

Arthur Ab Bergen

List of Publications by Year in descending order

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144
papers

12,077
citations

26630

56
h-index

27406

106
g-index

146
all docs

146
docs citations

146
times ranked

10223
citing authors

#	ARTICLE	IF	CITATIONS
1	CRB1-Associated Retinal Dystrophies: A Prospective Natural History Study in Anticipation of Future Clinical Trials. <i>American Journal of Ophthalmology</i> , 2022, 234, 37-48.	3.3	17
2	Stargardt disease: monitoring incidence and diagnostic trends in the Netherlands using a nationwide disease registry. <i>Acta Ophthalmologica</i> , 2022, 100, 395-402.	1.1	10
3	X-Linked Retinoschisis. <i>Ophthalmology</i> , 2022, 129, 191-202.	5.2	29
4	The Phenotypic and Mutational Spectrum of the FHONDA Syndrome and Oculocutaneous Albinism: Similarities and Differences. , 2022, 63, 19.		12
5	Circadian clocks, retinogenesis and ocular health in vertebrates: new molecular insights. <i>Developmental Biology</i> , 2022, 484, 40-56.	2.0	5
6	The Natural History of Leber Congenital Amaurosis and Cone-Rod Dystrophy Associated with Variants in the GUCY2D Gene. <i>Ophthalmology Retina</i> , 2022, 6, 711-722.	2.4	8
7	Sodium-Iodate Injection Can Replicate Retinal Degenerative Disease Stages in Pigmented Mice and Rats: Non-Invasive Follow-Up Using OCT and ERG. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2918.	4.1	9
8	Bioinformatic Prioritization and Functional Annotation of GWAS-Based Candidate Genes for Primary Open-Angle Glaucoma. <i>Genes</i> , 2022, 13, 1055.	2.4	12
9	An alternative approach to produce versatile retinal organoids with accelerated ganglion cell development. <i>Scientific Reports</i> , 2021, 11, 1101.	3.3	16
10	Defining inclusion criteria and endpoints for clinical trials: a prospective cross-sectional study in <i>CRB1</i> -associated retinal dystrophies. <i>Acta Ophthalmologica</i> , 2021, 99, e402-e414.	1.1	10
11	The Role of Small Molecules and Their Effect on the Molecular Mechanisms of Early Retinal Organoid Development. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7081.	4.1	23
12	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6419.	4.1	8
13	Core circadian clock genes <i>Per1</i> and <i>Per2</i> regulate the rhythm in photoreceptor outer segment phagocytosis. <i>FASEB Journal</i> , 2021, 35, e21722.	0.5	17
14	The <i>Lrat</i> ^{−/−} Rat: CRISPR/Cas9 Construction and Phenotyping of a New Animal Model for Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7234.	4.1	6
15	Genome-wide CNV investigation suggests a role for cadherin, Wnt, and p53 pathways in primary open-angle glaucoma. <i>BMC Genomics</i> , 2021, 22, 590.	2.8	10
16	The Phenotypic Spectrum of Patients with PHARC Syndrome Due to Variants in ABHD12: An Ophthalmic Perspective. <i>Genes</i> , 2021, 12, 1404.	2.4	7
17	CLINICAL CHARACTERISTICS AND NATURAL HISTORY OF RHO-ASSOCIATED RETINITIS PIGMENTOSA. <i>Retina</i> , 2021, 41, 213-223.	1.7	18
18	Dark-adapted light response in mice is regulated by a circadian clock located in rod photoreceptors. <i>Experimental Eye Research</i> , 2021, 213, 108807.	2.6	5

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19	Mitochondrial Genome Study Identifies Association Between Primary Open-Angle Glaucoma and Variants in MT-CYB, MT-ND4 Genes and Haplogroups. <i>Frontiers in Genetics</i> , 2021, 12, 781189.	2.3	13
20	The circadian clock regulates RPE-mediated lactate transport via SLC16A1 (MCT1). <i>Experimental Eye Research</i> , 2020, 190, 107861.	2.6	13
21	Core-clock genes Period 1 and 2 regulate visual cascade and cell cycle components during mouse eye development. <i>Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms</i> , 2020, 1863, 194623.	1.9	10
22	RPGR-Associated Dystrophies: Clinical, Genetic, and Histopathological Features. <i>International Journal of Molecular Sciences</i> , 2020, 21, 835.	4.1	23
23	A Systematic Review on Transplantation Studies of the Retinal Pigment Epithelium in Animal Models. <i>International Journal of Molecular Sciences</i> , 2020, 21, 2719.	4.1	19
24	Rev-Erb β and Photoreceptor Outer Segments modulate the Circadian Clock in Retinal Pigment Epithelial Cells. <i>Scientific Reports</i> , 2019, 9, 11790.	3.3	14
25	Long-Term Follow-Up of Retinal Degenerations Associated With<i>LRAT</i> Mutations and Their Comparability to Phenotypes Associated With<i>RPE65</i> Mutations. <i>Translational Vision Science and Technology</i> , 2019, 8, 24.	2.2	14
26	Does the circadian clock make RPE-mediated ion transport "œtick" via SLC12A2 (NKCC1)? <i>Chronobiology International</i> , 2019, 36, 1592-1598.	2.0	5
27	On the origin of proteins in human drusen: The meet, greet and stick hypothesis. <i>Progress in Retinal and Eye Research</i> , 2019, 70, 55-84.	15.5	77
28	Increased High-Density Lipoprotein Levels Associated with Age-Related Macular Degeneration. <i>Ophthalmology</i> , 2019, 126, 393-406.	5.2	88
29	CLINICAL AND GENETIC CHARACTERISTICS OF MALE PATIENTS WITH RPGR-ASSOCIATED RETINAL DYSTROPHIES. <i>Retina</i> , 2019, 39, 1186-1199.	1.7	56
30	Systemic and Ocular Determinants of Peripapillary Retinal Nerve Fiber Layer Thickness Measurements in the European Eye Epidemiology (E3) Population. <i>Ophthalmology</i> , 2018, 125, 1526-1536.	5.2	62
31	The Decreasing Prevalence of Nonrefractive Visual Impairment in Older Europeans. <i>Ophthalmology</i> , 2018, 125, 1149-1159.	5.2	20
32	LONG-TERM FOLLOW-UP OF PATIENTS WITH CHOROIDEREMIA WITH SCLERAL PITS AND TUNNELS AS A NOVEL OBSERVATION. <i>Retina</i> , 2018, 38, 1713-1724.	1.7	11
33	P1"291: BINDING PROPERTIES OF CURCLUMIN IN POSTMORTEM BRAIN TISSUE: TOWARD AMYLOID IMAGING IN THE RETINA?. <i>Alzheimer's and Dementia</i> , 2018, 14, P397.	0.8	0
34	P2"251: NEUROPATHOLOGICAL HALLMARKS OF ALZHEIMER'S DISEASE IN POSTMORTEM AD RETINAS. <i>Alzheimer's and Dementia</i> , 2018, 14, P770.	0.8	0
35	The Phenotypic Spectrum of Albinism. <i>Ophthalmology</i> , 2018, 125, 1953-1960.	5.2	78
36	Ocular albinism with infertility and late"onset sensorineural hearing loss. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1587-1593.	1.2	6

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37	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. <i>Nature Genetics</i> , 2018, 50, 834-848.	21.4	239
38	Delineation of Novel Autosomal Recessive Mutation in GJA3 and Autosomal Dominant Mutations in GJA8 in Pakistani Congenital Cataract Families. <i>Genes</i> , 2018, 9, 112.	2.4	19
39	The Spectrum of Structural and Functional Abnormalities in Female Carriers of Pathogenic Variants in the <i>RPGR</i> Gene. , 2018, 59, 4123.		41
40	Genotypic and Phenotypic Characteristics of CRB1 -Associated Retinal Dystrophies. <i>Ophthalmology</i> , 2017, 124, 884-895.	5.2	75
41	Development of Refractive Errors—What Can We Learn From Inherited Retinal Dystrophies?. <i>American Journal of Ophthalmology</i> , 2017, 182, 81-89.	3.3	61
42	Prevalence of Age-Related Macular Degeneration in Europe. <i>Ophthalmology</i> , 2017, 124, 1753-1763.	5.2	337
43	Nicotinamide, iPPE-in-a dish, and age-related macular degeneration therapy development. <i>Stem Cell Investigation</i> , 2017, 4, 81-81.	3.0	4
44	Comparative gene expression study and pathway analysis of the human iris- and the retinal pigment epithelium. <i>PLoS ONE</i> , 2017, 12, e0182983.	2.5	9
45	Ophthalmic epidemiology in Europe: the “European Eye Epidemiology” (E3) consortium. <i>European Journal of Epidemiology</i> , 2016, 31, 197-210.	5.7	32
46	Visual Prognosis in USH2A-Associated Retinitis Pigmentosa Is Worse for Patients with Usher Syndrome Type IIa Than for Those with Nonsyndromic Retinitis Pigmentosa. <i>Ophthalmology</i> , 2016, 123, 1151-1160.	5.2	76
47	Gene expression and functional annotation of human choroid plexus epithelium failure in Alzheimer’s disease. <i>BMC Genomics</i> , 2015, 16, 956.	2.8	48
48	Systematic review of the association between Alzheimer’s disease and chronic glaucoma. <i>Clinical Ophthalmology</i> , 2015, 9, 783.	1.8	4
49	Comparison of Mouse and Human Retinal Pigment Epithelium Gene Expression Profiles: Potential Implications for Age-Related Macular Degeneration. <i>PLoS ONE</i> , 2015, 10, e0141597.	2.5	47
50	Cyclosporine A Treatment Inhibits Abcc6-Dependent Cardiac Necrosis and Calcification following Cocksackievirus B3 Infection in Mice. <i>PLoS ONE</i> , 2015, 10, e0138222.	2.5	10
51	ABCC6-Mediated ATP Secretion by the Liver Is the Main Source of the Mineralization Inhibitor Inorganic Pyrophosphate in the Systemic Circulation—Brief Report. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2014, 34, 1985-1989.	2.4	246
52	Retinitis Pigmentosa, Cutis Laxa, and Pseudoxanthoma Elasticum—Like Skin Manifestations Associated with GGCX Mutations. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2331-2338.	0.7	20
53	Human ciliary epithelia do express genes with retinal progenitor cell characteristics in vivo. <i>Experimental Eye Research</i> , 2014, 121, 41.	2.6	0
54	Gene expression-based comparison of the human secretory neuroepithelia of the brain choroid plexus and the ocular ciliary body: potential implications for glaucoma. <i>Fluids and Barriers of the CNS</i> , 2014, 11, 2.	5.0	20

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55	The Level of Hepatic ABCC6 Expression Determines the Severity of Calcification after Cardiac Injury. <i>American Journal of Pathology</i> , 2014, 184, 159-170.	3.8	28
56	The vast complexity of primary open angle glaucoma: Disease genes, risks, molecular mechanisms and pathobiology. <i>Progress in Retinal and Eye Research</i> , 2013, 37, 31-67.	15.5	149
57	Genotype and Phenotype of 101 Dutch Patients with Congenital Stationary Night Blindness. <i>Ophthalmology</i> , 2013, 120, 2072-2081.	5.2	95
58	Autosomal Recessive Bestrophinopathy. <i>Ophthalmology</i> , 2013, 120, 809-820.	5.2	115
59	Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. <i>Nature Genetics</i> , 2013, 45, 314-318.	21.4	398
60	Physiological evidence for impairment in autosomal dominant optic atrophy at the pre-ganglion level. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2013, 251, 221-234.	1.9	11
61	Expanded Clinical Spectrum of Enhanced S-Cone Syndrome. <i>JAMA Ophthalmology</i> , 2013, 131, 1324.	2.5	59
62	ABCC6 prevents ectopic mineralization seen in pseudoxanthoma elasticum by inducing cellular nucleotide release. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 20206-20211.	7.1	218
63	In silico analysis of the molecular machinery underlying aqueous humor production: potential implications for glaucoma. <i>Journal of Clinical Bioinformatics</i> , 2013, 3, 21.	1.2	19
64	Pseudoxanthoma Elasticum: Cardiac Findings in Patients and Abcc6-Deficient Mouse Model. <i>PLoS ONE</i> , 2013, 8, e68700.	2.5	32
65	Gene Expression and Functional Annotation of the Human and Mouse Choroid Plexus Epithelium. <i>PLoS ONE</i> , 2013, 8, e83345.	2.5	50
66	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. <i>PLoS ONE</i> , 2013, 8, e51622.	2.5	16
67	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. <i>International Journal of Epidemiology</i> , 2012, 41, 250-262.	1.9	79
68	Abcc6 deficiency in the mouse leads to calcification of collagen fibers in Bruch's membrane. <i>Experimental Eye Research</i> , 2012, 104, 59-64.	2.6	12
69	Clinical Course, Genetic Etiology, and Visual Outcome in Cone and Cone-Rod Dystrophy. <i>Ophthalmology</i> , 2012, 119, 819-826.	5.2	115
70	Incidental finding of alpha-methylacyl-CoA racemase deficiency in a patient with oculocutaneous albinism type 4. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2931-2934.	1.2	8
71	GPR179 Is Required for Depolarizing Bipolar Cell Function and Is Mutated in Autosomal-Recessive Complete Congenital Stationary Night Blindness. <i>American Journal of Human Genetics</i> , 2012, 90, 331-339.	6.2	131
72	Simultaneous Mutation Detection in 90 Retinal Disease Genes in Multiple Patients Using a Custom-designed 300-kb Retinal Resequencing Chip. <i>Ophthalmology</i> , 2011, 118, 160-167.e3.	5.2	25

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73	Clinical course of cone dystrophy caused by mutations in the RPGR gene. Graefe's Archive for Clinical and Experimental Ophthalmology, 2011, 249, 1527-1535.	1.9	36
74	Vitamin K supplementation increases vitamin K tissue levels but fails to counteract ectopic calcification in a mouse model for pseudoxanthoma elasticum. Journal of Molecular Medicine, 2011, 89, 1125-1135.	3.9	45
75	<i>Abcc6</i> Deficiency Causes Increased Infarct Size and Apoptosis in a Mouse Cardiac Ischemia-Reperfusion Model. Arteriosclerosis, Thrombosis, and Vascular Biology, 2011, 31, 2806-2812.	2.4	38
76	Common genetic variants associated with open-angle glaucoma. Human Molecular Genetics, 2011, 20, 2464-2471.	2.9	152
77	Dietary magnesium, not calcium, prevents vascular calcification in a mouse model for pseudoxanthoma elasticum. Journal of Molecular Medicine, 2010, 88, 467-475.	3.9	58
78	Proposal for updating the pseudoxanthoma elasticum classification system and a review of the clinical findings. American Journal of Medical Genetics, Part A, 2010, 152A, 1049-1058.	1.2	128
79	A genome-wide association study identifies a susceptibility locus for refractive errors and myopia at 15q14. Nature Genetics, 2010, 42, 897-901.	21.4	200
80	A New Strategy to Identify and Annotate Human RPE-Specific Gene Expression. PLoS ONE, 2010, 5, e9341.	2.5	72
81	A Genome-Wide Association Study of Optic Disc Parameters. PLoS Genetics, 2010, 6, e1000978.	3.5	187
82	The Complement Component 5 Gene and Age-Related Macular Degeneration. Ophthalmology, 2010, 117, 500-511.	5.2	36
83	Course of Visual Decline in Relation to the Best1 Genotype in Vitelliform Macular Dystrophy. Ophthalmology, 2010, 117, 1415-1422.	5.2	32
84	Pseudoxanthoma elasticum: Wide phenotypic variation in homozygotes and no signs in heterozygotes for the c.3775delT mutation in ABCC6. Genetics in Medicine, 2009, 11, 852-858.	2.4	30
85	Functional annotation of the human retinal pigment epithelium transcriptome. BMC Genomics, 2009, 10, 164.	2.8	52
86	Mutations in TRPM1 Are a Common Cause of Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2009, 85, 730-736.	6.2	193
87	Complement Component C3 and Risk of Age-Related Macular Degeneration. Ophthalmology, 2009, 116, 474-480.e2.	5.2	89
88	The SERPING1 gene and age-related macular degeneration. Lancet, The, 2009, 374, 875-876.	13.7	25
89	Geographic atrophy in age-related macular degeneration and TLR3. New England Journal of Medicine, 2009, 360, 2252-4; author reply 2255-6.	27.0	25
90	The SERPING1 gene and age-related macular degeneration. Lancet, The, 2008, 372, 1788-1789.	13.7	5

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91	Comprehensive Analysis of the Candidate Genes <i>CCL2</i> , <i>CCR2</i> , and <i>TLR4</i> in Age-Related Macular Degeneration. , 2008, 49, 364.		58
92	Retinal Degeneration and Ionizing Radiation Hypersensitivity in a Mouse Model for Cockayne Syndrome. <i>Molecular and Cellular Biology</i> , 2007, 27, 1433-1441.	2.3	69
93	GAP-43 expression is upregulated in retinal ganglion cells after ischemia/reperfusion-induced damage. <i>Experimental Eye Research</i> , 2007, 84, 858-867.	2.6	49
94	A Common Polymorphism in the Complement Factor H Gene Is Associated With Increased Risk of Myocardial Infarction. <i>Journal of the American College of Cardiology</i> , 2006, 47, 1568-1575.	2.8	83
95	Pseudoxanthoma Elasticum: the End of the Autosomal Dominant Segregation Myth. <i>Journal of Investigative Dermatology</i> , 2006, 126, 704-705.	0.7	16
96	Reduced secretion of fibulin 5 in age-related macular degeneration and cutis laxa. <i>Human Mutation</i> , 2006, 27, 568-574.	2.5	73
97	Complement Factor H Polymorphism, Complement Activators, and Risk of Age-Related Macular Degeneration. <i>JAMA - Journal of the American Medical Association</i> , 2006, 296, 301.	7.4	306
98	Disruption of <i>Abcc6</i> in the mouse: novel insight in the pathogenesis of pseudoxanthoma elasticum. <i>Human Molecular Genetics</i> , 2005, 14, 1763-1773.	2.9	184
99	Identification of mutations in the <i>AIPL1</i> , <i>CRB1</i> , <i>GUCY2D</i> , <i>RPE65</i> , and <i>RPGRIP1</i> genes in patients with juvenile retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2005, 42, e67-e67.	3.2	77
100	Patients with premature coronary artery disease who carry the <i>ABCC6</i> R1141X mutation have no Pseudoxanthoma Elasticum phenotype. <i>International Journal of Cardiology</i> , 2005, 100, 389-393.	1.7	20
101	Circadian expression of clock genes and clock-controlled genes in the rat retina. <i>Biochemical and Biophysical Research Communications</i> , 2005, 330, 18-26.	2.1	95
102	Efficient Molecular Diagnostic Strategy for <i>ABCC6</i> in Pseudoxanthoma Elasticum. <i>Genetic Testing and Molecular Biomarkers</i> , 2004, 8, 292-300.	1.7	19
103	Does autosomal dominant pseudoxanthoma elasticum exist?. <i>American Journal of Medical Genetics Part A</i> , 2004, 126A, 403-412.	2.4	48
104	In patients with pseudoxanthoma elasticum a thicker and more elastic carotid artery is associated with elastin fragmentation and proteoglycans accumulation. <i>Ultrasound in Medicine and Biology</i> , 2004, 30, 1041-1048.	1.5	41
105	Evaluation of the <i>ARMD1</i> locus on 1q25 in patients with age-related maculopathy: genetic variation in laminin genes and in exon 104 of <i>HEMICENTIN-1</i> . <i>Ophthalmic Genetics</i> , 2004, 25, 111-119.	1.2	38
106	<i>ABCC6</i> / <i>MRP6</i> mutations: further insight into the molecular pathology of pseudoxanthoma elasticum. <i>European Journal of Human Genetics</i> , 2003, 11, 215-224.	2.8	57
107	Subcellular localization and N-glycosylation of human <i>ABCC6</i> , expressed in MDCKII cells. <i>Biochemical and Biophysical Research Communications</i> , 2003, 308, 263-269.	2.1	37
108	Pseudoxanthoma elasticum: a clinical, histopathological, and molecular update. <i>Survey of Ophthalmology</i> , 2003, 48, 424-438.	4.0	149

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109	Analysis of the Frequent R1141X Mutation in the ABCC6 Gene in Pseudoxanthoma Elasticum. , 2003, 44, 1824.		36
110	Frequent Mutation in the ABCC6 Gene (R1141X) Is Associated With a Strong Increase in the Prevalence of Coronary Artery Disease. Circulation, 2002, 106, 773-775.	1.6	124
111	Myocilin mutations in a population-based sample of cases with open-angle glaucoma: the Rotterdam Study. Graefe's Archive for Clinical and Experimental Ophthalmology, 2002, 240, 468-474.	1.9	27
112	MRP6 (ABCC6) Detection in Normal Human Tissues and Tumors. Laboratory Investigation, 2002, 82, 515-518.	3.7	458
113	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	3.8	31
114	A summary of 20 CACNA1F mutations identified in 36 families with incomplete X-linked congenital stationary night blindness, and characterization of splice variants. Human Genetics, 2001, 108, 91-97.	3.8	99
115	Mutations in ABCC6 cause pseudoxanthoma elasticum. Nature Genetics, 2000, 25, 228-231.	21.4	804
116	Mutations in NYX, encoding the leucine-rich proteoglycan nyctalopin, cause X-linked complete congenital stationary night blindness. Nature Genetics, 2000, 26, 319-323.	21.4	309
117	Mutations in a human homologue of Drosophila crumbs cause retinitis pigmentosa (RP12). Nature Genetics, 1999, 23, 217-221.	21.4	427
118	The mutation spectrum of the bestrophin protein - functional implications. Human Genetics, 1999, 104, 383-389.	3.8	100
119	Identification of a 5' splice site mutation in the RPGR gene in a family with X-linked retinitis pigmentosa (RP3). , 1999, 13, 141-145.		38
120	The 2588G→C Mutation in the ABCR Gene Is a Mild Frequent Founder Mutation in the Western European Population and Allows the Classification of ABCR Mutations in Patients with Stargardt Disease. American Journal of Human Genetics, 1999, 64, 1024-1035.	6.2	242
121	Retinitis Pigmentosa. Survey of Ophthalmology, 1999, 43, 321-334.	4.0	254
122	Pseudoxanthoma Elasticum Maps to an 820-kb Region of the p13.1 Region of Chromosome 16. Genomics, 1999, 62, 1-10.	2.9	61
123	Positional cloning of the gene for X-linked retinitis pigmentosa 2. Nature Genetics, 1998, 19, 327-332.	21.4	371
124	Identification of the gene responsible for Best macular dystrophy. Nature Genetics, 1998, 19, 241-247.	21.4	634
125	Autosomal recessive retinitis pigmentosa and cone-rod dystrophy caused by splice site mutations in the Stargardt's disease gene ABCR. Human Molecular Genetics, 1998, 7, 355-362.	2.9	475
126	Characterization of SCML1, a New Gene in Xp22, with Homology to Developmental Polycomb Genes. Genomics, 1998, 49, 96-102.	2.9	32

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127	A Gene for X-Linked Optic Atrophy Is Closely Linked to the Xp11.4-Xp11.2 Region of the X Chromosome. American Journal of Human Genetics, 1997, 61, 934-939.	6.2	60
128	Localization of a Novel X-Linked Progressive Cone Dystrophy Gene to Xq27: Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 1997, 60, 1468-1473.	6.2	43
129	A Locus for Autosomal Recessive Pseudoxanthoma Elasticum, with Penetrance of Vascular Symptoms in Carriers, Maps to Chromosome 16p13.1. Genome Research, 1997, 7, 830-834.	5.5	74
130	Positional cloning of the gene for X-linked retinitis pigmentosa 3: homology with the guanine-nucleotide-exchange factor RCC1. Human Molecular Genetics, 1996, 5, 1035-1041.	2.9	186
131	An Xp22.1-p22.2 YAC Contig Encompassing the Disease Loci for RS, KFSD, CLS, HYP and RP15: Refined Localization of RS. European Journal of Human Genetics, 1996, 4, 101-104.	2.8	19
132	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. Nature Genetics, 1995, 10, 13-19.	21.4	190
133	The Human Molecular Genetics Network. New England Journal of Medicine, 1995, 333, 1573-1573.	27.0	0
134	Localization of a novel X-linked congenital stationary night blindness locus: close linkage to the RP3 type retinitis pigmentosa gene region. Human Molecular Genetics, 1995, 4, 931-935.	2.9	11
135	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. Human Molecular Genetics, 1995, 4, 2319-2325.	2.9	80
136	X linked progressive cone dystrophy. Localisation of the gene locus to Xp21-p11.1 by linkage analysis.. British Journal of Ophthalmology, 1994, 78, 103-108.	3.9	23
137	A submicroscopic deletion in a patient with isolated X-linked ocular albinism (OA1). Human Molecular Genetics, 1994, 3, 647-648.	2.9	16
138	Nance-Horan Syndrome: Linkage Analysis in a Family from The Netherlands. Genomics, 1994, 21, 238-240.	2.9	15
139	Assignment of a Gene for Autosomal Recessive Retinitis Pigmentosa (RP12) to Chromosome 1q31-q32.1 in an Inbred and Genetically Heterogeneous Disease Population. Genomics, 1994, 22, 499-504.	2.9	123
140	Refinement of the Localization of the X-Linked Ocular Albinism Gene. Genomics, 1993, 16, 272-273.	2.9	15
141	Additional Evidence for a Gene Locus for Progressive Cone Dystrophy with Late Rod Involvement in Xp21.1-p11.3. Genomics, 1993, 18, 463-464.	2.9	16
142	Evidence for nonallelic genetic heterogeneity in autosomal recessive retinitis pigmentosa. Genomics, 1992, 14, 811-812.	2.9	4
143	Isolation of a candidate gene for Norrie disease by positional cloning. Nature Genetics, 1992, 1, 199-203.	21.4	239
144	Multipoint linkage analysis in X-linked ocular albinism of the Nettleship-Falls type. Human Genetics, 1991, 88, 162-166.	3.8	38