

# Daniel H Geschwind

## List of Publications by Year in descending order

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Version: 2024-02-01

242  
papers

62,972  
citations

1294

109  
h-index

1216

227  
g-index

289  
all docs

289  
docs citations

289  
times ranked

59694  
citing authors

#	ARTICLE	IF	CITATIONS
1	An anatomically comprehensive atlas of the adult human brain transcriptome. <i>Nature</i> , 2012, 489, 391-399.	13.7	2,321
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	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A $\beta$ , tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019, 51, 414-430.	9.4	1,962
4	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. <i>Nature</i> , 2012, 485, 237-241.	13.7	1,863
5	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. <i>Nature Genetics</i> , 2011, 43, 436-441.	9.4	1,676
6	Transcriptomic analysis of autistic brain reveals convergent molecular pathology. <i>Nature</i> , 2011, 474, 380-384.	13.7	1,654
7	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. <i>Nature Genetics</i> , 2019, 51, 63-75.	9.4	1,594
8	Identification of common genetic risk variants for autism spectrum disorder. <i>Nature Genetics</i> , 2019, 51, 431-444.	9.4	1,538
9	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
10	Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. <i>Nature Genetics</i> , 2018, 50, 381-389.	9.4	1,332
11	Functional cortical neurons and astrocytes from human pluripotent stem cells in 3D culture. <i>Nature Methods</i> , 2015, 12, 671-678.	9.0	1,220
12	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
13	Transcriptional landscape of the prenatal human brain. <i>Nature</i> , 2014, 508, 199-206.	13.7	1,147
14	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. <i>Neuron</i> , 2011, 70, 863-885.	3.8	1,146
15	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
16	The Central Nervous System and the Gut Microbiome. <i>Cell</i> , 2016, 167, 915-932.	13.5	985
17	Integrative Functional Genomic Analyses Implicate Specific Molecular Pathways and Circuits in Autism. <i>Cell</i> , 2013, 155, 1008-1021.	13.5	948
18	Sex differences in autism spectrum disorders. <i>Current Opinion in Neurology</i> , 2013, 26, 146-153.	1.8	895

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19	Absence of CNTNAP2 Leads to Epilepsy, Neuronal Migration Abnormalities, and Core Autism-Related Deficits. <i>Cell</i> , 2011, 147, 235-246.	13.5	870
20	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. <i>Science</i> , 2018, 359, 693-697.	6.0	851
21	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
22	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. <i>Science</i> , 2018, 362, .	6.0	805
23	Functional organization of the transcriptome in human brain. <i>Nature Neuroscience</i> , 2008, 11, 1271-1282.	7.1	743
24	Human Gut Microbiota from Autism Spectrum Disorder Promote Behavioral Symptoms in Mice. <i>Cell</i> , 2019, 177, 1600-1618.e17.	13.5	701
25	Advancing the understanding of autism disease mechanisms through genetics. <i>Nature Medicine</i> , 2016, 22, 345-361.	15.2	684
26	Comprehensive functional genomic resource and integrative model for the human brain. <i>Science</i> , 2018, 362, .	6.0	618
27	Genome-wide changes in lncRNA, splicing, and regional gene expression patterns in autism. <i>Nature</i> , 2016, 540, 423-427.	13.7	603
28	Conservation and evolution of gene coexpression networks in human and chimpanzee brains. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 17973-17978.	3.3	580
29	Evidence for $\alpha$ -synuclein prions causing multiple system atrophy in humans with parkinsonism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E5308-17.	3.3	578
30	Selenium Drives a Transcriptional Adaptive Program to Block Ferroptosis and Treat Stroke. <i>Cell</i> , 2019, 177, 1262-1279.e25.	13.5	576
31	Functional and Evolutionary Insights into Human Brain Development through Global Transcriptome Analysis. <i>Neuron</i> , 2009, 62, 494-509.	3.8	555
32	Genetics of autism spectrum disorders. <i>Trends in Cognitive Sciences</i> , 2011, 15, 409-416.	4.0	546
33	A Highly Conserved Program of Neuronal Microexons Is Misregulated in Autistic Brains. <i>Cell</i> , 2014, 159, 1511-1523.	13.5	546
34	A genome-wide scan for common alleles affecting risk for autism. <i>Human Molecular Genetics</i> , 2010, 19, 4072-4082.	1.4	538
35	Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. <i>Nature Medicine</i> , 2011, 17, 1657-1662.	15.2	521
36	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. <i>Science</i> , 2018, 362, .	6.0	516

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37	Chromosome conformation elucidates regulatory relationships in developing human brain. <i>Nature</i> , 2016, 538, 523-527.	13.7	507
38	Divergence of human and mouse brain transcriptome highlights Alzheimer disease pathways. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 12698-12703.	3.3	487
39	Cortical Evolution: Judge the Brain by Its Cover. <i>Neuron</i> , 2013, 80, 633-647.	3.8	444
40	Advances in Autism. <i>Annual Review of Medicine</i> , 2009, 60, 367-380.	5.0	411
41	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. <i>Nature Genetics</i> , 2018, 50, 538-548.	9.4	406
42	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. <i>Nature Neuroscience</i> , 2016, 19, 571-577.	7.1	388
43	A Multi-network Approach Identifies Protein-Specific Co-expression in Asymptomatic and Symptomatic Alzheimer's Disease. <i>Cell Systems</i> , 2017, 4, 60-72.e4.	2.9	381
44	A Systems Level Analysis of Transcriptional Changes in Alzheimer's Disease and Normal Aging. <i>Journal of Neuroscience</i> , 2008, 28, 1410-1420.	1.7	379
45	Systems biology and gene networks in neurodevelopmental and neurodegenerative disorders. <i>Nature Reviews Genetics</i> , 2015, 16, 441-458.	7.7	378
46	Gene hunting in autism spectrum disorder: on the path to precision medicine. <i>Lancet Neurology</i> , The, 2015, 14, 1109-1120.	4.9	374
47	The PsychENCODE project. <i>Nature Neuroscience</i> , 2015, 18, 1707-1712.	7.1	371
48	A Single-Cell Transcriptomic Atlas of Human Neocortical Development during Mid-gestation. <i>Neuron</i> , 2019, 103, 785-801.e8.	3.8	361
49	Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9.	2.6	357
50	Altered proliferation and networks in neural cells derived from idiopathic autistic individuals. <i>Molecular Psychiatry</i> , 2017, 22, 820-835.	4.1	349
51	Genetics and genomics of psychiatric disease. <i>Science</i> , 2015, 349, 1489-1494.	6.0	337
52	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
53	Defining the Genetic, Genomic, Cellular, and Diagnostic Architectures of Psychiatric Disorders. <i>Cell</i> , 2019, 177, 162-183.	13.5	331
54	Reliability of human cortical organoid generation. <i>Nature Methods</i> , 2019, 16, 75-78.	9.0	330

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55	Inherited and De Novo Genetic Risk for Autism Impacts Shared Networks. <i>Cell</i> , 2019, 178, 850-866.e26.	13.5	326
56	A Systems-Level Analysis of the Peripheral Nerve Intrinsic Axonal Growth Program. <i>Neuron</i> , 2016, 89, 956-970.	3.8	314
57	Exogenous and evoked oxytocin restores social behavior in the <i>Cntnap2</i> mouse model of autism. <i>Science Translational Medicine</i> , 2015, 7, 271ra8.	5.8	308
58	Self-Organized Cerebral Organoids with Human-Specific Features Predict Effective Drugs to Combat Zika Virus Infection. <i>Cell Reports</i> , 2017, 21, 517-532.	2.9	305
59	Autism: Many Genes, Common Pathways?. <i>Cell</i> , 2008, 135, 391-395.	13.5	293
60	Genome-wide expression profiling of lymphoblastoid cell lines distinguishes different forms of autism and reveals shared pathways. <i>Human Molecular Genetics</i> , 2007, 16, 1682-1698.	1.4	290
61	Strategies for aggregating gene expression data: The collapseRows R function. <i>BMC Bioinformatics</i> , 2011, 12, 322.	1.2	290
62	Astrocyte layers in the mammalian cerebral cortex revealed by a single-cell in situ transcriptomic map. <i>Nature Neuroscience</i> , 2020, 23, 500-509.	7.1	290
63	The Autism Genetic Resource Exchange: A Resource for the Study of Autism and Related Neuropsychiatric Conditions. <i>American Journal of Human Genetics</i> , 2001, 69, 463-466.	2.6	284
64	The Dynamic Landscape of Open Chromatin during Human Cortical Neurogenesis. <i>Cell</i> , 2018, 172, 289-304.e18.	13.5	281
65	Circuit-wide Transcriptional Profiling Reveals Brain Region-Specific Gene Networks Regulating Depression Susceptibility. <i>Neuron</i> , 2016, 90, 969-983.	3.8	272
66	The Human Brain in a Dish: The Promise of iPSC-Derived Neurons. <i>Cell</i> , 2011, 145, 831-834.	13.5	270
67	Genes and pathways underlying regional and cell type changes in Alzheimer's disease. <i>Genome Medicine</i> , 2013, 5, 48.	3.6	267
68	Identification of the Transcriptional Targets of FOXP2, a Gene Linked to Speech and Language, in Developing Human Brain. <i>American Journal of Human Genetics</i> , 2007, 81, 1144-1157.	2.6	262
69	Foxp2 Regulates Gene Networks Implicated in Neurite Outgrowth in the Developing Brain. <i>PLoS Genetics</i> , 2011, 7, e1002145.	1.5	256
70	Transcriptional Reprogramming of Distinct Peripheral Sensory Neuron Subtypes after Axonal Injury. <i>Neuron</i> , 2020, 108, 128-144.e9.	3.8	254
71	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. <i>Nature Genetics</i> , 2015, 47, 579-581.	9.4	237
72	Histone Acetylome-wide Association Study of Autism Spectrum Disorder. <i>Cell</i> , 2016, 167, 1385-1397.e11.	13.5	237

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73	The BRAIN Initiative Cell Census Consortium: Lessons Learned toward Generating a Comprehensive Brain Cell Atlas. <i>Neuron</i> , 2017, 96, 542-557.	3.8	235
74	De novo mutations in regulatory elements in neurodevelopmental disorders. <i>Nature</i> , 2018, 555, 611-616.	13.7	232
75	Gene expression in human brain implicates sexually dimorphic pathways in autism spectrum disorders. <i>Nature Communications</i> , 2016, 7, 10717.	5.8	227
76	Clinicopathological correlations in behavioural variant frontotemporal dementia. <i>Brain</i> , 2017, 140, 3329-3345.	3.7	226
77	The Emerging Picture of Autism Spectrum Disorder: Genetics and Pathology. <i>Annual Review of Pathology: Mechanisms of Disease</i> , 2015, 10, 111-144.	9.6	225
78	Transcriptome and epigenome landscape of human cortical development modeled in organoids. <i>Science</i> , 2018, 362, .	6.0	220
79	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. <i>JAMA Neurology</i> , 2014, 71, 1237.	4.5	211
80	Cytoplasmic Rbfox1 Regulates the Expression of Synaptic and Autism-Related Genes. <i>Neuron</i> , 2016, 89, 113-128.	3.8	205
81	Neuroscience in the era of functional genomics and systems biology. <i>Nature</i> , 2009, 461, 908-915.	13.7	201
82	The contribution of rare variants to risk of schizophrenia in individuals with and without intellectual disability. <i>Nature Genetics</i> , 2017, 49, 1167-1173.	9.4	200
83	Microglia-organized scar-free spinal cord repair in neonatal mice. <i>Nature</i> , 2020, 587, 613-618.	13.7	197
84	RBFOX1 regulates both splicing and transcriptional networks in human neuronal development. <i>Human Molecular Genetics</i> , 2012, 21, 4171-4186.	1.4	192
85	The road to precision psychiatry: translating genetics into disease mechanisms. <i>Nature Neuroscience</i> , 2016, 19, 1397-1407.	7.1	189
86	Long-term maturation of human cortical organoids matches key early postnatal transitions. <i>Nature Neuroscience</i> , 2021, 24, 331-342.	7.1	188
87	A Quantitative Framework to Evaluate Modeling of Cortical Development by Neural Stem Cells. <i>Neuron</i> , 2014, 83, 69-86.	3.8	184
88	Rare Inherited and De Novo CNVs Reveal Complex Contributions to ASD Risk in Multiplex Families. <i>American Journal of Human Genetics</i> , 2016, 99, 540-554.	2.6	179
89	A gut-derived metabolite alters brain activity and anxiety behaviour in mice. <i>Nature</i> , 2022, 602, 647-653.	13.7	179
90	Neurobehavioral phenotype of Klinefelter syndrome. <i>Mental Retardation and Developmental Disabilities Research Reviews</i> , 2000, 6, 107-116.	3.5	176

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91	Genetic Control of Expression and Splicing in Developing Human Brain Informs Disease Mechanisms. <i>Cell</i> , 2019, 179, 750-771.e22.	13.5	174
92	A genome-wide association study of shared risk across psychiatric disorders implicates gene regulation during fetal neurodevelopment. <i>Nature Neuroscience</i> , 2019, 22, 353-361.	7.1	173
93	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. <i>Alzheimer's and Dementia</i> , 2017, 13, 727-738.	0.4	166
94	Tet3 regulates synaptic transmission and homeostatic plasticity via DNA oxidation and repair. <i>Nature Neuroscience</i> , 2015, 18, 836-843.	7.1	164
95	Endocannabinoid signaling mediates oxytocin-driven social reward. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 14084-14089.	3.3	163
96	Genome-wide, integrative analysis implicates microRNA dysregulation in autism spectrum disorder. <i>Nature Neuroscience</i> , 2016, 19, 1463-1476.	7.1	163
97	Neuron-specific signatures in the chromosomal connectome associated with schizophrenia risk. <i>Science</i> , 2018, 362, .	6.0	162
98	Widespread RNA editing dysregulation in brains from autistic individuals. <i>Nature Neuroscience</i> , 2019, 22, 25-36.	7.1	161
99	Signaling to Transcription Networks in the Neuronal Retrograde Injury Response. <i>Science Signaling</i> , 2010, 3, ra53.	1.6	159
100	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel. <i>JAMA Neurology</i> , 2021, 78, 102.	4.5	144
101	Mechanistic Differences in Neuropathic Pain Modalities Revealed by Correlating Behavior with Global Expression Profiling. <i>Cell Reports</i> , 2018, 22, 1301-1312.	2.9	142
102	Transcriptomic and cellular decoding of regional brain vulnerability to neurogenetic disorders. <i>Nature Communications</i> , 2020, 11, 3358.	5.8	141
103	Altered network connectivity in frontotemporal dementia with C9orf72 hexanucleotide repeat expansion. <i>Brain</i> , 2014, 137, 3047-3060.	3.7	140
104	Sex-chromosome dosage effects on gene expression in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 7398-7403.	3.3	139
105	Timing and significance of pathological features in C9orf72 expansion-associated frontotemporal dementia. <i>Brain</i> , 2016, 139, 3202-3216.	3.7	136
106	Genetics of autism spectrum disorder. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 147, 321-329.	1.0	135
107	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133
108	Functional Genomic Analyses Identify Pathways Dysregulated by Progranulin Deficiency, Implicating Wnt Signaling. <i>Neuron</i> , 2011, 71, 1030-1042.	3.8	132

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109	A Genomic Screen for Modifiers of Tauopathy Identifies Puromycin-Sensitive Aminopeptidase as an Inhibitor of Tau-Induced Neurodegeneration. <i>Neuron</i> , 2006, 51, 549-560.	3.8	130
110	Tau Phosphorylation, Tangles, and Neurodegeneration. <i>Neuron</i> , 2003, 40, 457-460.	3.8	129
111	From genotype to phenotype: A clinical, pathological, and biochemical investigation of frontotemporal dementia and parkinsonism (FTDP-17) caused by the P301L tau mutation. <i>Annals of Neurology</i> , 1999, 45, 704-715.	2.8	128
112	Timing of the Diagnosis of Autism in African American Children. <i>Pediatrics</i> , 2020, 146, e20193629.	1.0	124
113	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. <i>Nature Neuroscience</i> , 2017, 20, 1661-1668.	7.1	122
114	Cerebellar associative sensory learning defects in five mouse autism models. <i>ELife</i> , 2015, 4, e06085.	2.8	120
115	Induced Pluripotent Stem Cell Models of Progranulin-Deficient Frontotemporal Dementia Uncover Specific Reversible Neuronal Defects. <i>Cell Reports</i> , 2012, 2, 789-798.	2.9	118
116	Correspondence between Resting-State Activity and Brain Gene Expression. <i>Neuron</i> , 2015, 88, 659-666.	3.8	117
117	Conserved brain myelination networks are altered in Alzheimer's and other neurodegenerative diseases. <i>Alzheimer's and Dementia</i> , 2018, 14, 352-366.	0.4	116
118	Neuropsychological profiles of adults with Klinefelter syndrome. <i>Journal of the International Neuropsychological Society</i> , 2001, 7, 446-456.	1.2	114
119	The organization of the transcriptional network in specific neuronal classes. <i>Molecular Systems Biology</i> , 2009, 5, 291.	3.2	114
120	Tau interactome maps synaptic and mitochondrial processes associated with neurodegeneration. <i>Cell</i> , 2022, 185, 712-728.e14.	13.5	114
121	Autism-like phenotype and risk gene mRNA deadenylation by CPEB4 mis-splicing. <i>Nature</i> , 2018, 560, 441-446.	13.7	113
122	Neuronal defects in a human cellular model of 22q11.2 deletion syndrome. <i>Nature Medicine</i> , 2020, 26, 1888-1898.	15.2	113
123	Late-Onset Friedreich Ataxia. <i>Archives of Neurology</i> , 2005, 62, 1865.	4.9	111
124	Identification of evolutionarily conserved gene networks mediating neurodegenerative dementia. <i>Nature Medicine</i> , 2019, 25, 152-164.	15.2	111
125	Functional genomic analysis of frataxin deficiency reveals tissue-specific alterations and identifies the PPAR $\delta$ pathway as a therapeutic target in Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2009, 18, 2452-2461.	1.4	109
126	p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR). <i>Cell</i> , 2021, 184, 689-708.e20.	13.5	104



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127	Prevalence of rearrangements in the 22q11.2 region and population-based risk of neuropsychiatric and developmental disorders in a Danish population: a case-cohort study. <i>Lancet Psychiatry</i> , 2018, 5, 573-580.	3.7	102
128	Human evolved regulatory elements modulate genes involved in cortical expansion and neurodevelopmental disease susceptibility. <i>Nature Communications</i> , 2019, 10, 2396.	5.8	98
129	Conservation and divergence of vulnerability and responses to stressors between human and mouse astrocytes. <i>Nature Communications</i> , 2021, 12, 3958.	5.8	94
130	Molecular approaches to cerebral laterality: Development and neurodegeneration. <i>American Journal of Medical Genetics Part A</i> , 2001, 101, 370-381.	2.4	93
131	Human iPSC-Derived Neuronal Model of Tau-A152T Frontotemporal Dementia Reveals Tau-Mediated Mechanisms of Neuronal Vulnerability. <i>Stem Cell Reports</i> , 2016, 7, 325-340.	2.3	92
132	Genetic architecture of epigenetic and neuronal ageing rates in human brain regions. <i>Nature Communications</i> , 2017, 8, 15353.	5.8	92
133	Neuronal CTGF/CCN2 negatively regulates myelination in a mouse model of tuberous sclerosis complex. <i>Journal of Experimental Medicine</i> , 2017, 214, 681-697.	4.2	91
134	A framework for the investigation of rare genetic disorders in neuropsychiatry. <i>Nature Medicine</i> , 2019, 25, 1477-1487.	15.2	90
135	Robust Axonal Regeneration Occurs in the Injured CAST/Ei Mouse CNS. <i>Neuron</i> , 2015, 86, 1215-1227.	3.8	87
136	Neurons selectively targeted in frontotemporal dementia reveal early stage TDP-43 pathobiology. <i>Acta Neuropathologica</i> , 2019, 137, 27-46.	3.9	87
137	Analysis of the immune response to sciatic nerve injury identifies efferocytosis as a key mechanism of nerve debridement. <i>ELife</i> , 2020, 9, .	2.8	85
138	A152T tau allele causes neurodegeneration that can be ameliorated in a zebrafish model by autophagy induction. <i>Brain</i> , 2017, 140, 1128-1146.	3.7	84
139	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. <i>Nature Medicine</i> , 2020, 26, 1754-1765.	15.2	84
140	Recurrence rates provide evidence for sex-differential, familial genetic liability for autism spectrum disorders in multiplex families and twins. <i>Molecular Autism</i> , 2015, 6, 27.	2.6	81
141	Reduced Prefrontal Synaptic Connectivity and Disturbed Oscillatory Population Dynamics in the CNTNAP2 Model of Autism. <i>Cell Reports</i> , 2019, 27, 2567-2578.e6.	2.9	80
142	Increased prevalence of autoimmune disease within C9 and FTD/MND cohorts. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2016, 3, e301.	3.1	78
143	C9orf72 deficiency promotes microglial-mediated synaptic loss in aging and amyloid accumulation. <i>Neuron</i> , 2021, 109, 2275-2291.e8.	3.8	78
144	Joint genome-wide association study of progressive supranuclear palsy identifies novel susceptibility loci and genetic correlation to neurodegenerative diseases. <i>Molecular Neurodegeneration</i> , 2018, 13, 41.	4.4	77

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145	Brain calcification process and phenotypes according to age and sex: Lessons from <i>SLC20A2</i> , <i>PDGFB</i> , and <i>PDGFRB</i> mutation carriers. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 586-594.	1.1	74
146	Alteration in basal and depolarization induced transcriptional network in iPSC derived neurons from Timothy syndrome. <i>Genome Medicine</i> , 2014, 6, 75.	3.6	72
147	Wnt genes define distinct boundaries in the developing human brain: Implications for human forebrain patterning. <i>Journal of Comparative Neurology</i> , 2004, 474, 276-288.	0.9	70
148	Genome-wide DNA methylation profiling identifies convergent molecular signatures associated with idiopathic and syndromic autism in post-mortem human brain tissue. <i>Human Molecular Genetics</i> , 2019, 28, 2201-2211.	1.4	70
149	Recent ultra-rare inherited variants implicate new autism candidate risk genes. <i>Nature Genetics</i> , 2021, 53, 1125-1134.	9.4	68
150	An Epigenetic Signature in Peripheral Blood Associated with the Haplotype on 17q21.31, a Risk Factor for Neurodegenerative Tauopathy. <i>PLoS Genetics</i> , 2014, 10, e1004211.	1.5	65
151	Inducible and reversible phenotypes in a novel mouse model of Friedreich's Ataxia. <i>ELife</i> , 2017, 6, .	2.8	64
152	Integrative genomics identifies a convergent molecular subtype that links epigenomic with transcriptomic differences in autism. <i>Nature Communications</i> , 2020, 11, 4873.	5.8	62
153	Novel Roles for Osteopontin and Clusterin in Peripheral Motor and Sensory Axon Regeneration. <i>Journal of Neuroscience</i> , 2014, 34, 1689-1700.	1.7	61
154	Dissecting the molecular basis of human interneuron migration in forebrain assembloids from Timothy syndrome. <i>Cell Stem Cell</i> , 2022, 29, 248-264.e7.	5.2	61
155	VolCE: A semi-automated pipeline for standardizing vocal analysis across models. <i>Scientific Reports</i> , 2015, 5, 10237.	1.6	59
156	DNA microarrays: translation of the genome from laboratory to clinic. <i>Lancet Neurology</i> , The, 2003, 2, 275-282.	4.9	58
157	A gene expression phenotype in lymphocytes from friedreich ataxia patients. <i>Annals of Neurology</i> , 2011, 70, 790-804.	2.8	58
158	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	1.1	58
159	Tau Pathology Drives Dementia Risk-Associated Gene Networks toward Chronic Inflammatory States and Immunosuppression. <i>Cell Reports</i> , 2020, 33, 108398.	2.9	57
160	Sharing gene expression data: an array of options. <i>Nature Reviews Neuroscience</i> , 2001, 2, 435-438.	4.9	55
161	A Rare Mutation of $\beta$ 21-Adrenergic Receptor Affects Sleep/Wake Behaviors. <i>Neuron</i> , 2019, 103, 1044-1055.e7.	3.8	54
162	Spatiotemporal dynamics of the postnatal developing primate brain transcriptome. <i>Human Molecular Genetics</i> , 2015, 24, 4327-4339.	1.4	53

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163	Molecular Genetics of Neurodegenerative Dementias. Cold Spring Harbor Perspectives in Biology, 2017, 9, a023705.	2.3	51
164	Strong correlation of downregulated genes related to synaptic transmission and mitochondria in post-mortem autism cerebral cortex. Journal of Neurodevelopmental Disorders, 2018, 10, 18.	1.5	51
165	Genetic correlates of phenotypic heterogeneity in autism. Nature Genetics, 2022, 54, 1293-1304.	9.4	51
166	Gene expression profiling in frataxin deficient mice: Microarray evidence for significant expression changes without detectable neurodegeneration. Neurobiology of Disease, 2006, 22, 302-311.	2.1	50
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