macular Pigment and Lutein Supplementation in Choroideremia. Experimental Eye Research, 2002, 74, 371-381.   | 1.2 | 96        |
| 188 | Spinocerebellar Ataxia Type 7 (SCA7) Shows a Cone–Rod Dystrophy Phenotype. Experimental Eye<br>Research, 2002, 74, 737-745.  | 1.2 | 94        |
| 189 | Early age-related maculopathy and self-reported visual difficulty in daily life2 2The authors have no commercial interests in any device or product mentioned in this article Ophthalmology, 2002, 109, 1235-1242.                                       | 2.5 | 151       |
| 190 | Novel Mutation in the TIMP3 Gene Causes Sorsby Fundus Dystrophy. JAMA Ophthalmology, 2002, 120, 376.   | 2.6 | 47        |
| 191 | Augmented rod bipolar cell function in partial receptor loss: an ERG study in P23H rhodopsin transgenic and aging normal rats. Vision Research, 2001, 41, 2779-2797.   | 0.7 | 91        |
| 192 | Melatonin delays photoreceptor degeneration in the rds/rds mouse. NeuroReport, 2001, 12, 1011-1014.  | 0.6 | 54        |
| 193 | Title is missing!. Nature Genetics, 2001, 28, 92-95.   | 9.4 | 132       |
| 194 | Gene therapy restores vision in a canine model of childhood blindness. Nature Genetics, 2001, 28, 92-95.   | 9.4 | 1,130     |
| 195 | [41] In vivo assessment of photoreceptor function in human diseases caused by photoreceptor-specific gene mutations. Methods in Enzymology, 2000, 316, 611-626.  | 0.4 | 17        |
| 196 | Rod and cone visual cycle consequences of a null mutation in the 11-cis-retinol dehydrogenase gene in man. Visual Neuroscience, 2000, 17, 667-678.   | 0.5 | 99        |
| 197 | Mutation of a nuclear receptor gene, NR2E3, causes enhanced S cone syndrome, a disorder of retinal cell fate. Nature Genetics, 2000, 24, 127-131.  | 9.4 | 439       |
| 198 | Rapid restoration of visual pigment and function with oral retinoid in a mouse model of childhood<br>blindness. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97,<br>8623-8628.                                 | 3.3 | 292       |

| #   | Article   | IF  | CITATIONS |
|-----|---|-----|-----------|
| 199 | Optical Coherence Tomography (OCT) Abnormalities in rhodopsin Mutant Transgenic Swine with<br>Retinal Degeneration. Experimental Eye Research, 2000, 70, 247-251.   | 1.2 | 37        |
| 200 | Pitfalls in Homozygosity Mapping. American Journal of Human Genetics, 2000, 67, 1348-1351.  | 2.6 | 72        |
| 201 | Dominant late-onset retinal degeneration with regional variation of sub-retinal pigment epithelium deposits, retinal function, and photoreceptor degeneration. Ophthalmology, 2000, 107, 2256-2266.                                       | 2.5 | 83        |
| 202 | A Nonsense Mutation in a Novel Gene Is Associated With Retinitis Pigmentosa in a Family Linked to the RP1 Locus. Human Molecular Genetics, 1999, 8, 1541-1546.  | 1.4 | 68        |
| 203 | Stable transgene expression in rod photoreceptors after recombinant adeno-associated virus-mediated gene transfer to monkey retina. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 9920-9925. | 3.3 | 226       |
| 204 | Retinal Rod Photoreceptor–Specific Gene Mutation Perturbs Cone Pathway Development. Neuron, 1999, 23, 549-557.  | 3.8 | 75        |
| 205 | Comparative study of mammalian phototransduction in vivo: a prelude to preclinical treatment trials in retinal degenerations. , 1999, , .   |     | 0         |
| 206 | A Novel Locus (RP24) for X-linked Retinitis Pigmentosa Maps to Xq26-27. American Journal of Human<br>Genetics, 1998, 63, 1439-1447.   | 2.6 | 65        |
| 207 | Disease sequence from mutant rhodopsin allele to rod and cone photoreceptor degeneration in man.<br>Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 7103-7108.                                 | 3.3 | 251       |
| 208 | Null mutation in the rhodopsin kinase gene slows recovery kinetics of rod and cone<br>phototransduction in man. Proceedings of the National Academy of Sciences of the United States of<br>America, 1998, 95, 328-333.                    | 3.3 | 172       |
| 209 | Genetically engineered large animal model for studying cone photoreceptor survival and degeneration in retinitis pigmentosa. Nature Biotechnology, 1997, 15, 965-970.   | 9.4 | 247       |
| 210 | Sites of disease action in a retinal dystrophy with supernormal and delayed rod electroretinogramb-waves. Vision Research, 1996, 36, 889-901.   | 0.7 | 44        |
| 211 | An Alternative Phototransduction Model for Human Rod and Cone ERG a -waves: Normal Parameters and Variation with Age. Vision Research, 1996, 36, 2609-2621.   | 0.7 | 116       |
| 212 | The Enhanced S Cone Syndrome: An Analysis of Receptoral and Post-receptoral Changes. Vision Research, 1996, 36, 3711-3722.  | 0.7 | 38        |
| 213 | Preferential Rod and Cone Photoreceptor Abnormalities in Heterozygotes with Point Mutations in theRDSGene. Experimental Eye Research, 1996, 63, 603-608.  | 1.2 | 20        |
| 214 | Photoreceptor function in heterozygotes with insertion or deletion mutations in the RDS gene.<br>Investigative Ophthalmology and Visual Science, 1996, 37, 1662-74.   | 3.3 | 43        |
| 215 | Night blindness in Sorsby's fundus dystrophy reversed by vitamin A. Nature Genetics, 1995, 11, 27-32.   | 9.4 | 197       |
| 216 | Automated imaging dark adaptometer for investigating hereditary retinal degenerations. , 1995, , .  |     | 2         |

| #   | Article  | IF  | CITATIONS |
|-----|--|-----|-----------|
| 217 | Enhanced S cone syndrome: Evidence for an abnormally large number of S cones. Vision Research, 1995, 35, 1473-1481.                                    | 0.7 | 98        |
| 218 | A Heterozygous Putative Null Mutation in ROM1 without a Mutation in Peripherin/RDS in a Family with Retinitis Pigmentosa. Genomics, 1995, 27, 384-386. | 1.3 | 37        |
| 219 | Pattern of Retinal Dysfunction in Acute Zonal Occult Outer Retinopathy. Ophthalmology, 1995, 102, 1187-1198.   | 2.5 | 119       |
| 220 | <title>Registration of high-resolution images of the retina</title> . , 1992, 1652, 310.   |     | 32        |
| 221 | Microcomputer-assisted determination of regional myocardial function. Medical and Biological Engineering and Computing, 1990, 28, 591-594.             | 1.6 | 0         |
| 222 | Intravitreal Sepofarsen for Leber Congenital Amaurosis Type 10 (LCA10). SSRN Electronic Journal, 0, , .  | 0.4 | 0         |