Sandro Banfi

List of Publications by Year
in descending order

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162 papers	18,269 citations	32410 55 h-index	15698 129 g-index
164 all docs	164 docs citations	164 times ranked	25152 citing authors

#	Article	IF	Citations
1	RPE65-Associated Retinopathies in the Italian Population: A Longitudinal Natural History Study. , 2022, 63, 13.		11
2	Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2021, 58, 570-578.	1.5	10
3	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	1.6	71
4	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. International Journal of Molecular Sciences, 2021, 22, 1681.	1.8	19
5	RPE65-associated inherited retinal diseases: consensus recommendations for eligibility to gene therapy. Orphanet Journal of Rare Diseases, 2021, 16, 257.	1.2	24
6	Inherited Retinal Diseases Due to RPE65 Variants: From Genetic Diagnostic Management to Therapy. International Journal of Molecular Sciences, 2021, 22, 7207.	1.8	19
7	Mild Clinical Presentation of Joubert Syndrome in a Male Adult Carrying Biallelic MKS1 Truncating Variants. Diagnostics, 2021, 11, 1218.	1.3	4
8	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3
9	Pupillometry via smartphone for low-resource settings. Biocybernetics and Biomedical Engineering, 2021, 41, 891-902.	3.3	9
10	Characteristics of Retinitis Pigmentosa Associated with ADGRV1 and Comparison with USH2A in Patients from a Multicentric Usher Syndrome Study Treatrush. International Journal of Molecular Sciences, 2021, 22, 10352.	1.8	3
11	Voretigene Neparvovec Gene Therapy in Clinical Practice: Treatment of the First Two Italian Pediatric Patients. Translational Vision Science and Technology, 2021, 10, 11.	1.1	15
12	VarGenius-HZD Allows Accurate Detection of Rare Homozygous or Hemizygous Deletions in Targeted Sequencing Leveraging Breadth of Coverage. Genes, 2021, 12, 1979.	1.0	4
13	ORÃO: RESTful Cloud-Based Ophthalmologic Medical Record for Chromatic Pupillometry. IFMBE Proceedings, 2020, , 713-720.	0.2	5
14	A collaborative RESTful cloud-based tool for management of chromatic pupillometry in a clinical trial. Health and Technology, 2020, 10, 25-38.	2.1	8
15	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	1.8	25
16	AAV-miR-204 Protects from Retinal Degeneration by Attenuation of Microglia Activation and Photoreceptor Cell Death. Molecular Therapy - Nucleic Acids, 2020, 19, 144-156.	2.3	28
17	Mutation-Independent Therapies for Retinal Diseases: Focus on Gene-Based Approaches. Frontiers in Neuroscience, 2020, 14, 588234.	1.4	9
18	Clinical Phenotype and Course of PDE6A-Associated Retinitis Pigmentosa Disease, Characterized in Preparation for a Gene Supplementation Trial. JAMA Ophthalmology, 2020, 138, 1241.	1.4	9

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19	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
20	The Pervasive Role of the miR-181 Family in Development, Neurodegeneration, and Cancer. International Journal of Molecular Sciences, 2020, 21, 2092.	1.8	93
21	Mild form of Zellweger Spectrum Disorders (ZSD) due to variants in PEX1: Detailed clinical investigation in a 9-years-old female. Molecular Genetics and Metabolism Reports, 2020, 24, 100615.	0.4	12
22	Lightâ€responsive microRNA miRâ€211 targets Ezrin to modulate lysosomal biogenesis and retinal cell clearance. EMBO Journal, 2020, 39, e102468.	3.5	30
23	Sophisticated Gene Regulation for a Complex Physiological System: The Role of Non-coding RNAs in Photoreceptor Cells. Frontiers in Cell and Developmental Biology, 2020, 8, 629158.	1.8	7
24	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17
25	Full-field electroretinography, visual acuity and visual fields in Usher syndrome: a multicentre European study. Documenta Ophthalmologica, 2019, 139, 151-160.	1.0	7
26	Efficacy, Safety, and Durability of Voretigene Neparvovec-rzyl in RPE65 Mutationâ€“Associated Inherited Retinal Dystrophy. Ophthalmology, 2019, 126, 1273-1285.	2.5	239
27	Microdeletion of pseudogene chr14.232.a affects LRFN5 expression in cells of a patient with autism spectrum disorder. European Journal of Human Genetics, 2019, 27, 1475-1480.	1.4	13
28	miRâ€181a/b downregulation exerts a protective action on mitochondrial disease models. EMBO Molecular Medicine, 2019, 11, .	3.3	58
29	Non-coding RNAs in retinal development and function. Human Genetics, 2019, 138, 957-971.	1.8	35
30	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10
31	Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. Genetics in Medicine, 2019, 21, 1319-1329.	1.1	15
32	ASSOCIATION BETWEEN GENOTYPE AND DISEASE PROGRESSION IN ITALIAN STARGARDT PATIENTS. Retina, 2019, 39, 1399-1409.	1.0	19
33	Toward a Novel Medical Device Based on Chromatic Pupillometry for Screening and Monitoring of Inherited Ocular Disease: A Pilot Study. IFMBE Proceedings, 2019, , 387-390.	0.2	5
34	Triple Vectors Expand AAV Transfer Capacity in the Retina. Molecular Therapy, 2018, 26, 524-541.	3.7	94
35	Usher Syndrome and Color Vision. Current Eye Research, 2018, 43, 1295-1301.	0.7	3
36	Intrafamilial heterogeneity of congenital optic disc pit maculopathy. Ophthalmic Genetics, 2017, 38, 267-272.	0.5	4

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37	CLINICAL PRESENTATION AND DISEASE COURSE OF USHER SYNDROME BECAUSE OF MUTATIONS IN MYO7A OR USH2A. Retina, 2017, 37, 1581-1590.	1.0	36
38	Next-Generation Sequencing Approaches to Define the Role of the Autophagy Lysosomal Pathway in Human Disease: The Example of LysoPlex. Methods in Molecular Biology, 2017, 1594, 227-241.	0.4	3
39	Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. European Journal of Human Genetics, 2017, 25, 651-655.	1.4	19
40	Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with RPE65 -mediated inherited retinal dystrophy: a randomised, controlled, open-label, phase 3 trial. Lancet, The, 2017, 390, 849-860.	6.3	1,250
41	MiR-211 is essential for adult cone photoreceptor maintenance and visual function. Scientific Reports, 2017, 7, 17004.	1.6	29
42	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. Genes, 2017, 8, 280.	1.0	23
43	Targeting and silencing of rhodopsin by ectopic expression of the transcription factor KLF15. JCI Insight, 2017, 2, .	2.3	12
44	En Face Spectral-Domain Optical Coherence Tomography for the Monitoring of Lesion Area Progression in Stargardt Disease. , 2016, 57, OCT247.		17
45	Safety and durability of effect of contralateral-eye administration of AAV2 gene therapy in patients with childhood-onset blindness caused by RPE65 mutations: a follow-on phase 1 trial. Lancet, The, 2016, 388, 661-672.	6.3	377
46	MIB2variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with MÃ©nÃ©trier-like gastropathy. Human Molecular Genetics, 2016, 26, ddw365.	1.4	7
47	Reproducibility of en-face Optical Coherence Tomography Imaging for Macular Atrophy Area Evaluation in Juvenile Macular Degeneration. IFMBE Proceedings, 2016, , 250-253.	0.2	1
48	An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. European Journal of Human Genetics, 2016, 24, 1730-1738.	1.4	77
49	Functional improvement assessed by multifocal electroretinogram after Ocriplasmin treatment for vitreomacular traction. BMC Ophthalmology, 2016, 16, 110.	0.6	7
50	An atlas of gene expression and gene co-regulation in the human retina. Nucleic Acids Research, 2016, 44, 5773-5784.	6.5	65
51	High-resolution analysis of the human retina miRNome reveals isomiR variations and novel microRNAs. Nucleic Acids Research, 2016, 44, 1525-1540.	6.5	98
52	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	9.4	50
53	Evaluation of Ocular Gene Therapy in an Italian Patient Affected by Congenital Leber Amaurosis Type 2 Treated in Both Eyes. Advances in Experimental Medicine and Biology, 2016, 854, 533-539.	0.8	6
54	Rhodopsin targeted transcriptional silencing by DNA-binding. ELife, 2016, 5, e12242.	2.8	33

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55	TGF-Î² Controls miR-181/ERK Regulatory Network during Retinal Axon Specification and Growth. PLoS ONE, 2015, 10, e0144129.	1.1	30
56	Gene Therapy of Inherited Retinal Degenerations: Prospects and Challenges. Human Gene Therapy, 2015, 26, 193-200.	1.4	39
57	Improved dual AAV vectors with reduced expression of truncated proteins are safe and effective in the retina of a mouse model of Stargardt disease. Human Molecular Genetics, 2015, 24, 6811-6825.	1.4	73
58	miRâ€•181a/b control the assembly of visual circuitry by regulating retinal axon specification and growth. Developmental Neurobiology, 2015, 75, 1252-1267.	1.5	22
59	MiR-204 is responsible for inherited retinal dystrophy associated with ocular coloboma. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E3236-45.	3.3	90
60	Lysoplex: An efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway. Autophagy, 2015, 11, 928-938.	4.3	47
61	Inherited Retinal Dystrophies: The role of gene expression regulators. International Journal of Biochemistry and Cell Biology, 2015, 61, 115-119.	1.2	7
62	Mutations in MFSD8, Encoding a Lysosomal Membrane Protein, Are Associated with Nonsyndromic Autosomal Recessive Macular Dystrophy. Ophthalmology, 2015, 122, 170-179.	2.5	60
63	miR-340 inhibits tumor cell proliferation and induces apoptosis by targeting multiple negative regulators of p27 in non-small cell lung cancer. Oncogene, 2015, 34, 3240-3250.	2.6	167
64	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
65	Macular abnormalities in Italian patients with retinitis pigmentosa. British Journal of Ophthalmology, 2014, 98, 946-950.	2.1	76
66	Cardiomyogenesis is controlled by the miR-99a/let-7c cluster and epigenetic modifications. Stem Cell Research, 2014, 12, 323-337.	0.3	57
67	Macular Function and Morphologic Features in Juvenile Stargardt Disease. Ophthalmology, 2014, 121, 2399-2405.	2.5	54
68	The combination of transcriptomics and informatics identifies pathways targeted by miR-204 during neurogenesis and axon guidance. Nucleic Acids Research, 2014, 42, 7793-7806.	6.5	31
69	The ADAMTS18 gene is responsible for autosomal recessive early onset severe retinal dystrophy. Orphanet Journal of Rare Diseases, 2013, 8, 16.	1.2	41
70	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	2.6	71
71	Combined Rod and Cone Transduction by Adeno-Associated Virus 2/8. Human Gene Therapy, 2013, 24, 982-992.	1.4	36
72	Recessive Mutations in SLC38A8 Cause Foveal Hypoplasia and Optic Nerve Misrouting without Albinism. American Journal of Human Genetics, 2013, 93, 1143-1150.	2.6	71

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73	Three-Year Follow-up after Unilateral Subretinal Delivery of Adeno-Associated Virus in Patients with Leber Congenital Amaurosis Type 2. Ophthalmology, 2013, 120, 1283-1291.	2.5	301
74	Non-coding RNAs in the development of sensory organs and related diseases. Cellular and Molecular Life Sciences, 2013, 70, 4141-4155.	2.4	29
75	Pax6 Regulates Gene Expression in the Vertebrate Lens through miR-204. PLoS Genetics, 2013, 9, e1003357.	1.5	86
76	Highly conserved elements discovered in vertebrates are present in non-syntenic loci of tunicates, act as enhancers and can be transcribed during development. Nucleic Acids Research, 2013, 41, 3600-3618.	6.5	24
77	Reverse engineering a mouse embryonic stem cell-specific transcriptional network reveals a new modulator of neuronal differentiation. Nucleic Acids Research, 2013, 41, 711-726.	6.5	24
78	miR-204 Targeting of Ankrd13A Controls Both Mesenchymal Neural Crest and Lens Cell Migration. PLoS ONE, 2013, 8, e61099.	1.1	30
79	Recombinant Vectors Based on Porcine Adeno-Associated Viral Serotypes Transduce the Murine and Pig Retina. PLoS ONE, 2013, 8, e59025.	1.1	13
80	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. PLoS ONE, 2013, 8, e51622.	1.1	16
81	The long noncoding RNA Vax2os1 controls the cell cycle progression of photoreceptor progenitors in the mouse retina. Rna, 2012, 18, 111-123.	1.6	91
82	Subretinal Fibrosis in StargardtÂ's Disease with Fundus Flavimaculatus and ABCA4 Gene Mutation. Case Reports in Ophthalmology, 2012, 3, 410-417.	0.3	17
83	BBS1 Mutations in a Wide Spectrum of Phenotypes Ranging From Nonsyndromic Retinitis Pigmentosa to Bardet-Biedl Syndrome. JAMA Ophthalmology, 2012, 130, 1425.	2.6	106
84	Identification of microRNA-regulated gene networks by expression analysis of target genes. Genome Research, 2012, 22, 1163-1172.	2.4	165
85	Pupillometric analysis for assessment of gene therapy in Leber Congenital Amaurosis patients. BioMedical Engineering OnLine, 2012, 11, 40.	1.3	27
86	AAV2 Gene Therapy Readministration in Three Adults with Congenital Blindness. Science Translational Medicine, 2012, 4, 120ra15.	5.8	340
87	Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. , 2012, 53, 4409.		62
88	The absence of dystrophin brain isoform expression in healthy human heart ventricles explains the pathogenesis of 5' X-linked dilated cardiomyopathy. BMC Medical Genetics, 2012, 13, 20.	2.1	20
89	Impact of Age at Administration, Lysosomal Storage, and Transgene Regulatory Elements on AAV2/8-Mediated Rat Liver Transduction. PLoS ONE, 2012, 7, e33286.	1.1	17
90	Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. PLoS ONE, 2012, 7, e43799.	1.1	29

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91	HOCTAR database: A unique resource for microRNA target prediction. Gene, 2011, 480, 51-58.	1.0	54
92	MicroRNA-Restricted Transgene Expression in the Retina. PLoS ONE, 2011, 6, e22166.	1.1	55
93	Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. , 2011, 52, 5618.		41
94	Vax2 regulates retinoic acid distribution and cone opsin expression in the vertebrate eye. Development (Cambridge), 2011, 138, 261-271.	1.2	39
95	A High-Resolution Anatomical Atlas of the Transcriptome in the Mouse Embryo. PLoS Biology, 2011, 9, e1000582.	2.6	552
96	The human visual cortex responds to gene therapyâ€“mediated recovery of retinal function. Journal of Clinical Investigation, 2011, 121, 2160-2168.	3.9	121
97	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	2.6	98
98	miRNeye: a microRNA expression atlas of the mouse eye. BMC Genomics, 2010, 11, 715.	1.2	140
99	UTRdb and UTRsite (RELEASE 2010): a collection of sequences and regulatory motifs of the untranslated regions of eukaryotic mRNAs. Nucleic Acids Research, 2010, 38, D75-D80.	6.5	285
100	miR-204 is required for lens and retinal development via Meis2 targeting. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 15491-15496.	3.3	151
101	Promiscuity of enhancer, coding and non-coding transcription functions in ultraconserved elements. BMC Genomics, 2010, 11, 151.	1.2	32
102	A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. Genome Biology, 2010, 11, R64.	13.9	16
103	Gene Therapy for Leber's Congenital Amaurosis is Safe and Effective Through 1.5 Years After Vector Administration. Molecular Therapy, 2010, 18, 643-650.	3.7	503
104	MicroRNA target prediction by expression analysis of host genes. Genome Research, 2009, 19, 481-490.	2.4	168
105	A Homozygous Missense Mutation in theIRBPGene (RBP3) Associated with Autosomal Recessive Retinitis Pigmentosa. , 2009, 50, 1864.		93
106	Two novel CYP7B1 mutations in Italian families with SPG5: a clinical and genetic study. Journal of Neurology, 2009, 256, 1252-1257.	1.8	39
107	A Gene Network Regulating Lysosomal Biogenesis and Function. Science, 2009, 325, 473-477.	6.0	1,958
108	Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial. Lancet, The, 2009, 374, 1597-1605.	6.3	774

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109	microRNAs and genetic diseases. PathoGenetics, 2009, 2, 7.	5.7	140
110	Screening for POLG1 mutations in a Southern Italian ataxia population. Journal of Neurology, 2008, 255, 454-455.	1.8	2
111	CoGemiR: A comparative genomics microRNA database. BMC Genomics, 2008, 9, 457.	1.2	35
112	Sonic Hedgehog deletion and distal trisomy 3p in a patient with microphthalmia and microcephaly, lacking cerebral anomalies typical of holoprosencephaly. European Journal of Medical Genetics, 2008, 51, 658-665.	0.7	14
113	Safety and Efficacy of Gene Transfer for Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2240-2248.	13.9	1,941
114	A High-Resolution RNA Expression Atlas of Retinitis Pigmentosa Genes in Human and Mouse Retinas. , 2008, 49, 2330.		24
115	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
116	Identification and expression analysis of novel Jakmip1 transcripts. Gene, 2007, 402, 1-8.	1.0	15
117	Identification and Characterization of MicroRNAs Expressed in the Mouse Eye. , 2007, 48, 509.		179
118	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	1.5	94
119	Ataxia with oculomotor apraxia type 2: A clinical, pathologic, and genetic study. Neurology, 2006, 66, 1207-1210.	1.5	114
120	An Autoregulatory Loop Directs the Tissue-Specific Expression of p63 through a Long-Range Evolutionarily Conserved Enhancer. Molecular and Cellular Biology, 2006, 26, 3308-3318.	1.1	73
121	A novel GJA1 mutation causes oculodentodigital dysplasia without syndactyly. American Journal of Medical Genetics, Part A, 2005, 133A, 58-60.	0.7	55
122	Very late onset in ataxia oculomotor apraxia type I. Annals of Neurology, 2005, 57, 777-777.	2.8	133
123	Novel mutation ofSACSgene in a Spanish family with autosomal recessive spastic ataxia. Movement Disorders, 2005, 20, 1358-1361.	2.2	49
124	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	1.8	8
125	Natural antisense transcripts associated with genes involved in eye development. Human Molecular Genetics, 2005, 14, 913-923.	1.4	101
126	Genotype-Phenotype Correlation in Italian Families with Stargardt Disease. Ophthalmic Research, 2005, 37, 159-167.	1.0	42

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127	Transcriptional regulation by Barhl1 and Brn-3c in organ of corti derived cell lines. Molecular Brain Research, 2005, 141, 174-180.	2.5	5
128	DG-CST (Disease Gene Conserved Sequence Tags), a database of human-mouse conserved elements associated to disease genes. Nucleic Acids Research, 2004, 33, D505-D510.	6.5	11
129	Pcp4l1, a novel gene encoding a Pcp4-like polypeptide, is expressed in specific domains of the developing brain. Gene Expression Patterns, 2004, 4, 297-301.	0.3	20
130	Identification and characterisation of the retinitis pigmentosa 1-like1 gene (RP1L1): a novel candidate for retinal degenerations. European Journal of Human Genetics, 2003, 11, 155-162.	1.4	39
131	Identification and characterization of C1orf36, a transcript highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Biochemical and Biophysical Research Communications, 2003, 308, 414-421.	1.0	13
132	Characterization of the OFD1/Ofd1 genes on the human and mouse sex chromosomes and exclusion of Ofd1 for the Xpl mouse mutantâ¯†. Genomics, 2003, 81, 560-569.	1.3	40
133	Clinical features of X linked juvenile retinoschisis associated with new mutations in the XLRS1 gene in Italian families. British Journal of Ophthalmology, 2003, 87, 1130-1134.	2.1	43
134	A 76-kb duplicon maps close to the BCR gene on chromosome 22 and the ABL gene on chromosome 9: Possible involvement in the genesis of the Philadelphia chromosome translocation. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 9882-9887.	3.3	71
135	Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Gene, 2002, 297, 33-38.	1.0	14
136	Human chromosome 21 gene expression atlas in the mouse. Nature, 2002, 420, 582-586.	13.7	208
137	Vax2inactivation in mouse determines alteration of the eye dorsal-ventral axis, misrouting of the optic fibres and eye coloboma. Development (Cambridge), 2002, 129, 805-813.	1.2	111
138	Vax2 inactivation in mouse determines alteration of the eye dorsal-ventral axis, misrouting of the optic fibres and eye coloboma. Development (Cambridge), 2002, 129, 805-13.	1.2	53
139	Expression of the Xvax2 gene demarcates presumptive ventral telencephalon and specific visual structures in Xenopus laevis. Mechanisms of Development, 2001, 100, 115-118.	1.7	22
140	Proprioceptor Pathway Development Is Dependent on MATH1. Neuron, 2001, 30, 411-422.	3.8	280
141	Identification and Characterization of YME1L1, a Novel Paraplegin-Related Gene. Genomics, 2000, 66, 48-54.	1.3	58
142	Cloning of PC3B, a Novel Member of the PC3/BTG/TOB Family of Growth Inhibitory Genes, Highly Expressed in the Olfactory Epithelium. Genomics, 2000, 68, 253-263.	1.3	66
143	EYA4, a novel vertebrate gene related to Drosophila eyes absent. Human Molecular Genetics, 1999, 8, 11-23.	1.4	149
144	A homeobox gene, vax2, controls the patterning of the eye dorsoventral axis. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 10729-10734.	3.3	144

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145	Identification and Characterization of CDS2, a Mammalian Homolog of theDrosophilaCDP-diacylglycerol Synthase Gene. Genomics, 1999, 55, 68-77.	1.3	40
146	Identification and Characterization of AFG3L2, a Novel Paraplegin-Related Gene. Genomics, 1999, 59, 51-58.	1.3	70
147	KCNE1-like Gene Is Deleted in AMME Contiguous Gene Syndrome: Identification and Characterization of the Human and Mouse Homologs. Genomics, 1999, 60, 251-257.	1.3	72
148	How to get the best of dbEST. Trends in Genetics, 1998, 14, 80-81.	2.9	27
149	A Human Homologue of the Drosophila melanogaster diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. American Journal of Human Genetics, 1998, 62, 533-541.	2.6	248
150	Characterization ofCxorf5(71-7A), a Novel Human cDNA Mapping to Xp22 and Encoding a Protein Containing Coiled-Coil Î±-Helical Domains. Genomics, 1998, 51, 243-250.	1.3	56
151	Sequencing Analysis of Forty-Eight Human Image cDNA Clones Similar to Drosophila Mutant Protein. DNA Sequence, 1998, 9, 307-315.	0.7	5
152	A practical guide to orient yourself in the labyrinth of genome databases. Human Molecular Genetics, 1998, 7, 1641-1648.	1.4	17
153	Mice Lacking Ataxin-1 Display Learning Deficits and Decreased Hippocampal Paired-Pulse Facilitation. Journal of Neuroscience, 1998, 18, 5508-5516.	1.7	197
154	Drosophila-related expressed sequences. Human Molecular Genetics, 1997, 6, 1745-1753.	1.4	22
155	A Novel Zinc Finger-Containing RNA-Binding Protein Conserved from Fruitflies to Humans. Genomics, 1997, 41, 444-452.	1.3	27
156	DRES search engine: of flies, men and ESTs. Trends in Genetics, 1997, 13, 79.	2.9	5
157	Identification and mapping of human cDNAs homologous to Drosophila mutant genes through EST database searching. Nature Genetics, 1996, 13, 167-174.	9.4	177
158	Cloning and Developmental Expression Analysis of the Murine Homolog of the Spinocerebellar Ataxia Type 1 Gene (Sca1). Human Molecular Genetics, 1996, 5, 33-40.	1.4	59
159	Identification and characterization of the gene causing type 1 spinocerebellar ataxia. Nature Genetics, 1994, 7, 513-520.	9.4	362
160	Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1. Nature Genetics, 1993, 4, 221-226.	9.4	1,673
161	Mapping and cloning of the critical region for the spinocerebellar ataxia type 1 gene (SCA1) in a yeast artificial chromosome contig spanning 1.2 Mb. Genomics, 1993, 18, 627-635.	1.3	42
162	An easy and rapid method for the detection of chimeric yeast artificial chromosome clones. Nucleic Acids Research, 1992, 20, 1814-1814.	6.5	8