

Anthony P Monaco

List of Publications by Year in descending order

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317
papers

47,213
citations

2203

99
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1895

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all docs

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docs citations

324
times ranked

31748
citing authors

#	ARTICLE	IF	CITATIONS
1	Complete cloning of the duchenne muscular dystrophy (DMD) cDNA and preliminary genomic organization of the DMD gene in normal and affected individuals. <i>Cell</i> , 1987, 50, 509-517.	13.5	2,324
2	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013, 45, 984-994.	9.4	2,067
3	A forkhead-domain gene is mutated in a severe speech and language disorder. <i>Nature</i> , 2001, 413, 519-523.	13.7	1,969
4	Functional impact of global rare copy number variation in autism spectrum disorders. <i>Nature</i> , 2010, 466, 368-372.	13.7	1,803
5	The complete sequence of dystrophin predicts a rod-shaped cytoskeletal protein. <i>Cell</i> , 1988, 53, 219-228.	13.5	1,509
6	Molecular evolution of FOXP2, a gene involved in speech and language. <i>Nature</i> , 2002, 418, 869-872.	13.7	1,481
7	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007, 39, 319-328.	9.4	1,272
8	An explanation for the phenotypic differences between patients bearing partial deletions of the DMD locus. <i>Genomics</i> , 1988, 2, 90-95.	1.3	1,103
9	Isolation of candidate cDNAs for portions of the Duchenne muscular dystrophy gene. <i>Nature</i> , 1986, 323, 646-650.	13.7	1,050
10	Cloning the gene for an inherited human disorderâ€”chronic granulomatous diseaseâ€”on the basis of its chromosomal location. <i>Nature</i> , 1986, 322, 32-38.	13.7	833
11	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2014, 94, 677-694.	2.6	819
12	An unusual member of the nuclear hormone receptor superfamily responsible for X-linked adrenal hypoplasia congenita. <i>Nature</i> , 1994, 372, 635-641.	13.7	796
13	Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. <i>Nature</i> , 1994, 372, 672-676.	13.7	722
14	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. <i>Nature Genetics</i> , 1993, 3, 14-19.	9.4	708
15	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. <i>Nature Neuroscience</i> , 2015, 18, 199-209.	7.1	701
16	Mutations in ATP2A2, encoding a Ca ²⁺ pump, cause Darier disease. <i>Nature Genetics</i> , 1999, 21, 271-277.	9.4	697
17	A Functional Genetic Link between Distinct Developmental Language Disorders. <i>New England Journal of Medicine</i> , 2008, 359, 2337-2345.	13.9	626
18	A genome-wide linkage and association scan reveals novel loci for autism. <i>Nature</i> , 2009, 461, 802-808.	13.7	570

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19	A genome-wide scan for common alleles affecting risk for autism. <i>Human Molecular Genetics</i> , 2010, 19, 4072-4082.	1.4	538
20	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. <i>PLoS Genetics</i> , 2014, 10, e1004580.	1.5	501
21	Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. <i>Nature</i> , 1986, 322, 73-77.	13.7	475
22	Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. <i>Nature Genetics</i> , 2000, 25, 17-19.	9.4	462
23	Localisation of a gene implicated in a severe speech and language disorder. <i>Nature Genetics</i> , 1998, 18, 168-170.	9.4	447
24	A Genomewide Screen for Autism: Strong Evidence for Linkage to Chromosomes 2q, 7q, and 16p. <i>American Journal of Human Genetics</i> , 2001, 69, 570-581.	2.6	439
25	Identification of FOXP2 Truncation as a Novel Cause of Developmental Speech and Language Deficits. <i>American Journal of Human Genetics</i> , 2005, 76, 1074-1080.	2.6	438
26	Specific cloning of DNA fragments absent from the DNA of a male patient with an X chromosome deletion.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1985, 82, 4778-4782.	3.3	432
27	Association between X-linked mixed deafness and mutations in the POU domain gene POU3F4. <i>Science</i> , 1995, 267, 685-688.	6.0	423
28	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. <i>Nature Genetics</i> , 2017, 49, 978-985.	9.4	401
29	Self-injection of amphetamine directly into the brain. <i>Psychopharmacology</i> , 1983, 81, 158-163.	1.5	399
30	Detection of deletions spanning the Duchenne muscular dystrophy locus using a tightly linked DNA segment. <i>Nature</i> , 1985, 316, 842-845.	13.7	396
31	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. <i>PLoS Genetics</i> , 2012, 8, e1002521.	1.5	358
32	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. <i>Nature Genetics</i> , 2001, 28, 119-120.	9.4	357
33	Yeast artificial chromosome libraries containing large inserts from mouse and human DNA.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1991, 88, 4123-4127.	3.3	345
34	Individual common variants exert weak effects on the risk for autism spectrum disorders. <i>Human Molecular Genetics</i> , 2012, 21, 4781-4792.	1.4	334
35	FOXP2 expression during brain development coincides with adult sites of pathology in a severe speech and language disorder. <i>Brain</i> , 2003, 126, 2455-2462.	3.7	313
36	A Genomewide Scan for Loci Involved in Attention-Deficit/Hyperactivity Disorder. <i>American Journal of Human Genetics</i> , 2002, 70, 1183-1196.	2.6	304

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37	LRRTM1 on chromosome 2p12 is a maternally suppressed gene that is associated paternally with handedness and schizophrenia. <i>Molecular Psychiatry</i> , 2007, 12, 1129-1139.	4.1	300
38	Familial Infantile Convulsions and Paroxysmal Choreoathetosis: A New Neurological Syndrome Linked to the Pericentromeric Region of Human Chromosome 16. <i>American Journal of Human Genetics</i> , 1997, 61, 889-898.	2.6	294
39	A missense mutation in connexin26, D66H, causes mutilating keratoderma with sensorineural deafness (Vohwinkel's syndrome) in three unrelated families. <i>Human Molecular Genetics</i> , 1999, 8, 1237-1243.	1.4	290
40	Isolation of the gene for McLeod syndrome that encodes a novel membrane transport protein. <i>Cell</i> , 1994, 77, 869-880.	13.5	272
41	Hailey-Hailey disease is caused by mutations in ATP2C1 encoding a novel Ca ²⁺ pump. <i>Human Molecular Genetics</i> , 2000, 9, 1131-1140.	1.4	264
42	A Quantitative-Trait Locus on Chromosome 6p Influences Different Aspects of Developmental Dyslexia. <i>American Journal of Human Genetics</i> , 1999, 64, 146-156.	2.6	260
43	Conservation of the Duchenne muscular dystrophy gene in mice and humans. <i>Science</i> , 1987, 238, 347-350.	6.0	248
44	McLeod neuroacanthocytosis: Genotype and phenotype. <i>Annals of Neurology</i> , 2001, 50, 755-764.	2.8	244
45	Independent genome-wide scans identify a chromosome 18 quantitative-trait locus influencing dyslexia. <i>Nature Genetics</i> , 2002, 30, 86-91.	9.4	240
46	The chromosome 6p22 haplotype associated with dyslexia reduces the expression of KIAA0319, a novel gene involved in neuronal migration. <i>Human Molecular Genetics</i> , 2006, 15, 1659-1666.	1.4	240
47	Isolation of a candidate gene for Norrie disease by positional cloning. <i>Nature Genetics</i> , 1992, 1, 199-203.	9.4	239
48	Expression of four alternative dystrophin transcripts in brain regions regulated by different promoters. <i>Human Molecular Genetics</i> , 1992, 1, 505-510.	1.4	236
49	A molecular defect in lorixin, the major component of the cornified cell envelope, underlies Vohwinkel's syndrome. <i>Nature Genetics</i> , 1996, 13, 70-77.	9.4	236
50	A 77-Kilobase Region of Chromosome 6p22.2 Is Associated with Dyslexia in Families From the United Kingdom and From the United States. <i>American Journal of Human Genetics</i> , 2004, 75, 1046-1058.	2.6	222
51	The UTX gene escapes X inactivation in mice and humans. <i>Human Molecular Genetics</i> , 1998, 7, 737-742.	1.4	218
52	The SPCH1 Region on Human 7q31: Genomic Characterization of the Critical Interval and Localization of Translocations Associated with Speech and Language Disorder. <i>American Journal of Human Genetics</i> , 2000, 67, 357-368.	2.6	214
53	Genetic Linkage of Attention-Deficit/Hyperactivity Disorder on Chromosome 16p13, in a Region Implicated in Autism. <i>American Journal of Human Genetics</i> , 2002, 71, 959-963.	2.6	210
54	A Genomewide Scan for Attention-Deficit/Hyperactivity Disorder in an Extended Sample: Suggestive Linkage on 17p11. <i>American Journal of Human Genetics</i> , 2003, 72, 1268-1279.	2.6	206

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55	Investigation of Dyslexia and SLI Risk Variants in Reading- and Language-Impaired Subjects. <i>Behavior Genetics</i> , 2011, 41, 90-104.	1.4	200
56	FOXP2 Is Not a Major Susceptibility Gene for Autism or Specific Language Impairment. <i>American Journal of Human Genetics</i> , 2002, 70, 1318-1327.	2.6	197
57	Mutations in the connexin 32 gene in X-linked dominant Charcot- Marie - Tooth disease (CMTX1). <i>Human Molecular Genetics</i> , 1994, 3, 29-34.	1.4	191
58	Analysis of the human VPS13 gene family. <i>Genomics</i> , 2004, 84, 536-549.	1.3	190
59	Chorein detection for the diagnosis of chorea-acanthocytosis. <i>Annals of Neurology</i> , 2004, 56, 299-302.	2.8	186
60	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. <i>Human Genetics</i> , 2012, 131, 565-579.	1.8	180
61	Disruption at the <i>PTCHD1</i> Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. <i>Science Translational Medicine</i> , 2010, 2, 49ra68.	5.8	178
62	Severity of disease and risk of malignant change in hereditary multiple exostoses. <i>Journal of Bone and Joint Surgery: British Volume</i> , 2004, 86-B, 1041-1046.	3.4	175
63	Clinical features and molecular bases of neuroacanthocytosis. <i>Journal of Molecular Medicine</i> , 2002, 80, 475-491.	1.7	174
64	CMIP and ATP2C2 Modulate Phonological Short-Term Memory in Language Impairment. <i>American Journal of Human Genetics</i> , 2009, 85, 264-272.	2.6	173
65	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. <i>European Journal of Human Genetics</i> , 2002, 10, 773-781.	1.4	172
66	A Whole-Genome Scan and Fine-Mapping Linkage Study of Auditory-Visual Synesthesia Reveals Evidence of Linkage to Chromosomes 2q24, 5q33, 6p12, and 12p12. <i>American Journal of Human Genetics</i> , 2009, 84, 279-285.	2.6	170
67	Highly Significant Linkage to the SLI1 Locus in an Expanded Sample of Individuals Affected by Specific Language Impairment. <i>American Journal of Human Genetics</i> , 2004, 74, 1225-1238.	2.6	169
68	Genetic Advances in the Study of Speech and Language Disorders. <i>Neuron</i> , 2010, 68, 309-320.	3.8	167
69	Candidate-Gene Screening and Association Analysis at the Autism-Susceptibility Locus on Chromosome 16p: Evidence of Association at GRIN2A and ABAT. <i>American Journal of Human Genetics</i> , 2005, 76, 950-966.	2.6	165
70	Construction, arraying, and high-density screening of large insert libraries of human chromosomes X and 21: their potential use as reference libraries.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1991, 88, 3233-3237.	3.3	163
71	Further characterization of the autism susceptibility locus AUTS1 on chromosome 7q. <i>Human Molecular Genetics</i> , 2001, 10, 973-982.	1.4	159
72	DCDC2, KIAA0319 and CMIP Are Associated with Reading-Related Traits. <i>Biological Psychiatry</i> , 2011, 70, 237-245.	0.7	156

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73	A physical map of 4 million bp around the Duchenne muscular dystrophy gene on the human X-chromosome. <i>Cell</i> , 1986, 47, 499-504.	13.5	155
74	YACs, BACs, PACs and MACs: Artificial chromosomes as research tools. <i>Trends in Biotechnology</i> , 1994, 12, 280-286.	4.9	151
75	Further evidence that the KIAA0319 gene confers susceptibility to developmental dyslexia. <i>Molecular Psychiatry</i> , 2006, 11, 1085-1091.	4.1	140
76	Serotonin transporter (5-HTT) and γ -aminobutyric acid receptor subunit $\beta 3$ (GABRB3) gene polymorphisms are not associated with autism in the IMGSA families. , 1999, 88, 492-496.		139
77	Localization and cloning of Xp21 deletion breakpoints involved in muscular dystrophy. <i>Human Genetics</i> , 1987, 75, 221-227.	1.8	136
78	Distal transcript of the dystrophin gene initiated from an alternative first exon and encoding a 75-kDa protein widely distributed in nonmuscle tissues.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1992, 89, 7506-7510.	3.3	136
79	DECIPHERING THE GENETIC BASIS OF SPEECH AND LANGUAGE DISORDERS. <i>Annual Review of Neuroscience</i> , 2003, 26, 57-80.	5.0	135
80	The Genetic Lexicon of Dyslexia. <i>Annual Review of Genomics and Human Genetics</i> , 2007, 8, 57-79.	2.5	131
81	Characterization of a Family with Rare Deletions in CNTNAP5 and DOCK4 Suggests Novel Risk Loci for Autism and Dyslexia. <i>Biological Psychiatry</i> , 2010, 68, 320-328.	0.7	131
82	Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. <i>Human Mutation</i> , 2010, 31, E1851-E1860.	1.1	130
83	A 15q13.3 microdeletion segregating with autism. <i>European Journal of Human Genetics</i> , 2009, 17, 687-692.	1.4	129
84	Common Variants in Left/Right Asymmetry Genes and Pathways Are Associated with Relative Hand Skill. <i>PLoS Genetics</i> , 2013, 9, e1003751.	1.5	129
85	Autism: recent molecular genetic advances. <i>Human Molecular Genetics</i> , 2000, 9, 861-868.	1.4	128
86	Mutation Screening of the EXT1 and EXT2 Genes in Patients with Hereditary Multiple Exostoses. <i>American Journal of Human Genetics</i> , 1997, 61, 520-528.	2.6	127
87	Chorea-Acanthocytosis: Genetic Linkage to Chromosome 9q21. <i>American Journal of Human Genetics</i> , 1997, 61, 899-908.	2.6	126
88	High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the IMMP2L-DOCK4 gene region in autism susceptibility. <i>Molecular Psychiatry</i> , 2010, 15, 954-968.	4.1	126
89	Linkage of Benign Familial Infantile Convulsions to Chromosome 16p12-q12 Suggests Allelism to the Infantile Convulsions and Choreoathetosis Syndrome. <i>American Journal of Human Genetics</i> , 2001, 68, 788-794.	2.6	125
90	A 10-megabase physical map of human Xp21, including the Duchenne muscular dystrophy gene. <i>Genomics</i> , 1988, 2, 189-202.	1.3	120

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91	Association of the KIAA0319 Dyslexia Susceptibility Gene With Reading Skills in the General Population. <i>American Journal of Psychiatry</i> , 2008, 165, 1576-1584.	4.0	120
92	Two Members of the Human MAGEB Gene Family Located in Xp21.3 Are Expressed in Tumors of Various Histological Origins. <i>Genomics</i> , 1997, 46, 397-408.	1.3	119
93	Analysis of reelin as a candidate gene for autism. <i>Molecular Psychiatry</i> , 2003, 8, 885-892.	4.1	119
94	Use of Multivariate Linkage Analysis for Dissection of a Complex Cognitive Trait. <i>American Journal of Human Genetics</i> , 2003, 72, 561-570.	2.6	119
95	PCSK6 is associated with handedness in individuals with dyslexia. <i>Human Molecular Genetics</i> , 2011, 20, 608-614.	1.4	119
96	Analysis of IMGSAC autism susceptibility loci: evidence for sex limited and parent of origin specific effects. <i>Journal of Medical Genetics</i> , 2005, 42, 132-137.	1.5	114
97	Human VPS13A is associated with multiple organelles and influences mitochondrial morphology and lipid droplet motility. <i>ELife</i> , 2019, 8, .	2.8	114
98	Genome-wide screening for DNA variants associated with reading and language traits. <i>Genes, Brain and Behavior</i> , 2014, 13, 686-701.	1.1	112
99	A Genomewide Linkage Screen for Relative Hand Skill in Sibling Pairs. <i>American Journal of Human Genetics</i> , 2002, 70, 800-805.	2.6	111
100	Screening of nine candidate genes for autism on chromosome 2q reveals rare nonsynonymous variants in the cAMP-GEFII gene. <i>Molecular Psychiatry</i> , 2003, 8, 916-924.	4.1	108
101	Genetic influences on language impairment and phonological short-term memory. <i>Trends in Cognitive Sciences</i> , 2005, 9, 528-534.	4.0	105
102	DNA linkage analysis of X chromosome-linked chronic granulomatous disease.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1986, 83, 3398-3401.	3.3	101
103	Autism and multiple exostoses associated with an X;8 translocation occurring within the GRPR gene and 3' to the SDC2 gene. <i>Human Molecular Genetics</i> , 1997, 6, 1241-1250.	1.4	100
104	Association of infantile convulsions with paroxysmal dyskinesias (ICCA syndrome): confirmation of linkage to human chromosome 16p12-q12 in a Chinese family. <i>Human Genetics</i> , 1998, 103, 608.	1.8	100
105	Complete coverage of the <i>Schizosaccharomyces pombe</i> genome in yeast artificial chromosomes. <i>Nature Genetics</i> , 1992, 1, 273-277.	9.4	99
106	Reconstruction of the 204 Mb human DMD-gene by homologous YAC recombination. <i>Human Molecular Genetics</i> , 1992, 1, 19-28.	1.4	95
107	Adenomatous polyps of the stomach. A clinical and pathological study of 153 cases. <i>Cancer</i> , 1962, 15, 456-467.	2.0	94
108	Isolation and characterization of a MAGE gene family in the Xp21.3 region.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1995, 92, 4987-4991.	3.3	94

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109	Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. <i>Journal of Medical Genetics</i> , 2011, 48, 48-54.	1.5	94
110	A giant locus for the Duchenne and Becker muscular dystrophy gene. <i>Trends in Genetics</i> , 1987, 3, 33-37.	2.9	92
111	A Common Variant Associated with Dyslexia Reduces Expression of the KIAA0319 Gene. <i>PLoS Genetics</i> , 2009, 5, e1000436.	1.5	92
112	The utrophin and dystrophin genes share similarities in genomic structure. <i>Human Molecular Genetics</i> , 1993, 2, 1765-1772.	1.4	91
113	Characterization of the exon structure of the Menkes disease gene using vectorette PCR. <i>Genomics</i> , 1995, 26, 437-442.	1.3	91
114	Putative functional alleles of DYX1C1 are not associated with dyslexia susceptibility in a large sample of sibling pairs from the UK. <i>Journal of Medical Genetics</i> , 2004, 41, 853-857.	1.5	91
115	Genetic and phenotypic effects of phonological short-term memory and grammatical morphology in specific language impairment. <i>Genes, Brain and Behavior</i> , 2008, 7, 393-402.	1.1	91
116	MET and autism susceptibility: family and case-control studies. <i>European Journal of Human Genetics</i> , 2009, 17, 749-758.	1.4	86
117	A Golgi localization signal identified in the Menkes recombinant protein. <i>Human Molecular Genetics</i> , 1998, 7, 1245-1252.	1.4	85
118	Comparison of fluorescent single-strand conformation polymorphism analysis and denaturing high-performance liquid chromatography for detection of EXT1 and EXT2 mutations in hereditary multiple exostoses. <i>European Journal of Human Genetics</i> , 2000, 8, 24-32.	1.4	85
119	Confirmatory Evidence for Linkage of Relative Hand Skill to 2p12-q11. <i>American Journal of Human Genetics</i> , 2003, 72, 499-501.	2.6	83
120	Bivariate linkage scan for reading disability and attention-deficit/hyperactivity disorder localizes pleiotropic loci. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2005, 46, 1045-1056.	3.1	83
121	Genome-wide association scan identifies new variants associated with a cognitive predictor of dyslexia. <i>Translational Psychiatry</i> , 2019, 9, 77.	2.4	82
122	Hailey-Hailey Disease: Molecular and Clinical Characterization of Novel Mutations in the ATP2C1 Gene. <i>Journal of Investigative Dermatology</i> , 2002, 118, 338-343.	0.3	81
123	Linkage and candidate gene studies of autism spectrum disorders in European populations. <i>European Journal of Human Genetics</i> , 2010, 18, 1013-1019.	1.4	80
124	Construction and Characterization of a 10-fold Genome Equivalent Rat P1-Derived Artificial Chromosome Library. <i>Genomics</i> , 1998, 50, 306-316.	1.3	78
125	Copy number variation and association analysis of SHANK3 as a candidate gene for autism in the IMGSAC collection. <i>European Journal of Human Genetics</i> , 2009, 17, 1347-1353.	1.4	76
126	Recent advances in the genetics of language impairment. <i>Genome Medicine</i> , 2010, 2, 6.	3.6	76

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127	Genome-wide association analyses of child genotype effects and parent-of-origin effects in specific language impairment. <i>Genes, Brain and Behavior</i> , 2014, 13, 418-429.	1.1	76
128	The Human 5'-Oligoadenylate Synthetase Locus Is Composed of Three Distinct Genes Clustered on Chromosome 12q24.2 Encoding the 100-, 69-, and 40-kDa Forms. <i>Genomics</i> , 1998, 52, 267-277.	1.3	75
129	Mutation screening and association analysis of six candidate genes for autism on chromosome 7q. <i>European Journal of Human Genetics</i> , 2005, 13, 198-207.	1.4	74
130	A yeast artificial chromosome contig containing the complete Duchenne muscular dystrophy gene. <i>Genomics</i> , 1992, 12, 465-473.	1.3	72
131	Absence of the XIST gene from late-replicating isodicentric X chromosomes in leukaemia. <i>Human Molecular Genetics</i> , 1994, 3, 1053-1059.	1.4	72
132	Genome-wide scan of reading ability in affected sibling pairs with attention-deficit/hyperactivity disorder: unique and shared genetic effects. <i>Molecular Psychiatry</i> , 2004, 9, 485-493.	4.1	72
133	Characterization of a yeast artificial chromosome contig spanning the Huntington's disease gene candidate region. <i>Nature Genetics</i> , 1992, 1, 180-187.	9.4	71
134	Early Developmental Regression in Autism Spectrum Disorder: Evidence from an International Multiplex Sample. <i>Journal of Autism and Developmental Disorders</i> , 2011, 41, 332-340.	1.7	71
135	A contig of non-chimaeric YACs containing the spinal muscular atrophy gene in 5q13. <i>Human Molecular Genetics</i> , 1993, 2, 1161-1167.	1.4	70
136	Characterization of the Menkes Protein Copper-Binding Domains and Their Role in Copper-Induced Protein Relocalization. <i>Human Molecular Genetics</i> , 1999, 8, 1473-1478.	1.4	67
137	Myotubularin-related 2 protein phosphatase and neurofilament light chain protein, both mutated in CMT neuropathies, interact in peripheral nerve. <i>Human Molecular Genetics</i> , 2003, 12, 1713-1723.	1.4	67
138	Associations of HLA alleles with specific language impairment. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 1.	1.5	67
139	Refined Linkage Disequilibrium and Physical Mapping of the Gene Locus for X-Linked Dystonia-Parkinsonism (DYT3). <i>Genomics</i> , 1999, 60, 320-329.	1.3	66
140	The genetic basis of dyslexia. <i>Lancet Neurology</i> , The, 2002, 1, 483-490.	4.9	65
141	Fine mapping of the chromosome 2p12-16 dyslexia susceptibility locus: quantitative association analysis and positional candidate genes SEMA4F and OTX1. <i>Psychiatric Genetics</i> , 2002, 12, 35-41.	0.6	64
142	Identifying Autism Susceptibility Genes. <i>Neuron</i> , 2000, 28, 19-24.	3.8	63
143	Familial Temporal Lobe Epilepsy as a Presenting Feature of Chorea-acanthocytosis. <i>Epilepsia</i> , 2005, 46, 1256-1263.	2.6	62
144	Parent-of-origin effects on handedness and schizophrenia susceptibility on chromosome 2p12-q11. <i>Human Molecular Genetics</i> , 2003, 12, 3225-3230.	1.4	61

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145	Early Clinical Heterogeneity in Choreoacanthocytosis. <i>Archives of Neurology</i> , 2005, 62, 611.	4.9	61
146	Three genes that escape X chromosome inactivation are clustered within a 6 Mb YAC contig and STS map in Xp11.21-p11.22. <i>Human Molecular Genetics</i> , 1995, 4, 731-739.	1.4	60
147	Oxidised LDL internalisation by the LOX-1 scavenger receptor is dependent on a novel cytoplasmic motif and is regulated by dynamin-2. <i>Journal of Cell Science</i> , 2008, 121, 2136-2147.	1.2	60
148	Further evidence for a parent-of-origin effect at the NOP9 locus on language-related phenotypes. <i>Journal of Neurodevelopmental Disorders</i> , 2016, 8, 24.	1.5	60
149	Genetic analysis of dyslexia candidate genes in the European cross-linguistic NeuroDys cohort. <i>European Journal of Human Genetics</i> , 2014, 22, 675-680.	1.4	59
150	Cloning the Wilson disease gene. <i>Nature Genetics</i> , 1993, 5, 317-318.	9.4	58
151	2.6 Mb YAC contig of the human X inactivation center region in Xq13: physical linkage of the RPS4X, PHKA1, XIST and DXS128E genes. <i>Human Molecular Genetics</i> , 1993, 2, 1105-1115.	1.4	57
152	The dyslexia-associated gene KIAA0319 encodes highly N- and O-glycosylated plasma membrane and secreted isoforms. <i>Human Molecular Genetics</i> , 2008, 17, 859-871.	1.4	56
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