Colin Eric Willoughby

List of Publications by Year
in descending order

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	A Severe Case of Spondylometaphyseal Dysplasia Algerian Type with Two Mutations in COL2A1. Journal of Pediatric Genetics, 2023, 12, 339-341.	0.3	0
2	Delivering Endothelial Keratoplasty Grafts: Modern Day Transplant Devices. Current Eye Research, 2022, 47, 493-504.	0.7	7
3	Prognostic value of miR-21 for prostate cancer: a systematic review and meta-analysis. Bioscience Reports, 2022, 42, .	1.1	15
4	Corneal Endothelial Cell Loss in Glaucoma and Glaucoma Surgery and the Utility of Management with Descemet Membrane Endothelial Keratoplasty (DMEK). Journal of Ophthalmology, 2022, 2022, 1-17.	0.6	5
5	Consensus Recommendation for Mouse Models of Ocular Hypertension to Study Aqueous Humor Outflow and Its Mechanisms. , 2022, 63, 12.		20
6	Clinical and Genetic Aspects of Phelanâ€“McDermid Syndrome: An Interdisciplinary Approach to Management. Genes, 2022, 13, 504.	1.0	9
7	Multisystemic Manifestations in Rare Diseases: The Experience of Dyskeratosis Congenita. Genes, 2022, 13, 496.	1.0	7
8	A Comparative Genome-Wide Transcriptome Analysis of Glucocorticoid Responder and Non-Responder Primary Human Trabecular Meshwork Cells. Genes, 2022, 13, 882.	1.0	5
9	Short and long-term effect of dexamethasone on the transcriptome profile of primary human trabecular meshwork cells in vitro. Scientific Reports, 2022, 12, 8299.	1.6	3
10	Genome-wide transcriptome profiling of human trabecular meshwork cells treated with TGF-Î²2. Scientific Reports, 2022, 12, .	1.6	8
11	Pierquin Syndrome: Report of a New Case. Journal of Pediatric Neurology, 2021, 19, 046-049.	0.0	0
12	Assessment of differential intraocular pressure response to dexamethasone treatment in perfusion cultured Indian cadaveric eyes. Scientific Reports, 2021, 11, 605.	1.6	4
13	Biomaterials for corneal endothelial cell culture and tissue engineering. Journal of Tissue Engineering, 2021, 12, 204173142199053.	2.3	32
14	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. Human Molecular Genetics, 2021, 30, 658-671.	1.4	12
15	Exploiting biomaterial approaches to manufacture an artificial trabecular meshwork: A progress report. Biomaterials and Biosystems, 2021, 1, 100011.	1.0	5
16	Chemical Cross-Linking of Corneal Tissue to Reduce Progression of Loss of Sight in Patients With Keratoconus. Translational Vision Science and Technology, 2021, 10, 6.	1.1	2
17	EffUnet-SpaGen: An Efficient and Spatial Generative Approach to Glaucoma Detection. Journal of Imaging, 2021, 7, 92.	1.7	8
18	Targeting the NLRP3 Inflammasome in Glaucoma. Biomolecules, 2021, 11, 1239.	1.8	22

#	Article	IF	Citations
19	Replacement of the Trabecular Meshwork Cellsâ€"A Way Ahead in IOP Control?. Biomolecules, 2021, 11, 1371.	1.8	10
20	Corneal biomechanics and biomechanically corrected intraocular pressure in primary open-angle glaucoma, ocular hypertension and controls. British Journal of Ophthalmology, 2020, 104, 121-126.	2.1	67
21	An Intraocular Pressure Polygenic Risk Score Stratifies Multiple Primary Open-Angle Glaucoma Parameters Including Treatment Intensity. Ophthalmology, 2020, 127, 901-907.	2.5	37
22	Association of Genetic Variation With Keratoconus. JAMA Ophthalmology, 2020, 138, 174.	1.4	34
23	Membranous aplasia cutis congenita in trisomy 18. Italian Journal of Pediatrics, 2020, 46, 120.	1.0	1
24	Clinical, etiopathogenic, and therapeutic aspects of<scp>KID</scp>syndrome. Dermatologic Therapy, 2020, 33, e13507.	0.8	7
25	COVID â€19 and ectodermal dysplasias. Recommendations are necessary. Dermatologic Therapy, 2020, 33, e13702.	0.8	1
26	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. Nature Genetics, 2020, 52, 160-166.	9.4	192
27	Cross-Country Transportation Efficacy and Clinical Outcomes of Preloaded Large-Diameter Ultra-Thin Descemet Stripping Automated Endothelial Keratoplasty Grafts. Cornea, 2019, 38, 30-34.	0.9	20
28	Accurate, fast, data efficient and interpretable glaucoma diagnosis with automated spatial analysis of the whole cup to disc profile. PLoS ONE, 2019, 14, e0209409.	1.1	27
29	Novel clinical features associated with Clouston syndrome. International Journal of Dermatology, 2019, 58, e143-e146.	0.5	8
30	A Venezuelan Case of Schmid-Type Metaphyseal Chondrodysplasia with a Novel Mutation inCOL10A1. Molecular Syndromology, 2019, 10, 167-170.	0.3	4
31	Bandage contact lens and topical steroids are risk factors for the development of microbial keratitis after epithelium-off CXL. BMJ Open Ophthalmology, 2019, 4, e000231.	0.8	25
32	Detecting Change in Conjunctival Hyperemia Using a Pixel Densitometry Index. Ocular Immunology and Inflammation, 2019, 27, 276-281.	1.0	4
33	Gene-based antiangiogenic applications for corneal neovascularization. Survey of Ophthalmology, 2018, 63, 193-213.	1.7	33
34	Comparison of preservation and transportation protocols for preloaded Descemet membrane endothelial keratoplasty. British Journal of Ophthalmology, 2018, 102, 549-555.	2.1	58
35	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	5.8	63
36	Improving precision for detecting change in the shape of the cornea in patients with keratoconus. Scientific Reports, 2018, 8, 12345.	1.6	45

#	Article	IF	Citations
37	Imaging of Corneal Neovascularization: Optical Coherence Tomography Angiography and Fluorescence Angiography. , 2018, 59, 1263.		47
38	The Relationship Between Mechanical Properties, Ultrastructural Changes, and Intrafibrillar Bond Formation in Corneal UVA/Riboflavin Cross-linking Treatment for Keratoconus. Journal of Refractive Surgery, 2018, 34, 264-272.	1.1	38
39	Mitochondrial dysfunction and oxidative stress in corneal disease. Mitochondrion, 2017, 36, 103-113.	1.6	73
40	Outcome of Descemet stripping automated endothelial keratoplasty in eyes with an Ahmed glaucoma valve. Graefe's Archive for Clinical and Experimental Ophthalmology, 2017, 255, 987-993.	1.0	10
41	Preparation of ultrathin grafts for Descemet-stripping endothelial keratoplasty with a single microkeratome pass. Journal of Cataract and Refractive Surgery, 2017, 43, 12-15.	0.7	42
42	Avoiding Complications Associated With Preloaded Ultrathin Descemet Stripping Automated Endothelial Keratoplasty. Cornea, 2017, 36, e12-e13.	0.9	2
43	In Vivo Early Corneal Biomechanical Changes After Corneal Cross-linking in Patients With Progressive Keratoconus. Journal of Refractive Surgery, 2017, 33, 840-846.	1.1	79
44	Defining Ocular Surface Disease Activity and Damage Indices by an International Delphi Consultation. Ocular Surface, 2017, 15, 97-111.	2.2	11
45	Deformation velocity imaging using optical coherence tomography and its applications to the cornea. Biomedical Optics Express, 2017, 8, 5579.	1.5	22
46	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	1.1	54
47	Method for Angiographically Guided Fine-Needle Diathermy in the Treatment of Corneal Neovascularization. Cornea, 2016, 35, 1029-1032.	0.9	18
48	Femtosecond Laserâ€“Assisted Lamellar Keratectomy for Corneal Opacities Secondary to Anterior Corneal Dystrophies. Cornea, 2016, 35, 6-13.	0.9	15
49	Correction of Mutant p63 in EEC Syndrome Using siRNA Mediated Allele-Specific Silencing Restores Defective Stem Cell Function. Stem Cells, 2016, 34, 1588-1600.	1.4	17
50	High resolution corneal and single pulse imaging with line field spectral domain optical coherence tomography. Optics Express, 2016, 24, 12395.	1.7	31
51	The ocular phenotype of stiff-skin syndrome. Eye, 2016, 30, 156-159.	1.1	4
52	A COL17A1 Splice-Altering Mutation Is Prevalent in Inherited Recurrent Corneal Erosions. Ophthalmology, 2016, 123, 709-722.	2.5	37
53	A novel <scp>INDEL</scp> mutation in the <scp>EDA</scp> gene resulting in a distinct Xâ€•linked hypohidrotic ectodermal dysplasia phenotype in an Italian family. Journal of the European Academy of Dermatology and Venereology, 2016, 30, 341-343.	1.3	5
54	Further evidence for heredity of pterygium. Ophthalmic Genetics, 2016, 37, 434-436.	0.5	11

#	Article	IF	Citations
55	Angiographic and In Vivo Confocal Microscopic Characterization of Human Corneal Blood and Presumed Lymphatic Neovascularization. Cornea, 2015, 34, 1459-1465.	0.9	25
56	Reliability of the Effect of Artificial Anterior Chamber Pressure and Corneal Drying on Corneal Graft Thickness. Cornea, 2015, 34, 866-869.	0.9	18
57	Next-generation sequencing-based molecular diagnosis of 82 retinitis pigmentosa probands from Northern Ireland. Human Genetics, 2015, 134, 217-230.	1.8	85
58	Identification of a novel frameshift mutation in the <scp>EDAR</scp> gene causing autosomal dominant hypohidrotic ectodermal dysplasia. Journal of the European Academy of Dermatology and Venereology, 2015, 29, 1032-1034.	1.3	6
59	Corneal Angiography for Guiding and Evaluating Fine-Needle Diathermy Treatment of Corneal Neovascularization. Ophthalmology, 2015, 122, 1079-1084.	2.5	53
60	Influence of graft size on graft survival following Descemet stripping automated endothelial keratoplasty. British Journal of Ophthalmology, 2015, 99, 784-788.	2.1	35
61	Iris Flocculi as an Ocular Marker ofACTA2Mutation in Familial Thoracic Aortic Aneurysms and Dissections. Ophthalmic Genetics, 2015, 36, 86-88.	0.5	12
62	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
63	Whole-mitochondrial genome sequencing in primary open-angle glaucoma using massively parallel sequencing identifies novel and known pathogenic variants. Genetics in Medicine, 2015, 17, 279-284.	1.1	38
64	Phenotypic heterogeneity and mutational spectrum in a cohort of 45 Italian males subjects with Xâ€linked ectodermal dysplasia. Clinical Genetics, 2015, 87, 338-342.	1.0	16
65	Mutational Analysis of theRhodopsinGene in Sector Retinitis Pigmentosa. Ophthalmic Genetics, 2015, 36, 239-243.	0.5	14
66	Enrichment of pathogenic alleles in the brittle cornea gene, ZNF469, in keratoconus. Human Molecular Genetics, 2014, 23, 5527-5535.	1.4	56
67	Corneal endothelial dysfunction in Pearson syndrome. Ophthalmic Genetics, 2013, 34, 55-57.	0.5	13
68	Infantile bilateral glaucoma in a child with ectodermal dysplasia. Ophthalmic Genetics, 2013, 34, 58-60.	0.5	15
69	Amniotic band syndrome associated with an atypical iris and optic nerve defect. Journal of AAPOS, 2013, 17, 539-541.	0.2	4
70	Heredity of Keratoconus. , 2013, , 37-52.		0
71	Mutational Analysis ofMIR184in Sporadic Keratoconus and Myopia. , 2013, 54, 5266.		73
72	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	9.4	269

#	Article	IF	Citations
73	Mutational Spectrum of theZEB1Gene in Corneal Dystrophies Supports a Genotypeâ€“Phenotype Correlation. , 2013, 54, 3215.		65
74	Marginal Corneal Vascular Arcades. , 2013, 54, 7470.		22
75	Bilateral keratoconus in tuberous sclerosis: is there a molecular link?. Canadian Journal of Ophthalmology, 2012, 47, e41-e42.	0.4	5
76	Development of an Allele-Specific Real-Time PCR Assay for Discrimination and Quantification of p63 R279H Mutation in EEC Syndrome. Journal of Molecular Diagnostics, 2012, 14, 38-45.	1.2	8
77	Limbal Stem Cell Deficiency and Ocular Phenotype in Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome Caused by p63 Mutations. Ophthalmology, 2012, 119, 74-83.	2.5	94
78	Mitochondrial dysfunction in glaucoma: Understanding genetic influences. Mitochondrion, 2012, 12, 202-212.	1.6	85
79	A novel de novo missense mutation in TP63 underlying germline mosaicism in AEC syndrome: Implications for recurrence risk and prenatal diagnosis. American Journal of Medical Genetics, Part A, 2012, 158A, 1957-1961.	0.7	19
80	Response to Iliff etÂal.. American Journal of Human Genetics, 2012, 90, 934-935.	2.6	3
81	Keratoconus in 18 pairs of twins. Acta Ophthalmologica, 2012, 90, e482-6.	0.6	102
82	Oral manifestations in a boy with X-linked reticulate pigmentary disorder. Head & Face Medicine, 2012, 8, .	0.8	1
83	Association of Polymorphisms in the Hepatocyte Growth Factor Gene Promoter with Keratoconus. , 2011, 52, 8514.		114
84	Mutation Altering the miR-184 Seed Region Causes Familial Keratoconus with Cataract. American Journal of Human Genetics, 2011, 89, 628-633.	2.6	234
85	Molecular diagnosis for heterogeneous genetic diseases with targeted high-throughput DNA sequencing applied to retinitis pigmentosa. Journal of Medical Genetics, 2011, 48, 145-151.	1.5	81
86	Anatomy and physiology of the human eye: effects of mucopolysaccharidoses disease on structure and function â€“ a review. Clinical and Experimental Ophthalmology, 2010, 38, 2-11.	1.3	127
87	Monozygotic twins discordant for phacomatosis pigmentovascularis: Evidence for the concept of twin spotting. American Journal of Medical Genetics, Part A, 2010, 152A, 718-720.	0.7	14
88	Development of a Diagnostic Genetic Test for Simplex and Autosomal Recessive Retinitis Pigmentosa. Ophthalmology, 2010, 117, 2169-2177.e3.	2.5	42
89	Replication of the Recessive STBMS1 Locus but with Dominant Inheritance. , 2009, 50, 3210.		25
90	Mutations in smooth muscle Î±-actin (ACTA2) lead to thoracic aortic aneurysms and dissections. Nature Genetics, 2007, 39, 1488-1493.	9.4	767

#	Article	IF	Citations
91	Corneal Topographic Features in a Family With Nanophthalmos. Cornea, 2006, 25, 750-756.	0.9	10
92	CRYBB1 mutation associated with congenital cataract and microcornea. Molecular Vision, 2005, 11, 587-93.	1.1	41
93	Defining the Pathogenicity ofOptineurinin Juvenile Open-Angle Glaucoma. , 2004, 45, 3122.		52
94	The Importance of Screening for Sight-Threatening Retinopathy in Incontinentia Pigmenti. Pediatric Dermatology, 2004, 21, 242-245.	0.5	13
95	Lensectomy in the Management of Glaucoma in Spherophakia: Is It Enough?. Journal of Cataract and Refractive Surgery, 2003, 29, 1053.	0.7	0
96	Simplifying Collection of Corneal Specimens in Cases of Suspected Bacterial Keratitis. Journal of Clinical Microbiology, 2003, 41, 3192-3197.	1.8	59
97	Missense Mutations in GJB2 Encoding Connexin-26 Cause the Ectodermal Dysplasia Keratitis-Ichthyosis-Deafness Syndrome. American Journal of Human Genetics, 2002, 70, 1341-1348.	2.6	345
98	Lensectomy in the management of glaucoma in spherophakia. Journal of Cataract and Refractive Surgery, 2002, 28, 1061-1064.	0.7	45
99	Common Mutations in Arg304 of the p63 Gene in Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome: Lack of Genotypeâ€“Phenotype Correlation and Implications for Mutation Detection Strategies. Journal of Investigative Dermatology, 2002, 119, 1202-1203.	0.3	12