Stanley F Nelson

List of Publications by Year
in descending order

298 papers	43,282 citations	1883 102 h-index	2375 198 g-index
316 all docs	316 docs citations	316 times ranked	57838 citing authors

#	Article	IF	Citations
1	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
2	Shotgun bisulphite sequencing of the Arabidopsis genome reveals DNA methylation patterning. Nature, 2008, 452, 215-219.	13.7	2,039
3	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. Nature, 2010, 468, 973-977.	13.7	1,944
4	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
5	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	6.0	1,654
6	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	9.4	1,272
7	Assessing the significance of chromosomal aberrations in cancer: Methodology and application to glioma. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 20007-20012.	3.3	927
8	High-throughput oncogene mutation profiling in human cancer. Nature Genetics, 2007, 39, 347-351.	9.4	927
9	Resolving Individuals Contributing Trace Amounts of DNA to Highly Complex Mixtures Using High-Density SNP Genotyping Microarrays. PLoS Genetics, 2008, 4, e1000167.	1.5	892
10	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	3.8	842
11	DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. Genetics in Medicine, 2011, 13, 913-920.	1.1	809
12	Linkage, Association, and Gene-Expression Analyses Identify CNTNAP2 as an Autism-Susceptibility Gene. American Journal of Human Genetics, 2008, 82, 150-159.	2.6	738
13	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	7.1	701
14	Gene Expression Profiling of Gliomas Strongly Predicts Survival. Cancer Research, 2004, 64, 6503-6510.	0.4	659
15	Analysis of oncogenic signaling networks in glioblastoma identifies ASPM as a molecular target. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 17402-17407.	3.3	606
16	Melanoma whole-exome sequencing identifies V600EB-RAF amplification-mediated acquired B-RAF inhibitor resistance. Nature Communications, 2012, 3, 724.	5.8	567
17	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
18	Genome Sequencing Highlights the Dynamic Early History of Dogs. PLoS Genetics, 2014, 10, e1004016.	1.5	481

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19	DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. Genetics in Medicine, 2012, 14, 296-305.	1.1	471
20	Targeted Therapy Resistance Mediated by Dynamic Regulation of Extrachromosomal Mutant EGFR DNA. Science, 2014, 343, 72-76.	6.0	460
21	BFAST: An Alignment Tool for Large Scale Genome Resequencing. PLoS ONE, 2009, 4, e7767.	1.1	444
22	Cold Urticaria, Immunodeficiency, and Autoimmunity Related toPLCG2Deletions. New England Journal of Medicine, 2012, 366, 330-338.	13.9	391
23	Gene Expression Profile Correlates with T-Cell Infiltration and Relative Survival in Glioblastoma Patients Vaccinated with Dendritic Cell Immunotherapy. Clinical Cancer Research, 2011, 17, 1603-1615.	3.2	378
24	C-terminal truncations in human 3â€²-5â€² DNA exonuclease TREX1 cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy. Nature Genetics, 2007, 39, 1068-1070.	9.4	366
25	Mutations in a Human ROBO Gene Disrupt Hindbrain Axon Pathway Crossing and Morphogenesis. Science, 2004, 304, 1509-1513.	6.0	361
26	Exome sequencing-based copy-number variation and loss of heterozygosity detection: ExomeCNV. Bioinformatics, 2011, 27, 2648-2654.	1.8	357
27	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	1.4	334
28	Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. Nature Genetics, 2012, 44, 78-84.	9.4	334
29	Gene connectivity, function, and sequence conservation: predictions from modular yeast co-expression networks. BMC Genomics, 2006, 7, 40.	1.2	327
30	Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. Nature Genetics, 2008, 40, 999-1003.	9.4	320
31	A Single CRISPR-Cas9 Deletion Strategy that Targets the Majority of DMD Patients Restores Dystrophin Function in hiPSC-Derived Muscle Cells. Cell Stem Cell, 2016, 18, 533-540.	5.2	307
32	A Genomewide Scan for Loci Involved in Attention-Deficit/Hyperactivity Disorder. American Journal of Human Genetics, 2002, 70, 1183-1196.	2.6	304
33	Identification of inflammatory gene modules based on variations of human endothelial cell responses to oxidized lipids. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 12741-12746.	3.3	303
34	Epidermal Growth Factor Receptor Activation in Glioblastoma through Novel Missense Mutations in the Extracellular Domain. PLoS Medicine, 2006, 3, e485.	3.9	298
35	Gene expression profiling identifies molecular subtypes of gliomas. Oncogene, 2003, 22, 4918-4923.	2.6	264
36	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	13.9	261

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37	Whole genome amplification using a degenerate oligonucleotide primer allows hundreds of genotypes to be performed on less than one nanogram of genomic DNA. Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 14676-14679.	3.3	256
38	Oxysterols Are Novel Activators of the Hedgehog Signaling Pathway in Pluripotent Mesenchymal Cells. Journal of Biological Chemistry, 2007, 282, 8959-8968.	1.6	254
39	Identification of molecular subtypes of glioblastoma by gene expression profiling. Oncogene, 2003, 22, 2361-2373.	2.6	247
40	A Genetic Analysis of Neural Progenitor Differentiation. Neuron, 2001, 29, 325-339.	3.8	243
41	Genome-Wide Analysis of Copy Number Variants in Attention Deficit Hyperactivity Disorder: The Role of Rare Variants and Duplications at 15q13.3. American Journal of Psychiatry, 2012, 169, 195-204.	4.0	242
42	U87MG Decoded: The Genomic Sequence of a Cytogenetically Aberrant Human Cancer Cell Line. PLoS Genetics, 2010, 6, e1000832.	1.5	229
43	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature Genetics, 2012, 44, 704-708.	9.4	216
44	Primary Glioblastomas Express Mesenchymal Stem-Like Properties. Molecular Cancer Research, 2006, 4, 607-619.	1.5	215
45	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	9.4	212
46	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
47	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	4.5	211
48	Genetic Linkage of Attention-Deficit/Hyperactivity Disorder on Chromosome 16p13, in a Region Implicated in Autism. American Journal of Human Genetics, 2002, 71, 959-963.	2.6	210
49	A Genomewide Scan for Attention-Deficit/Hyperactivity Disorder in an Extended Sample: Suggestive Linkage on 17p11. American Journal of Human Genetics, 2003, 72, 1268-1279.	2.6	206
50	Evidence that the dopamine D4 receptor is a susceptibility gene in attention deficit hyperactivity disorder. Molecular Psychiatry, 1998, 3, 427-430.	4.1	198
51	SGK196 Is a Glycosylation-Specific O -Mannose Kinase Required for Dystroglycan Function. Science, 2013, 341, 896-899.	6.0	197
52	Progressive Ataxia Due to a Missense Mutation in a Calcium-Channel Gene. American Journal of Human Genetics, 1997, 61, 1078-1087.	2.6	191
53	Joint mouseâ€“human phenome-wide association to test gene function and disease risk. Nature Communications, 2016, 7, 10464.	5.8	190
54	DNA-microarray analysis of brain cancer: molecular classification for therapy. Nature Reviews Neuroscience, 2004, 5, 782-792.	4.9	189

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55	Identification of EpCAM as the Gene for Congenital Tufting Enteropathy. Gastroenterology, 2008, 135, 429-437.	0.6	185
56	Distinct Transcription Profiles of Primary and Secondary Glioblastoma Subgroups. Cancer Research, 2006, 66, 159-167.	0.4	182
57	Gene expression analysis of glioblastomas identifies the major molecular basis for the prognostic benefit of younger age. BMC Medical Genomics, 2008, 1, 52.	0.7	181
58	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	1.8	180
59	Insulin growth factor-binding protein 2 is a candidate biomarker for PTEN status and PI3K/Akt pathway activation in glioblastoma and prostate cancer. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 5563-5568.	3.3	173
60	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E333-E344.	1.8	172
61	Detection of differentially expressed genes in primary tumor tissues using representational differences analysis coupled to microarray hybridization. Nucleic Acids Research, 1998, 26, 3059-3065.	6.5	169
62	Evidence for linkage of a tandem duplication polymorphism upstream of the dopamine D4 receptor gene (DRD4) with attention deficit hyperactivity disorder (ADHD). Molecular Psychiatry, 2000, 5, 531-536.	4.1	169
63	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. Nature Genetics, 2012, 44, 788-792.	9.4	169
64	Joint Analysis of the DRD5 Marker Concludes Association with Attention-Deficit/Hyperactivity Disorder Confined to the Predominantly Inattentive and Combined Subtypes. American Journal of Human Genetics, 2004, 74, 348-356.	2.6	168
65	Evidence for Sex-Specific Risk Alleles in Autism Spectrum Disorder. American Journal of Human Genetics, 2004, 75, 1117-1123.	2.6	165
66	Metaâ€analysis of genomeâ€wide linkage scans of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1392-1398.	1.1	160
67	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
68	FXR regulates organic solute transporters Î± and Î² in the adrenal gland, kidney, and intestine. Journal of Lipid Research, 2006, 47, 201-214.	2.0	153
69	ERBB3 and NGFR mark a distinct skeletal muscle progenitor cell in human development and hPSCs. Nature Cell Biology, 2018, 20, 46-57.	4.6	151
70	Ciliary Abnormalities Due to Defects in the Retrograde Transport Protein DYNC2H1 in Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2009, 84, 542-549.	2.6	149
71	Genome-wide analysis of single-nucleotide polymorphisms in human expressed sequences. Nature Genetics, 2000, 26, 233-236.	9.4	147
72	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. Nature Genetics, 2011, 43, 365-369.	9.4	147

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73	High-throughput profiling of influenza A virus hemagglutinin gene at single-nucleotide resolution. Scientific Reports, 2014, 4, 4942.	1.6	147
74	Relationship between Gene Expression and Enhancement in Glioblastoma Multiforme: Exploratory DNA Microarray Analysis. Radiology, 2008, 249, 268-277.	3.6	146
75	Maternal embryonic leucine zipper kinase is a key regulator of the proliferation of malignant brain tumors, including brain tumor stem cells. Journal of Neuroscience Research, 2008, 86, 48-60.	1.3	144
76	Familial episodic ataxia: Clinical heterogeneity in four families linked to chromosome 19p. Annals of Neurology, 1997, 41, 8-16.	2.8	143
77	Replication of Autism Linkage: Fine-Mapping Peak at 17q21. American Journal of Human Genetics, 2005, 76, 1050-1056.	2.6	142
78	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
79	Prevalence and Psychiatric Comorbidity of Attention-Deficit/Hyperactivity Disorder in an Adolescent Finnish Population. Journal of the American Academy of Child and Adolescent Psychiatry, 2007, 46, 1575-1583.	0.3	141
80	The apolipoprotein E ?4 allele is not a significant risk factor for frontotemporal dementia. Annals of Neurology, 1998, 44, 134-138.	2.8	136
81	Maternal embryonic leucine zipper kinase (MELK) regulates multipotent neural progenitor proliferation. Journal of Cell Biology, 2005, 170, 413-427.	2.3	136
82	Fyn and Src Are Effectors of Oncogenic Epidermal Growth Factor Receptor Signaling in Glioblastoma Patients. Cancer Research, 2009, 69, 6889-6898.	0.4	136
83	Biomarkers to Predict Antidepressant Response. Current Psychiatry Reports, 2010, 12, 553-562.	2.1	136
84	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	1.1	136
85	Genomic mismatch scanning: a new approach to genetic linkage mapping. Nature Genetics, 1993, 4, 11-18.	9.4	134
86	EGFR Mutation-Induced Alternative Splicing of Max Contributes to Growth of Glycolytic Tumors in Brain Cancer. Cell Metabolism, 2013, 17, 1000-1008.	7.2	130
87	Exome Sequencing Identifies PDE4D Mutations in Acrodysostosis. American Journal of Human Genetics, 2012, 90, 746-751.	2.6	128
88	Genomic Landscape of Meningiomas. Brain Pathology, 2010, 20, 751-762.	2.1	124
89	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	13.9	122
90	Family-Based Genome-Wide Association Scan of Attention-Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 898-905.e3.	0.3	122

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91	Attention Deficit Hyperactivity Disorder: Fine Mapping Supports Linkage to 5p13, 6q12, 16p13, and 17p11. American Journal of Human Genetics, 2004, 75, 661-668.	2.6	121
92	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. Genetics in Medicine, 2014, 16, 510-515.	1.1	121
93	A Recessive Skeletal Dysplasia, SEMD Aggrecan Type, Results from a Missense Mutation Affecting the C-Type Lectin Domain of Aggrecan. American Journal of Human Genetics, 2009, 84, 72-79.	2.6	120
94	Accuracy of phenotyping of autistic children based on internet implemented parent report. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1119-1126.	1.1	120
95	Annexin A6 modifies muscular dystrophy by mediating sarcolemmal repair. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 6004-6009.	3.3	117
96	Disruption of POF1B Binding to Nonmuscle Actin Filaments Is Associated with Premature Ovarian Failure. American Journal of Human Genetics, 2006, 79, 113-119.	2.6	116
97	De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay. American Journal of Human Genetics, 2015, 96, 498-506.	2.6	115
98	Differential Induction of Glioblastoma Migration and Growth by Two Forms of Pleiotrophin. Journal of Biological Chemistry, 2005, 280, 26953-26964.	1.6	112
99	Molecular properties of CD133+ glioblastoma stem cells derived from treatment-refractory recurrent brain tumors. Journal of Neuro-Oncology, 2009, 94, 1-19.	1.4	111
100	Relationship between Survival and Edema in Malignant Gliomas: Role of Vascular Endothelial Growth Factor and Neuronal Pentraxin 2. Clinical Cancer Research, 2007, 13, 2592-2598.	3.2	108
101	Biased paternal transmission of SNAP-25 risk alleles in attention-deficit hyperactivity disorder. Molecular Psychiatry, 2003, 8, 309-315.	4.1	107
102	Transmission disequilibrium testing of dopamine-related candidate gene polymorphisms in ADHD: confirmation of association of ADHD with DRD4 and DRD5. Molecular Psychiatry, 2004, 9, 711-717.	4.1	105
103	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	1.4	101
104	Association of the cannabinoid receptor gene (CNR1) with ADHD and postâ€traumatic stress disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1488-1494.	1.1	99
105	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	2.8	98
106	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	3.8	95
107	Temperament and Character Profiles and the Dopamine D4 Receptor Gene in ADHD. American Journal of Psychiatry, 2005, 162, 906-913.	4.0	94
108	PDE5 inhibition alleviates functional muscle ischemia in boys with Duchenne muscular dystrophy. Neurology, 2014, 82, 2085-2091.	1.5	94

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109	Exome sequencing identifies de novo gain of function missense mutation in KCND2 in identical twins with autism and seizures that slows potassium channel inactivation. Human Molecular Genetics, 2014, 23, 3481-3489.	1.4	90
110	Detecting tissue-specific regulation of alternative splicing as a qualitative change in microarray data. Nucleic Acids Research, 2004, 32, e180-e180.	6.5	87
111	Stem cell associated gene expression in glioblastoma multiforme: relationship to survival and the subventricular zone. Journal of Neuro-Oncology, 2010, 96, 359-367.	1.4	86
112	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. Genome Medicine, 2017, 9, 90.	3.6	86
113	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.5	84
114	Linkage-disequilibrium mapping without genotyping. Nature Genetics, 1998, 18, 225-230.	9.4	82
115	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. American Journal of Human Genetics, 2010, 87, 768-778.	2.6	82
116	Collaborative analysis of DRD4 and DAT genotypes in population-defined ADHD subtypes. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2005, 46, 1067-1073.	3.1	79
117	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	5.8	79
118	Familial migraine with vertigo: No mutations found in CACNA1A. , 1998, 79, 148-151.		78
119	SeqWare Query Engine: storing and searching sequence data in the cloud. BMC Bioinformatics, 2010, 11, S2.	1.2	78
120	Klinefelter's syndrome as a model of anomalous cerebral laterality: Testing gene dosage in the X chromosome pseudoautosomal region using a DNA microarray. , 1998, 23, 215-229.		77
121	Dantrolene Enhances Antisense-Mediated Exon Skipping in Human and Mouse Models of Duchenne Muscular Dystrophy. Science Translational Medicine, 2012, 4, 164ra160.	5.8	77
122	Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. PLoS Genetics, 2016, 12, e1005851.	1.5	77
123	Sequence variant in the laminin Î³1 (LAMC1) gene associated with familial pelvic organ prolapse. Human Genetics, 2007, 120, 847-856.	1.8	76
124	Proteinase and Growth Factor Alterations Revealed by Gene Microarray Analysis of Human Diabetic Corneas. , 2005, 46, 3604.		75
125	Identification of allele-specific alternative mRNA processing via transcriptome sequencing. Nucleic Acids Research, 2012, 40, e104-e104.	6.5	74
126	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	3.7	74

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127	De novo mutation in CACNA1A caused acetazolamide-responsive episodic ataxia. , 1998, 77, 298-301.		73
128	Genome-wide scan of reading ability in affected sibling pairs with attention-deficit/hyperactivity disorder: unique and shared genetic effects. Molecular Psychiatry, 2004, 9, 485-493.	4.1	72
129	Targeted massively parallel sequencing provides comprehensive genetic diagnosis for patients with disorders of sex development. Clinical Genetics, 2013, 83, 35-43.	1.0	72
130	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
131	Loss of annexin A1 expression in human breast cancer detected by multiple high-throughput analyses. Biochemical and Biophysical Research Communications, 2004, 326, 218-227.	1.0	68
132	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	9.4	68
133	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	1.1	68
134	High-density SNP association study of the 17q21 chromosomal region linked to autism identifies CACNA1G as a novel candidate gene. Molecular Psychiatry, 2010, 15, 996-1005.	4.1	67
135	Association of Sex With Recurrence of Autism Spectrum Disorder Among Siblings. JAMA Pediatrics, 2017, 171, 1107.	3.3	66
136	A New Episodic Ataxia Syndrome With Linkage to Chromosome 19q13. Archives of Neurology, 2007, 64, 749.	4.9	65
137	Expanding the phenotype of mutations in DICER1: mosaic missense mutations in the RNase IIIb domain ofDICER1cause GLOW syndrome. Journal of Medical Genetics, 2014, 51, 294-302.	1.5	65
138	DMD genotype correlations from the Duchenne Registry: Endogenous exon skipping is a factor in prolonged ambulation for individuals with a defined mutation subtype. Human Mutation, 2018, 39, 1193-1202.	1.1	65
139	CBCL Pediatric Bipolar Disorder Profile and ADHD: Comorbidity and Quantitative Trait Loci Analysis. Journal of the American Academy of Child and Adolescent Psychiatry, 2008, 47, 1151-1157.	0.3	64
140	Differential Gene Expression in Glioblastoma Defined by ADC Histogram Analysis: Relationship to Extracellular Matrix Molecules and Survival. American Journal of Neuroradiology, 2012, 33, 1059-1064.	1.2	64
141	Pleiotropic Mechanisms Indicated for Sex Differences in Autism. PLoS Genetics, 2016, 12, e1006425.	1.5	64
142	Emerging genetic therapies to treat Duchenne muscular dystrophy. Current Opinion in Neurology, 2009, 22, 532-538.	1.8	63
143	Homozygous mutation inNUP107leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. Journal of Medical Genetics, 2017, 54, 399-403.	1.5	62
144	Improved variant discovery through local re-alignment of short-read next-generation sequencing data using SRMA. Genome Biology, 2010, 11, R99.	3.8	61

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145	Detection of a MicroRNA Signal in an In Vivo Expression Set of mRNAs. PLoS ONE, 2007, 2, e804.	1.1	61
146	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
147	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	2.6	59
148	Expanding the mutational spectrum of LZTR1 in schwannomatosis. European Journal of Human Genetics, 2015, 23, 963-968.	1.4	58
149	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
150	Molecular Analysis of Glioblastoma: Pathway Profiling and Its Implications for Patient Therapy. Cancer Biology and Therapy, 2003, 2, 242-247.	1.5	57
151	High density SNP association study of a major autism linkage region on chromosome 17. Human Molecular Genetics, 2007, 16, 704-715.	1.4	57
152	Celsius: a community resource for Affymetrix microarray data. Genome Biology, 2007, 8, R112.	13.9	57
153	A genome-wide linkage scan of familial benign recurrent vertigo: linkage to 22q12 with evidence of heterogeneity. Human Molecular Genetics, 2006, 15, 251-258.	1.4	56
154	Identification of Retinol Binding Protein 1 Promoter Hypermethylation in Isocitrate Dehydrogenase 1 and 2 Mutant Gliomas. Journal of the National Cancer Institute, 2012, 104, 1458-1469.	3.0	56
155	Characterization of transformation related genes in oral cancer cells. Oncogene, 1998, 16, 1921-1930.	2.6	55
156	Pooled genome-wide linkage data on 424 ADHD ASPs suggests genetic heterogeneity and a common risk locus at 5p13. Molecular Psychiatry, 2006, 11, 5-8.	4.1	55
157	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. Frontiers in Psychiatry, 2013, 4, 154.	1.3	54
158	A comparison of gene expression profiles produced by SAGE, long SAGE, and oligonucleotide chips. Genomics, 2004, 84, 631-636.	1.3	53
159	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	2.8	53
160	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€“binding region. Epilepsia, 2019, 60, 406-418.	2.6	53
161	Ribosomal Proteins RPS11 and RPS20, Two Stress-Response Markers of Glioblastoma Stem Cells, Are Novel Predictors of Poor Prognosis in Glioblastoma Patients. PLoS ONE, 2015, 10, e0141334.	1.1	52
162	ADHD Candidate Gene Study in a Population-Based Birth Cohort: Association with DBH and DRD2. Journal of the American Academy of Child and Adolescent Psychiatry, 2007, 46, 1614-1621.	0.3	50

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163	De Novo variants in the KMT2A (MLL) gene causing atypical Wiedemann-Steiner syndrome in two unrelated individuals identified by clinical exome sequencing. BMC Medical Genetics, 2014, 15, 49.	2.1	49
164	Familial Clustering and DRD4 Effects on Electroencephalogram Measures in Multiplex Families With Attention Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 368-377.	0.3	48
165	Protective Properties of Radio-Chemoresistant Glioblastoma Stem Cell Clones Are Associated with Metabolic Adaptation to Reduced Glucose Dependence. PLoS ONE, 2013, 8, e80397.	1.1	48
166	A novel mutation in KCNA1 causes episodic ataxia without myokymia. Human Mutation, 2004, 24, 536-536.	1.1	47
167	Repetitive sequence environment distinguishes housekeeping genes. Gene, 2007, 390, 153-165.	1.0	47
168	Phenotypic and Genetic Analysis of a Large Family With Migraineâ€Associated Vertigo. Headache, 2008, 48, 1460-1467.	1.8	46
169	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
170	Distinct gene expression profiles in adult mouse heart following targeted MAP kinase activation. Physiological Genomics, 2006, 25, 50-59.	1.0	41
171	Rich annotation of DNA sequencing variants by leveraging the Ensembl Variant Effect Predictor with plugins. Briefings in Bioinformatics, 2015, 16, 255-264.	3.2	41
172	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. Npj Genomic Medicine, 2016, 1, .	1.7	41
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