

Colin Eric Willoughby

List of Publications by Year in descending order

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Version: 2024-02-01

99
papers

4,664
citations

147566

31
h-index

118652

62
g-index

106
all docs

106
docs citations

106
times ranked

6268
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in smooth muscle α -actin (ACTA2) lead to thoracic aortic aneurysms and dissections. <i>Nature Genetics</i> , 2007, 39, 1488-1493.	9.4	767
2	Missense Mutations in GJB2 Encoding Connexin-26 Cause the Ectodermal Dysplasia Keratitis-Ichthyosis-Deafness Syndrome. <i>American Journal of Human Genetics</i> , 2002, 70, 1341-1348.	2.6	345
3	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. <i>Nature Genetics</i> , 2013, 45, 155-163.	9.4	269
4	Mutation Altering the miR-184 Seed Region Causes Familial Keratoconus with Cataract. <i>American Journal of Human Genetics</i> , 2011, 89, 628-633.	2.6	234
5	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	4.6	215
6	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. <i>Nature Genetics</i> , 2020, 52, 160-166.	9.4	192
7	Anatomy and physiology of the human eye: effects of mucopolysaccharidoses disease on structure and function – a review. <i>Clinical and Experimental Ophthalmology</i> , 2010, 38, 2-11.	1.3	127
8	Association of Polymorphisms in the Hepatocyte Growth Factor Gene Promoter with Keratoconus. , 2011, 52, 8514.		114
9	Keratoconus in 18 pairs of twins. <i>Acta Ophthalmologica</i> , 2012, 90, e482-6.	0.6	102
10	Limbal Stem Cell Deficiency and Ocular Phenotype in Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome Caused by p63 Mutations. <i>Ophthalmology</i> , 2012, 119, 74-83.	2.5	94
11	Mitochondrial dysfunction in glaucoma: Understanding genetic influences. <i>Mitochondrion</i> , 2012, 12, 202-212.	1.6	85
12	Next-generation sequencing-based molecular diagnosis of 82 retinitis pigmentosa probands from Northern Ireland. <i>Human Genetics</i> , 2015, 134, 217-230.	1.8	85
13	Molecular diagnosis for heterogeneous genetic diseases with targeted high-throughput DNA sequencing applied to retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2011, 48, 145-151.	1.5	81
14	In Vivo Early Corneal Biomechanical Changes After Corneal Cross-linking in Patients With Progressive Keratoconus. <i>Journal of Refractive Surgery</i> , 2017, 33, 840-846.	1.1	79
15	Mutational Analysis of <i>MIR184</i> in Sporadic Keratoconus and Myopia. , 2013, 54, 5266.		73
16	Mitochondrial dysfunction and oxidative stress in corneal disease. <i>Mitochondrion</i> , 2017, 36, 103-113.	1.6	73
17	Corneal biomechanics and biomechanically corrected intraocular pressure in primary open-angle glaucoma, ocular hypertension and controls. <i>British Journal of Ophthalmology</i> , 2020, 104, 121-126.	2.1	67
18	Mutational Spectrum of the <i>ZEB1</i> Gene in Corneal Dystrophies Supports a Genotype-Phenotype Correlation. , 2013, 54, 3215.		65

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19	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. <i>Nature Communications</i> , 2018, 9, 1864.	5.8	63
20	Simplifying Collection of Corneal Specimens in Cases of Suspected Bacterial Keratitis. <i>Journal of Clinical Microbiology</i> , 2003, 41, 3192-3197.	1.8	59
21	Comparison of preservation and transportation protocols for preloaded Descemet membrane endothelial keratoplasty. <i>British Journal of Ophthalmology</i> , 2018, 102, 549-555.	2.1	58
22	Enrichment of pathogenic alleles in the brittle cornea gene, ZNF469, in keratoconus. <i>Human Molecular Genetics</i> , 2014, 23, 5527-5535.	1.4	56
23	Genetic Analysis of $\hat{PAX6}$ -Negative™ Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016, 11, e0153757.	1.1	54
24	Corneal Angiography for Guiding and Evaluating Fine-Needle Diathermy Treatment of Corneal Neovascularization. <i>Ophthalmology</i> , 2015, 122, 1079-1084.	2.5	53
25	Defining the Pathogenicity of Optineurin in Juvenile Open-Angle Glaucoma. , 2004, 45, 3122.		52
26	Imaging of Corneal Neovascularization: Optical Coherence Tomography Angiography and Fluorescence Angiography. , 2018, 59, 1263.		47
27	Lensectomy in the management of glaucoma in spherophakia. <i>Journal of Cataract and Refractive Surgery</i> , 2002, 28, 1061-1064.	0.7	45
28	Improving precision for detecting change in the shape of the cornea in patients with keratoconus. <i>Scientific Reports</i> , 2018, 8, 12345.	1.6	45
29	Development of a Diagnostic Genetic Test for Simplex and Autosomal Recessive Retinitis Pigmentosa. <i>Ophthalmology</i> , 2010, 117, 2169-2177.e3.	2.5	42
30	Preparation of ultrathin grafts for Descemet-stripping endothelial keratoplasty with a single microkeratome pass. <i>Journal of Cataract and Refractive Surgery</i> , 2017, 43, 12-15.	0.7	42
31	CRYBB1 mutation associated with congenital cataract and microcornea. <i>Molecular Vision</i> , 2005, 11, 587-93.	1.1	41
32	Whole-mitochondrial genome sequencing in primary open-angle glaucoma using massively parallel sequencing identifies novel and known pathogenic variants. <i>Genetics in Medicine</i> , 2015, 17, 279-284.	1.1	38
33	The Relationship Between Mechanical Properties, Ultrastructural Changes, and Intrafibrillar Bond Formation in Corneal UVA/Riboflavin Cross-linking Treatment for Keratoconus. <i>Journal of Refractive Surgery</i> , 2018, 34, 264-272.	1.1	38
34	A COL17A1 Splice-Altering Mutation Is Prevalent in Inherited Recurrent Corneal Erosions. <i>Ophthalmology</i> , 2016, 123, 709-722.	2.5	37
35	An Intraocular Pressure Polygenic Risk Score Stratifies Multiple Primary Open-Angle Glaucoma Parameters Including Treatment Intensity. <i>Ophthalmology</i> , 2020, 127, 901-907.	2.5	37
36	Influence of graft size on graft survival following Descemet stripping automated endothelial keratoplasty. <i>British Journal of Ophthalmology</i> , 2015, 99, 784-788.	2.1	35

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37	Association of Genetic Variation With Keratoconus. <i>JAMA Ophthalmology</i> , 2020, 138, 174.	1.4	34
38	Gene-based antiangiogenic applications for corneal neovascularization. <i>Survey of Ophthalmology</i> , 2018, 63, 193-213.	1.7	33
39	Biomaterials for corneal endothelial cell culture and tissue engineering. <i>Journal of Tissue Engineering</i> , 2021, 12, 204173142199053.	2.3	32
40	High resolution corneal and single pulse imaging with line field spectral domain optical coherence tomography. <i>Optics Express</i> , 2016, 24, 12395.	1.7	31
41	Accurate, fast, data efficient and interpretable glaucoma diagnosis with automated spatial analysis of the whole cup to disc profile. <i>PLoS ONE</i> , 2019, 14, e0209409.	1.1	27
42	Replication of the Recessive STBMS1 Locus but with Dominant Inheritance. , 2009, 50, 3210.		25
43	Angiographic and In Vivo Confocal Microscopic Characterization of Human Corneal Blood and Presumed Lymphatic Neovascularization. <i>Cornea</i> , 2015, 34, 1459-1465.	0.9	25
44	Bandage contact lens and topical steroids are risk factors for the development of microbial keratitis after epithelium-off CXL. <i>BMJ Open Ophthalmology</i> , 2019, 4, e000231.	0.8	25
45	Marginal Corneal Vascular Arcades. , 2013, 54, 7470.		22
46	Deformation velocity imaging using optical coherence tomography and its applications to the cornea. <i>Biomedical Optics Express</i> , 2017, 8, 5579.	1.5	22
47	Targeting the NLRP3 Inflammasome in Glaucoma. <i>Biomolecules</i> , 2021, 11, 1239.	1.8	22
48	Cross-Country Transportation Efficacy and Clinical Outcomes of Preloaded Large-Diameter Ultra-Thin Descemet Stripping Automated Endothelial Keratoplasty Grafts. <i>Cornea</i> , 2019, 38, 30-34.	0.9	20
49	Consensus Recommendation for Mouse Models of Ocular Hypertension to Study Aqueous Humor Outflow and Its Mechanisms. , 2022, 63, 12.		20
50	A novel de novo missense mutation in <i>TP63</i> underlying germline mosaicism in AEC syndrome: Implications for recurrence risk and prenatal diagnosis. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1957-1961.	0.7	19
51	Reliability of the Effect of Artificial Anterior Chamber Pressure and Corneal Drying on Corneal Graft Thickness. <i>Cornea</i> , 2015, 34, 866-869.	0.9	18
52	Method for Angiographically Guided Fine-Needle Diathermy in the Treatment of Corneal Neovascularization. <i>Cornea</i> , 2016, 35, 1029-1032.	0.9	18
53	Correction of Mutant p63 in EEC Syndrome Using siRNA Mediated Allele-Specific Silencing Restores Defective Stem Cell Function. <i>Stem Cells</i> , 2016, 34, 1588-1600.	1.4	17
54	Phenotypic heterogeneity and mutational spectrum in a cohort of 45 Italian males subjects with X-linked ectodermal dysplasia. <i>Clinical Genetics</i> , 2015, 87, 338-342.	1.0	16

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55	Infantile bilateral glaucoma in a child with ectodermal dysplasia. <i>Ophthalmic Genetics</i> , 2013, 34, 58-60.	0.5	15
56	Femtosecond Laser-Assisted Lamellar Keratectomy for Corneal Opacities Secondary to Anterior Corneal Dystrophies. <i>Cornea</i> , 2016, 35, 6-13.	0.9	15
57	Prognostic value of miR-21 for prostate cancer: a systematic review and meta-analysis. <i>Bioscience Reports</i> , 2022, 42, .	1.1	15
58	Monozygotic twins discordant for phacomatosis pigmentovascularis: Evidence for the concept of twin spotting. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 718-720.	0.7	14
59	Mutational Analysis of the <i>Rhodopsin</i> Gene in Sector Retinitis Pigmentosa. <i>Ophthalmic Genetics</i> , 2015, 36, 239-243.	0.5	14
60	The Importance of Screening for Sight-Threatening Retinopathy in Incontinentia Pigmenti. <i>Pediatric Dermatology</i> , 2004, 21, 242-245.	0.5	13
61	Corneal endothelial dysfunction in Pearson syndrome. <i>Ophthalmic Genetics</i> , 2013, 34, 55-57.	0.5	13
62	Common Mutations in Arg304 of the p63 Gene in Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome: Lack of Genotype-Phenotype Correlation and Implications for Mutation Detection Strategies. <i>Journal of Investigative Dermatology</i> , 2002, 119, 1202-1203.	0.3	12
63	Iris Flocculi as an Ocular Marker of <i>ACTA2</i> Mutation in Familial Thoracic Aortic Aneurysms and Dissections. <i>Ophthalmic Genetics</i> , 2015, 36, 86-88.	0.5	12
64	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. <i>Human Molecular Genetics</i> , 2021, 30, 658-671.	1.4	12
65	Further evidence for heredity of pterygium. <i>Ophthalmic Genetics</i> , 2016, 37, 434-436.	0.5	11
66	Defining Ocular Surface Disease Activity and Damage Indices by an International Delphi Consultation. <i>Ocular Surface</i> , 2017, 15, 97-111.	2.2	11
67	Corneal Topographic Features in a Family With Nanophthalmos. <i>Cornea</i> , 2006, 25, 750-756.	0.9	10
68	Outcome of Descemet stripping automated endothelial keratoplasty in eyes with an Ahmed glaucoma valve. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2017, 255, 987-993.	1.0	10
69	Replacement of the Trabecular Meshwork Cells—A Way Ahead in IOP Control?. <i>Biomolecules</i> , 2021, 11, 1371.	1.8	10
70	Clinical and Genetic Aspects of Phelan-McDermid Syndrome: An Interdisciplinary Approach to Management. <i>Genes</i> , 2022, 13, 504.	1.0	9
71	Development of an Allele-Specific Real-Time PCR Assay for Discrimination and Quantification of p63 R279H Mutation in EEC Syndrome. <i>Journal of Molecular Diagnostics</i> , 2012, 14, 38-45.	1.2	8
72	Novel clinical features associated with Clouston syndrome. <i>International Journal of Dermatology</i> , 2019, 58, e143-e146.	0.5	8

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73	EffUnet-SpaGen: An Efficient and Spatial Generative Approach to Glaucoma Detection. <i>Journal of Imaging</i> , 2021, 7, 92.	1.7	8
74	Genome-wide transcriptome profiling of human trabecular meshwork cells treated with TGF- β 2. <i>Scientific Reports</i> , 2022, 12, .	1.6	8
75	Clinical, etiopathogenic, and therapeutic aspects of<sc>KID</sc> syndrome. <i>Dermatologic Therapy</i> , 2020, 33, e13507.	0.8	7
76	Delivering Endothelial Keratoplasty Grafts: Modern Day Transplant Devices. <i>Current Eye Research</i> , 2022, 47, 493-504.	0.7	7
77	Multisystemic Manifestations in Rare Diseases: The Experience of Dyskeratosis Congenita. <i>Genes</i> , 2022, 13, 496.	1.0	7
78	Identification of a novel frameshift mutation in the <i><sc>EDAR</sc></i> gene causing autosomal dominant hypohidrotic ectodermal dysplasia. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2015, 29, 1032-1034.	1.3	6
79	Bilateral keratoconus in tuberous sclerosis: is there a molecular link?. <i>Canadian Journal of Ophthalmology</i> , 2012, 47, e41-e42.	0.4	5
80	A novel <sc>INDEL</sc> mutation in the <sc>EDA</sc> gene resulting in a distinct Xâ€•linked hypohidrotic ectodermal dysplasia phenotype in an Italian family. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2016, 30, 341-343.	1.3	5
81	Exploiting biomaterial approaches to manufacture an artificial trabecular meshwork: A progress report. <i>Biomaterials and Biosystems</i> , 2021, 1, 100011.	1.0	5
82	Corneal Endothelial Cell Loss in Glaucoma and Glaucoma Surgery and the Utility of Management with Descemet Membrane Endothelial Keratoplasty (DMEK). <i>Journal of Ophthalmology</i> , 2022, 2022, 1-17.	0.6	5
83	A Comparative Genome-Wide Transcriptome Analysis of Glucocorticoid Responder and Non-Responder Primary Human Trabecular Meshwork Cells. <i>Genes</i> , 2022, 13, 882.	1.0	5
84	Amniotic band syndrome associated with an atypical iris and optic nerve defect. <i>Journal of AAPOS</i> , 2013, 17, 539-541.	0.2	4
85	The ocular phenotype of stiff-skin syndrome. <i>Eye</i> , 2016, 30, 156-159.	1.1	4
86	A Venezuelan Case of Schmid-Type Metaphyseal Chondrodysplasia with a Novel Mutation in<i>COL10A1</i>. <i>Molecular Syndromology</i> , 2019, 10, 167-170.	0.3	4
87	Detecting Change in Conjunctival Hyperemia Using a Pixel Densitometry Index. <i>Ocular Immunology and Inflammation</i> , 2019, 27, 276-281.	1.0	4
88	Assessment of differential intraocular pressure response to dexamethasone treatment in perfusion cultured Indian cadaveric eyes. <i>Scientific Reports</i> , 2021, 11, 605.	1.6	4
89	Response to Iliff etÂal.. <i>American Journal of Human Genetics</i> , 2012, 90, 934-935.	2.6	3
90	Short and long-term effect of dexamethasone on the transcriptome profile of primary human trabecular meshwork cells in vitro. <i>Scientific Reports</i> , 2022, 12, 8299.	1.6	3

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91	Avoiding Complications Associated With Preloaded Ultrathin Descemet Stripping Automated Endothelial Keratoplasty. <i>Cornea</i> , 2017, 36, e12-e13.	0.9	2
92	Chemical Cross-Linking of Corneal Tissue to Reduce Progression of Loss of Sight in Patients With Keratoconus. <i>Translational Vision Science and Technology</i> , 2021, 10, 6.	1.1	2
93	Oral manifestations in a boy with X-linked reticulate pigmentary disorder. <i>Head & Face Medicine</i> , 2012, 8, .	0.8	1
94	Membranous aplasia cutis congenita in trisomy 18. <i>Italian Journal of Pediatrics</i> , 2020, 46, 120.	1.0	1
95	COVID â€19 and ectodermal dysplasias. Recommendations are necessary. <i>Dermatologic Therapy</i> , 2020, 33, e13702.	0.8	1
96	Lensectomy in the Management of Glaucoma in Spherophakia: Is It Enough?. <i>Journal of Cataract and Refractive Surgery</i> , 2003, 29, 1053.	0.7	0
97	Heredity of Keratoconus. , 2013, , 37-52.		0
98	Pierquin Syndrome: Report of a New Case. <i>Journal of Pediatric Neurology</i> , 2021, 19, 046-049.	0.0	0
99	A Severe Case of Spondylometaphyseal Dysplasia Algerian Type with Two Mutations in COL2A1. <i>Journal of Pediatric Genetics</i> , 2023, 12, 339-341.	0.3	0