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List of Publications by Year in descending order

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157 157 20161 all docs docs citations times ranked citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Identification of a Gene That Causes Primary Open Angle Glaucoma. Science, 1997, 275, 668-670.	12.6	1,274
3	Primary Open-Angle Glaucoma. New England Journal of Medicine, 2009, 360, 1113-1124.	27.0	747
4	Analysis of Myocilin Mutations in 1703 Glaucoma Patients From Five Different Populations. Human Molecular Genetics, 1999, 8, 899-905.	2.9	496
5	Clinical Features Associated with Mutations in the Chromosome 1 Open-Angle Glaucoma Gene (<i>GLC1A</i>). New England Journal of Medicine, 1998, 338, 1022-1027.	27.0	423
6	Automated Segmentation of the Optic Disc from Stereo Color Photographs Using Physiologically Plausible Features., 2007, 48, 1665.		275
7	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. Nature Genetics, 2014, 46, 1126-1130.	21.4	212
8	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. Nature Genetics, 2016, 48, 189-194.	21.4	211
9	Myocilin Glaucoma. Survey of Ophthalmology, 2002, 47, 547-561.	4.0	201
10	Retinal Ganglion Cell Death in Glaucoma: Mechanisms and Neuroprotective Strategies. Ophthalmology Clinics of North America, 2005, 18, 383-395.	1.8	192
11	Copy number variations on chromosome $12q14$ in patients with normal tension glaucoma. Human Molecular Genetics, $2011, 20, 2482-2494$.	2.9	189
12	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. Nature Genetics, 2014, 46, 1120-1125.	21.4	186
13	Primary open-angle glaucoma genes. Eye, 2011, 25, 587-595.	2.1	184
14	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. Nature Genetics, 2016, 48, 556-562.	21.4	147
15	Increased expression of the WNT antagonist sFRP-1 in glaucoma elevates intraocular pressure. Journal of Clinical Investigation, 2008, 118, 1056-64.	8.2	143
16	CRISPR-Cas9â€"based treatment of myocilin-associated glaucoma. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 11199-11204.	7.1	137
17	New insights into the genetics of primary open-angle glaucoma based on meta-analyses of intraocular pressure and optic disc characteristics Human Molecular Genetics, 2017, 26, ddw399.	2.9	120
18	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	21.4	114

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19	LOXL1 Mutations Are Associated with Exfoliation Syndrome in Patients from the Midwestern United States. American Journal of Ophthalmology, 2007, 144, 974-975.e1.	3.3	111
20	Evidence for genetic heterogeneity within eight glaucoma families, with the GLC1A Gln368STOP mutation being an important phenotypic modifier11None of the authors has a financial interest relating to this article Ophthalmology, 2001, 108, 1607-1620.	5.2	106
21	Evidence for a Novel X-Linked Modifier Locus for Leber Hereditary Optic Neuropathy. Ophthalmic Genetics, 2008, 29, 17-24.	1.2	105
22	A common variant near TGFBR3 is associated with primary open angle glaucoma. Human Molecular Genetics, 2015, 24, 3880-3892.	2.9	105
23	North Carolina Macular Dystrophy Is Caused by Dysregulation of the Retinal Transcription Factor PRDM13. Ophthalmology, 2016, 123, 9-18.	5.2	105
24	Characterization and Comparison of the Human and Mouse <i>GLC1A</i> Glaucoma Genes. Genome Research, 1998, 8, 377-384.	5.5	103
25	Complement Component C5a Activates ICAM-1 Expression on Human Choroidal Endothelial Cells. , 2010, 51, 5336.		101
26	Genome-wide association study identifies three novel loci in Fuchs endothelial corneal dystrophy. Nature Communications, 2017, 8, 14898.	12.8	101
27	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	21.4	97
28	Variations in the Myocilin Gene in Patients With Open-Angle Glaucoma. JAMA Ophthalmology, 2002, 120, 1189.	2.4	96
29	Association of CAV1/CAV2 Genomic Variants with Primary Open-Angle Glaucoma Overall and by Gender and Pattern of Visual Field Loss. Ophthalmology, 2014, 121, 508-516.	5.2	91
30	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. Nature Communications, 2014, 5, 4883.	12.8	89
31	Primary congenital and developmental glaucomas. Human Molecular Genetics, 2017, 26, R28-R36.	2.9	85
32	Discovery and Functional Annotation of SIX6 Variants in Primary Open-Angle Glaucoma. PLoS Genetics, 2014, 10, e1004372.	3.5	78
33	Leber's hereditary optic neuropathy triggered by antiretroviral therapy for human immunodeficiency virus. Eye, 2003, 17, 312-317.	2.1	77
34	<i>TBK1</i> Gene Duplication and Normal-Tension Glaucoma. JAMA Ophthalmology, 2014, 132, 544.	2.5	77
35	Glaucoma-associated corneal endothelial cell damage: A review. Survey of Ophthalmology, 2018, 63, 500-506.	4.0	77
36	Calpain-5 Mutations Cause Autoimmune Uveitis, Retinal Neovascularization, and Photoreceptor Degeneration. PLoS Genetics, 2012, 8, e1003001.	3.5	76

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37	Duplication of TBK1 Stimulates Autophagy in iPSC-derived Retinal Cells from a Patient with Normal Tension Glaucoma. Journal of Stem Cell Research & Therapy, 2014, 04, 161.	0.3	75
38	Investigation of Known Genetic Risk Factors for Primary Open Angle Glaucoma in Two Populations of African Ancestry., 2013, 54, 6248.		73
39	Metaâ€nnalysis of Genomeâ€Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. Genetic Epidemiology, 2015, 39, 207-216.	1.3	72
40	Confirmation of TBK1 duplication in normal tension glaucoma. Experimental Eye Research, 2012, 96, 178-180.	2.6	71
41	Copy Number Variations of TBK1 in Australian Patients With Primary Open-Angle Glaucoma. American Journal of Ophthalmology, 2015, 159, 124-130.e1.	3.3	68
42	Ethnic variation in AMD-associated complement factor H polymorphism p.Tyr402His. Human Mutation, 2006, 27, 921-925.	2.5	66
43	CRISPR-Cas9 genome engineering: Treating inherited retinal degeneration. Progress in Retinal and Eye Research, 2018, 65, 28-49.	15.5	64
44	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	12.8	63
45	No Association Between Variations in the WDR36 Gene and Primary Open-Angle Glaucoma. JAMA Ophthalmology, 2007, 125, 434.	2.4	58
46	Increased Expression of Serum Amyloid A in Glaucoma and Its Effect on Intraocular Pressure. , 2008, 49, 1916.		50
47	Systems genetics identifies a role for Cacna2d1 regulation in elevated intraocular pressure and glaucoma susceptibility. Nature Communications, 2017, 8, 1755.	12.8	50
48	Association of Genetic Variants With Primary Open-Angle Glaucoma Among Individuals With African Ancestry. JAMA - Journal of the American Medical Association, 2019, 322, 1682.	7.4	50
49	Glaucoma Phenotype in Pedigrees With the Myocilin Thr377Met Mutation. JAMA Ophthalmology, 2003, 121, 1172.	2.4	47
50	Association of a Novel Mutation in the Retinol Dehydrogenase 12 (RDH12) Gene With Autosomal Dominant Retinitis Pigmentosa. JAMA Ophthalmology, 2008, 126, 1301.	2.4	47
51	Expression of the glaucoma gene myocilin (MYOC) in the human optic nerve head. FASEB Journal, 2001, 15, 1251-1253.	0.5	46
52	<i>Lyst</i> Mutation in Mice Recapitulates Iris Defects of Human Exfoliation Syndrome., 2009, 50, 1205.		46
53	Assessing the Association of Mitochondrial Genetic Variation With Primary Open-Angle Glaucoma Using Gene-Set Analyses., 2016, 57, 5046.		44
54	A Common Variant in <i>MIR182</i> Is Associated With Primary Open-Angle Glaucoma in the NEIGHBORHOOD Consortium., 2016, 57, 4528.		42

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55	Chromosome 7q31 POAG locus: ocular expression of caveolins and lack of association with POAG in a US cohort. Molecular Vision, 2011, 17, 430-5.	1.1	41
56	Estrogen pathway polymorphisms in relation to primary open angle glaucoma: an analysis accounting for gender from the United States. Molecular Vision, 2013, 19, 1471-81.	1.1	40
57	Mendelian genes in primary open angle glaucoma. Experimental Eye Research, 2019, 186, 107702.	2.6	39
58	Cell–Matrix Interactions in the Eye: From Cornea to Choroid. Cells, 2021, 10, 687.	4.1	39
59	Genome-wide analysis of copy number variants in age-related macular degeneration. Human Genetics, 2011, 129, 91-100.	3.8	36
60	A Genome-Wide Association Study for Primary Open Angle Glaucoma and Macular Degeneration Reveals Novel Loci. PLoS ONE, 2013, 8, e58657.	2.5	35
61	Mutation in the <i>SLC4A11 </i> Gene Associated with Autosomal Recessive Congenital Hereditary Endothelial Dystrophy in a Large Saudi Family. Ophthalmic Genetics, 2008, 29, 41-45.	1.2	34
62	Optical Coherence Tomography Analysis Based Prediction of Humphrey 24-2 Visual Field Thresholds in Patients With Glaucoma., 2017, 58, 3975.		34
63	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. Scientific Reports, 2018, 8, 3124.	3.3	33
64	Clinical and genetic characterization of a Danish family with North Carolina macular dystrophy. Molecular Vision, 2010, 16, 2659-68.	1.1	33
65	Hypothesis-independent pathway analysis implicates GABA and Acetyl-CoA metabolism in primary open-angle glaucoma and normal-pressure glaucoma. Human Genetics, 2014, 133, 1319-1330.	3.8	32
66	Association of a Primary Open-Angle Glaucoma Genetic Risk Score With Earlier Age at Diagnosis. JAMA Ophthalmology, 2019, 137, 1190.	2.5	32
67	Copy Number Variations and Primary Open-Angle Glaucoma. , 2011, 52, 7122.		31
68	Genomic locus modulating corneal thickness in the mouse identifies POU6F2 as a potential risk of developing glaucoma. PLoS Genetics, 2018, 14, e1007145.	3.5	31
69	Broad phenotypic variability in a single pedigree with a novel 1410delC mutation in the PST domain of the PAX6 gene. Human Mutation, 2002, 20, 322-322.	2.5	30
70	Automated Axon Counting in Rodent Optic Nerve Sections with AxonJ. Scientific Reports, 2016, 6, 26559.	3.3	30
71	Case of Stargardt Disease Caused by Uniparental Isodisomy. JAMA Ophthalmology, 2006, 124, 744.	2.4	27
72	DNA Copy Number Variants of Known Glaucoma Genes in Relation to Primary Open-Angle Glaucoma. Investigative Ophthalmology and Visual Science, 2014, 55, 8251-8258.	3.3	27

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73	Genetics and genetic testing for glaucoma. Current Opinion in Ophthalmology, 2017, 28, 133-138.	2.9	26
74	Glaucoma Risk Alleles in the Ocular Hypertension Treatment Study. Ophthalmology, 2016, 123, 2527-2536.	5.2	25
75	CRISPRâ€Cas9â€Based Genome Editing of Human Induced Pluripotent Stem Cells. Current Protocols in Stem Cell Biology, 2018, 44, 5B.7.1-5B.7.22.	3.0	25
76	Novel TMEM98 mutations in pedigrees with autosomal dominant nanophthalmos. Molecular Vision, 2015, 21, 1017-23.	1.1	24
77	Familial Cavitary Optic Disk Anomalies: Identification of a Novel Genetic Locus. American Journal of Ophthalmology, 2007, 143, 795-800.e1.	3.3	23
78	The C677T Variant in the Methylenetetrahydrofolate Reductase Gene Is Not Associated with Disease in Cohorts of Pseudoexfoliation Glaucoma and Primary Open-Angle Glaucoma Patients from Iowa. Ophthalmic Genetics, 2006, 27, 39-41.	1.2	22
79	Heritable Features of the Optic Disc: A Novel Twin Method for Determining Genetic Significance. , 2007, 48, 2469.		22
80	Confirmation of the association between the <i>TCF4 < /i> risk allele and Fuchs endothelial corneal dystrophy in patients from the Midwestern United States. Ophthalmic Genetics, 2013, 34, 32-34.</i>	1.2	22
81	Multi-trait genome-wide association study identifies new loci associated with optic disc parameters. Communications Biology, 2019, 2, 435.	4.4	22
82	Familial Cavitary Optic Disk Anomalies: Clinical Features of a Large Family with Examples of Progressive Optic Nerve Head Cupping. American Journal of Ophthalmology, 2007, 143, 788-794.e1.	3.3	21
83	Analysis of ASB10 variants in open angle glaucoma. Human Molecular Genetics, 2012, 21, 4543-4548.	2.9	20
84	Novel IntragenicFRMD7Deletion in a Pedigree with Congenital X-Linked Nystagmus. Ophthalmic Genetics, 2010, 31, 77-80.	1.2	19
85	The Utility of Diaton Tonometer Measurements in Patients With Ocular Hypertension, Glaucoma, and Glaucoma Tube Shunts: A Preliminary Study for its Potential Use in Keratoprosthesis Patients. Journal of Glaucoma, 2016, 25, 643-647.	1.6	19
86	Transgenic <i>TBK1</i> mice have features of normal tension glaucoma. Human Molecular Genetics, 2017, 26, ddw372.	2.9	19
87	Reduced frequency of known mutations in a cohort of LHON patients from India. Ophthalmic Genetics, 2010, 31, 196-199.	1.2	18
88	Genetic correlations between intraocular pressure, blood pressure and primary open-angle glaucoma: a multi-cohort analysis. European Journal of Human Genetics, 2017, 25, 1261-1267.	2.8	18
89	Assessment of SNPs associated with the human glucocorticoid receptor in primary open-angle glaucoma and steroid responders. Molecular Vision, 2010, 16, 596-601.	1.1	18
90	<i>TBK1</i> and Flanking Genes in Human Retina. Ophthalmic Genetics, 2014, 35, 35-40.	1.2	17

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91	Myocilin Mutations in Patients With Normal-Tension Glaucoma. JAMA Ophthalmology, 2019, 137, 559.	2.5	17
92	Clinical and molecular characterization of a family affected with X-linked ocular albinism(OA1). Ophthalmic Genetics, 1997, 18, 175-184.	1.2	16
93	The Optic Nerve Head in Myocilin Glaucoma., 2007, 48, 238.		16
94	Familial non-arteritic anterior ischemic optic neuropathy. Graefe's Archive for Clinical and Experimental Ophthalmology, 2008, 246, 1295-1305.	1.9	16
95	Clinical correlates to the goniodysgensis among juvenile-onset primary open-angle glaucoma patients. Graefe's Archive for Clinical and Experimental Ophthalmology, 2013, 251, 1571-1576.	1.9	16
96	The Heritability of Pigment Dispersion Syndrome and Pigmentary Glaucoma. American Journal of Ophthalmology, 2019, 202, 55-61.	3.3	16
97	The genetic aspects of adult-onset glaucoma: a perspective from the Greater Toronto area. Canadian Journal of Ophthalmology, 2000, 35, 12-17.	0.7	15
98	Evaluation of variants in the selectin genes in age-related macular degeneration. BMC Medical Genetics, 2011, 12, 58.	2.1	15
99	Mitochondrial Variant G4132A is Associated with Familial Non-Arteritic Anterior Ischemic Optic Neuropathy in One Large Pedigree. Ophthalmic Genetics, 2007, 28, 1-7.	1.2	14
100	Microarray Analysis of Iris Gene Expression in Mice with Mutations Influencing Pigmentation., 2011, 52, 237.		14
101	Vascular tone pathway polymorphisms in relation to primary open-angle glaucoma. Eye, 2014, 28, 662-671.	2.1	14
102	Testosterone Pathway Genetic Polymorphisms in Relation to Primary Open-Angle Glaucoma: An Analysis in Two Large Datasets., 2018, 59, 629.		14
103	Tank-Binding Kinase 1 () Gene and Open-Angle Glaucomas (An American Ophthalmological Society) Tj ETQq1 1 C).784314 r 1.4	gBT/Overlo
104	Myocilin Gly252Arg Mutation and Glaucoma of Intermediate Severity in Caucasian Individuals. JAMA Ophthalmology, 2007, 125, 98.	2.4	13
105	Primary Open-Angle Glaucoma. New England Journal of Medicine, 2009, 360, 2679-2680.	27.0	12
106	Genetic Correlations Between Diabetes and Glaucoma: An Analysis of Continuous and Dichotomous Phenotypes. American Journal of Ophthalmology, 2019, 206, 245-255.	3.3	12
107	Family-Based Genome-Wide Association Study of South Indian Pedigrees Supports <i>WNT7B</i> as a Central Corneal Thickness Locus., 2018, 59, 2495.		11
108	Histochemical Analysis of Glaucoma Caused by a Myocilin Mutation in a Human Donor Eye. Ophthalmology Glaucoma, 2018, $1,132\text{-}138$.	1.9	11

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109	Animal Models of Exfoliation Syndrome, Now and Future. Journal of Glaucoma, 2014, 23, S68-S72.	1.6	10
110	Heterozygous Triplication of Upstream Regulatory Sequences Leads to Dysregulation of Matrix Metalloproteinase 19 in Patients with Cavitary Optic Disc Anomaly. Human Mutation, 2015, 36, 369-378.	2.5	10
111	Update on Animal Models of Exfoliation Syndrome. Journal of Glaucoma, 2018, 27, S78-S82.	1.6	10
112	Novel Intragenic <i>PAX6</i> Deletion in a Pedigree with Aniridia, Morbid Obesity, and Diabetes. Current Eye Research, 2020, 45, 91-96.	1.5	10
113	Circumferential Iris Transillumination Defects in Exfoliation Syndrome. Journal of Glaucoma, 2013, 22, 555-558.	1.6	9
114	SQSTM1 Mutations and Glaucoma. PLoS ONE, 2016, 11, e0156001.	2.5	9
115	Exome-based investigation of the genetic basis of human pigmentary glaucoma. BMC Genomics, 2021, 22, 477.	2.8	9
116	Diffusion Tensor Imaging of Visual Pathway Abnormalities in Five Glaucoma Animal Models., 2021, 62, 21.		9
117	Automated Quantification of Inherited Phenotypes from Color Images: A Twin Study of the Variability of Optic Nerve Head Shape., 2010, 51, 5870.		8
118	Localization of SH3PXD2B in human eyes and detection of rare variants in patients with anterior segment diseases and glaucoma. Molecular Vision, 2012, 18, 705-13.	1.1	8
119	Statistical Tests for Detecting Rare Variants Using Varianceâ€Stabilising Transformations. Annals of Human Genetics, 2012, 76, 402-409.	0.8	7
120	Identification of Proteins that Interact with TANK Binding Kinase 1 and Testing for Mutations Associated with Glaucoma. Current Eye Research, 2013, 38, 310-315.	1.5	7
121	Genomic Organization of TBK1 Copy Number Variations in Glaucoma Patients. Journal of Glaucoma, 2017, 26, 1063-1067.	1.6	6
122	Age at natural menopause genetic risk score in relation to age at natural menopause and primary open-angle glaucoma in a US-based sample. Menopause, 2017, 24, 150-156.	2.0	6
123	Penetrance of Myocilin Mutations—Who Gets Glaucoma?. JAMA Ophthalmology, 2019, 137, 35.	2.5	6
124	Genetic Association between MMP9 and Choroidal Neovascularization in Age-Related Macular Degeneration. Ophthalmology Science, 2021, 1, 100002.	2.5	6
125	Automated segmentation of choroidal layers from 3-dimensional macular optical coherence tomography scans. Journal of Neuroscience Methods, 2021, 360, 109267.	2.5	5
126	Nanophthalmos patient with a THR518MET mutation in MYRF, a case report. BMC Ophthalmology, 2020, 20, 388.	1.4	5

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127	LADD syndrome with glaucoma is caused by a novel gene. Molecular Vision, 2017, 23, 179-184.	1.1	5
128	Normal range of hearing associated with myocilin Thr377Met. Ophthalmic Genetics, 1999, 20, 205-207.	1.2	4
129	Clinical and genetic characterization of a large primary open angle glaucoma pedigree. Ophthalmic Genetics, 2017, 38, 222-225.	1.2	4
130	Gonioscopy-Assisted Transluminal Trabeculotomy for Myocilin Juvenile Glaucoma. Ophthalmology Glaucoma, 2022, 5, 369-370.	1.9	4
131	Thin Central Corneal Thickness and Early-Onset Glaucoma in Lacrimo-auriculo-dento-digital Syndrome. JAMA Ophthalmology, 2014, 132, 782.	2.5	3
132	Stereo Photo Measured ONH Shape Predicts Development of POAG in Subjects With Ocular Hypertension., 2015, 56, 4470.		3
133	Enhanced depth imaging optical coherence tomography of congenital cavitary optic disc anomaly (CODA). British Journal of Ophthalmology, 2015, 99, 549-555.	3.9	3
134	Long-Term Follow-Up of Normal Tension Glaucoma Patients With TBK1 Gene Mutations in One Large Pedigree. American Journal of Ophthalmology, 2020, 214, 52-62.	3.3	3
135	Prevalence of Open-angle Glaucoma in the Faroese Population. Journal of Glaucoma, 2022, 31, 72-78.	1.6	3
136	MMP19 expression in the human optic nerve. Molecular Vision, 2016, 22, 1429-1436.	1.1	3
137	Automated discovery of structural features of the optic nerve head on the basis of image and genetic data., 2014,,.		2
138	Assessment of a three-generation pedigree with Fuchs endothelial corneal dystrophy with anticipation for expansion of the triplet repeat in the TCF4 gene. Eye, 2017, 31, 1250-1252.	2.1	2
139	A novel mutation (LEU396ARG) in OPA1 is associated with a severe phenotype in a large dominant optic atrophy pedigree. Eye, 2018, 32, 843-845.	2.1	2
140	Evaluation of sFLT1 protein levels in human eyes with the FLT1 rs9943922 polymorphism. Ophthalmic Genetics, 2018, 39, 68-72.	1.2	2
141	Aqueous Misdirection After Trabeculectomy in a Down Syndrome Patient With Angle-closure Glaucoma. Journal of Glaucoma, 2021, 30, e269-e270.	1.6	2
142	Familial Glaucomaâ€"A Pedigree Revisited With Genetic Testing After 70 Years. JAMA Ophthalmology, 2022, 140, 543.	2.5	2
143	Changes in quantitative 3D shape features of the optic nerve head associated with age. Proceedings of SPIE, 2013, , .	0.8	1
144	Progressive optic nerve changes in cavitary optic disc anomaly: integration of copy number alteration and cis-expression quantitative trait loci to assess disease etiology. BMC Medical Genetics, 2019, 20, 63.	2.1	1

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145	Early-onset glaucoma., 2020,, 95-116.		1
146	Progressive Optic Disc Cupping Over 20 Years in a Patient with TBK1-Associated Glaucoma. Ophthalmology Glaucoma, 2020, 3, 167-168.	1.9	0
147	High Iris Insertion in Axenfeld-Rieger Syndrome. Ophthalmology, 2020, 127, 768.	5.2	O
148	Recombinant adenovirus causes prolonged mobilization of macrophages in the anterior chambers of mice Molecular Vision, 2021, 27, 741-756.	1.1	0