Joseph D Buxbaum

List of Publications by Year
in descending order

367	64,478	1370 108	1089 232
papers	citations	h-index	g-index

449	449	449	55884
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458.	9.4	3,741
2	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
3	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
4	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
5	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
6	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. Nature Genetics, 2011, 43, 436-441.	9.4	1,676
7	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
8	Identification of common genetic risk variants for autism spectrum disorder. Nature Genetics, 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. Cell, 2020, 180, 568-584.e23.	13.5	1,422
10	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	9.4	1,272
11	Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. Nature, 2009, 459, 569-573.	13.7	1,270
12	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
13	Most genetic risk for autism resides with common variation. Nature Genetics, 2014, 46, 881-885.	9.4	977
14	Gene expression elucidates functional impact of polygenic risk for schizophrenia. Nature Neuroscience, 2016, 19, 1442-1453.	7.1	952
15	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
16	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
17	Intraneuronal AÎ²42 Accumulation in Human Brain. American Journal of Pathology, 2000, 156, 15-20.	1.9	930
18	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	13.7	929

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19	Common genetic variants on 5p14.1 associate with autism spectrum disorders. Nature, 2009, 459, 528-533.	13.7	912
20	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. Science, 2018, 359, 693-697.	6.0	851
21	Evidence That Tumor Necrosis Factor Î± Converting Enzyme Is Involved in Regulated Î±-Secretase Cleavage of the Alzheimer Amyloid Protein Precursor. Journal of Biological Chemistry, 1998, 273, 27765-27767.	1.6	848
22	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	9.4	838
23	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
24	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
25	White Matter Changes in Schizophrenia. Archives of General Psychiatry, 2003, 60, 443.	13.8	761
26	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Cell, 2018, 173, 1705-1715.e16.	13.5	623
27	Sequencing of the sea lamprey (Petromyzon marinus) genome provides insights into vertebrate evolution. Nature Genetics, 2013, 45, 415-421.	9.4	588
28	Estrogen reduces neuronal generation of Alzheimer Î²-amyloid peptides. Nature Medicine, 1998, 4, 447-451.	15.2	545
29	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
30	Haploinsufficiency of the autism-associated Shank3 gene leads to deficits in synaptic function, social interaction, and social communication. Molecular Autism, 2010, 1, 15.	2.6	521
31	A spectral approach integrating functional genomic annotations for coding and noncoding variants. Nature Genetics, 2016, 48, 214-220.	9.4	506
32	Sequence Kernel Association Tests for the Combined Effect of Rare and Common Variants. American Journal of Human Genetics, 2013, 92, 841-853.	2.6	393
33	Altered ultrasonic vocalization in mice with a disruption in the Foxp2 gene. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 9643-9648.	3.3	389
34	Meta-analysis Confirms CR1, CLU, and PICALM as Alzheimer Disease Risk Loci and Reveals Interactions With APOE Genotypes. Archives of Neurology, 2010, 67, 1473.	4.9	376
35	Genome-Wide Analyses of Exonic Copy Number Variants in a Family-Based Study Point to Novel Autism Susceptibility Genes. PLoS Genetics, 2009, 5, e1000536.	1.5	374
36	Neuregulin 1-erbB signaling and the molecular/cellular basis of schizophrenia. Nature Neuroscience, 2004, 7, 575-580.	7.1	361

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37	Variants in the ATP-Binding Cassette Transporter (ABCA7), Apolipoprotein E Ïµ4, and the Risk of Late-Onset Alzheimer Disease in African Americans. JAMA - Journal of the American Medical Association, 2013, 309, 1483.	3.8	360
38	Reduced Excitatory Neurotransmission and Mild Autism-Relevant Phenotypes in AdolescentShank3Null Mutant Mice. Journal of Neuroscience, 2012, 32, 6525-6541.	1.7	342
39	AÎ² localization in abnormal endosomes: association with earliest AÎ² elevations in AD and Down syndrome. Neurobiology of Aging, 2004, 25, 1263-1272.	1.5	338
40	Autism spectrum disorder: neuropathology and animal models. Acta Neuropathologica, 2017, 134, 537-566.	3.9	335
41	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	1.4	334
42	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
43	Calsenilin: A calcium-binding protein that interacts with the presenilins and regulates the levels of a presenilin fragment. Nature Medicine, 1998, 4, 1177-1181.	15.2	331
44	PGC-1Î± Expression Decreases in the Alzheimer Disease Brain as a Function of Dementia. Archives of Neurology, 2009, 66, 352-61.	4.9	323
45	The Mount Sinai cohort of large-scale genomic, transcriptomic and proteomic data in Alzheimer's disease. Scientific Data, 2018, 5, 180185.	2.4	320
46	Association of Genetic and Environmental Factors With Autism in a 5-Country Cohort. JAMA Psychiatry, 2019, 76, 1035.	6.0	319
47	Enhanced Striatal Dopamine Transmission and Motor Performance with LRRK2 Overexpression in Mice Is Eliminated by Familial Parkinson's Disease Mutation G2019S. Journal of Neuroscience, 2010, 30, 1788-1797.	1.7	309
48	Genome-Wide Association Meta-analysis of Neuropathologic Features of Alzheimer's Disease and Related Dementias. PLoS Genetics, 2014, 10, e1004606.	1.5	305
49	Evidence for a Susceptibility Gene for Autism on Chromosome 2 and for Genetic Heterogeneity. American Journal of Human Genetics, 2001, 68, 1514-1520.	2.6	304
50	The Sac1 Domain of <scp>SYNJ</scp> 1 Identified Mutated in a Family with Earlyâ€Onset Progressive <scp>P</scp> arkinsonism with Generalized Seizures. Human Mutation, 2013, 34, 1200-1207.	1.1	302
51	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	9.4	298
52	Prospective investigation of autism and genotype-phenotype correlations in 22q13 deletion syndrome and SHANK3 deficiency. Molecular Autism, 2013, 4, 18.	2.6	278
53	Tumor Necrosis Factor-Î±-converting Enzyme Is Required for Cleavage of erbB4/HER4. Journal of Biological Chemistry, 2000, 275, 10379-10387.	1.6	277
54	Gene Expression Patterns Associated with Posttraumatic Stress Disorder Following Exposure to the World Trade Center Attacks. Biological Psychiatry, 2009, 66, 708-711.	0.7	273

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55	The emerging role of synaptic cell-adhesion pathways in the pathogenesis of autism spectrum disorders. Trends in Neurosciences, 2009, 32, 402-412.	4.2	271
56	Linking oligodendrocyte and myelin dysfunction to neurocircuitry abnormalities in schizophrenia. Progress in Neurobiology, 2011, 93, 13-24.	2.8	263
57	Epigenetic Biomarkers as Predictors and Correlates of Symptom Improvement Following Psychotherapy in Combat Veterans with PTSD. Frontiers in Psychiatry, 2013, 4, 118.	1.3	263
58	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	1.5	253
59	Alzheimer Amyloid Protein Precursor in the Rat Hippocampus: Transport and Processing through the Perforant Path. Journal of Neuroscience, 1998, 18, 9629-9637.	1.7	249
60	Mutation screening of thePTEN gene in patients with autism spectrum disorders and macrocephaly. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 484-491.	1.1	248
61	HDAC2 regulates atypical antipsychotic responses through the modulation of mGlu2 promoter activity. Nature Neuroscience, 2012, 15, 1245-1254.	7.1	247
62	Exceptionally low likelihood of Alzheimerâ€™s dementia in APOE2 homozygotes from a 5,000-person neuropathological study. Nature Communications, 2020, 11, 667.	5.8	246
63	Autism-like Deficits in Shank3-Deficient Mice Are Rescued by Targeting Actin Regulators. Cell Reports, 2015, 11, 1400-1413.	2.9	245
64	A Replication of the Autism Diagnostic Observation Schedule (ADOS) Revised Algorithms. Journal of the American Academy of Child and Adolescent Psychiatry, 2008, 47, 642-651.	0.3	243
65	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
66	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
67	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
68	Integrative network analysis of nineteen brain regions identifies molecular signatures and networks underlying selective regional vulnerability to Alzheimerâ€™s disease. Genome Medicine, 2016, 8, 104.	3.6	224
69	Strong synaptic transmission impact by copy number variations in schizophrenia. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 10584-10589.	3.3	212
70	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	7.1	212
71	cGAS drives noncanonical-inflammasome activation in age-related macular degeneration. Nature Medicine, 2018, 24, 50-61.	15.2	205
72	The Alzheimer Amyloid Precursor Protein (APP) and Fe65, an APP-Binding Protein, Regulate Cell Movement. Journal of Cell Biology, 2001, 153, 1403-1414.	2.3	204

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73	The increasing prevalence of reported diagnoses of childhood psychiatric disorders: a descriptive multinational comparison. European Child and Adolescent Psychiatry, 2015, 24, 173-183.	2.8	201
74	Regulation of Î²-Amyloid Secretion by FE65, an Amyloid Protein Precursor-binding Protein. Journal of Biological Chemistry, 1999, 274, 7952-7957.	1.6	200
75	Linkage and Association of the Mitochondrial Aspartate/Glutamate Carrier SLC25A12 Gene With Autism. American Journal of Psychiatry, 2004, 161, 662-669.	4.0	185
76	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	1.8	180
77	Molecular and cellular evidence for an oligodendrocyte abnormality in schizophrenia. Neurochemical Research, 2002, 27, 1193-1200.	1.6	175
78	Assessment of the genetic variance of late-onset Alzheimer's disease. Neurobiology of Aging, 2016, 41, 200.e13-200.e20.	1.5	174
79	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.4	173
80	Putative biological mechanisms for the association between early life adversity and the subsequent development of PTSD. Psychopharmacology, 2010, 212, 405-417.	1.5	172
81	Effects of Multiple Genetic Loci on Age at Onset in Late-Onset Alzheimer Disease. JAMA Neurology, 2014, 71, 1394.	4.5	166
82	Transethnic genomeâ€wide scan identifies novel Alzheimer's disease loci. Alzheimer's and Dementia, 2017, 13, 727-738.	0.4	166
83	The Amyloid Precursor Protein and Its Regulatory Protein, FE65, in Growth Cones and SynapsesIn VitroandIn Vivo. Journal of Neuroscience, 2003, 23, 5407-5415.	1.7	160
84	Genetics and genomics of autism spectrum disorder: embracing complexity. Human Molecular Genetics, 2015, 24, R24-R31.	1.4	160
85	Whole-Exome Sequencing Links a Variant in DHDDS to Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 88, 201-206.	2.6	155
86	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	1.1	155
87	The Autism Sequencing Consortium: Large-Scale, High-Throughput Sequencing in Autism Spectrum Disorders. Neuron, 2012, 76, 1052-1056.	3.8	153
88	SHANK3 haploinsufficiency: a â€œcommonâ€•but underdiagnosed highly penetrant monogenic cause of autism spectrum disorders. Molecular Autism, 2013, 4, 17.	2.6	152
89	Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by SHANK3 point mutations. Molecular Autism, 2018, 9, 31.	2.6	152
90	Insulin-like growth factor-1 rescues synaptic and motor deficits in a mouse model of autism and developmental delay. Molecular Autism, 2013, 4, 9.	2.6	150

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91	CommonMind Consortium provides transcriptomic and epigenomic data for Schizophrenia and Bipolar Disorder. Scientific Data, 2019, 6, 180.	2.4	149
92	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. Neurology, 2012, 79, 221-228.	1.5	144
93	Characterization of New Polyclonal Antibodies Specific for 40 and 42 Amino Acid-Long Amyloid Î² Peptides: Their Use to Examine the Cell Biology of Presenilins and the Immunohistochemistry of Sporadic Alzheimerâ€™s Disease and Cerebral Amyloid Angiopathy Cases. Molecular Medicine, 1997, 3, 695-707.	1.9	142
94	Interaction of the Phosphotyrosine Interaction/Phosphotyrosine Binding-related Domains of Fe65 with Wild-type and Mutant Alzheimer's Î²-Amyloid Precursor Proteins. Journal of Biological Chemistry, 1997, 272, 6399-6405.	1.6	141
95	A New Testing Strategy to Identify Rare Variants with Either Risk or Protective Effect on Disease. PLoS Genetics, 2011, 7, e1001289.	1.5	141
96	Identification of Small Exonic CNV from Whole-Exome Sequence Data and Application to Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 93, 607-619.	2.6	136
97	Oxytocin improves behavioral and electrophysiological deficits in a novel Shank3-deficient rat. ELife, 2017, 6, .	2.8	136
98	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
99	A critical role for the protein tyrosine phosphatase receptor type Z in functional recovery from demyelinating lesions. Nature Genetics, 2002, 32, 411-414.	9.4	132
100	Dementia Revealed: Novel Chromosome 6 Locus for Late-Onset Alzheimer Disease Provides Genetic Evidence for Folate-Pathway Abnormalities. PLoS Genetics, 2010, 6, e1001130.	1.5	130
101	Regulatory consequences of neuronal ELAV-like protein binding to coding and non-coding RNAs in human brain. ELife, 2016, 5, .	2.8	128
102	Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS. American Journal of Human Genetics, 2018, 102, 1169-1184.	2.6	128
103	Phelan-McDermid syndrome: a review of the literature and practice parameters for medical assessment and monitoring. Journal of Neurodevelopmental Disorders, 2014, 6, 39.	1.5	122
104	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
105	Genetic Markers for PTSD Risk and Resilience Among Survivors of the World Trade Center Attacks. Disease Markers, 2011, 30, 101-110.	0.6	117
106	Calcium-regulated DNA Binding and Oligomerization of the Neuronal Calcium-sensing Protein, Calsenilin/DREAM/KChIP3. Journal of Biological Chemistry, 2001, 276, 41005-41013.	1.6	116
107	Convergent evidence that oligodendrocyte lineage transcription factor 2 (OLIG2) and interacting genes influence susceptibility to schizophrenia. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 12469-12474.	3.3	116
108	Convergent Evidence for 2â€²,3â€²-Cyclic Nucleotide 3â€²-Phosphodiesterase as a Possible Susceptibility Gene for Schizophrenia. Archives of General Psychiatry, 2006, 63, 18.	13.8	115

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109	Multiple rare variants in the etiology of autism spectrum disorders. Dialogues in Clinical Neuroscience, 2009, 11, 35-43.	1.8	115
110	Expression profiling associates blood and brain glucocorticoid receptor signaling with trauma-related individual differences in both sexes. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 13529-13534.	3.3	113
111	Symptom domains in autism and related conditions: Evidence for familiality. American Journal of Medical Genetics Part A, 2002, 114, 64-73.	2.4	111
112	Generation and Regulation of Î²â€Amyloid Peptide Variants by Neurons. Journal of Neurochemistry, 1998, 71, 1920-1925.	2.1	111
113	Insulin degrading enzyme activity selectively decreases in the hippocampal formation of cases at high risk to develop Alzheimer's disease. Neurobiology of Aging, 2007, 28, 824-830.	1.5	111
114	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. Journal of Molecular Medicine, 2013, 91, 1399-1406.	1.7	111
115	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. Molecular Autism, 2014, 5, 22.	2.6	111
116	Haploinsufficiency of Gtf2i, a gene deleted in Williams Syndrome, leads to increases in social interactions. Autism Research, 2011, 4, 28-39.	2.1	109
117	A pilot controlled trial of insulin-like growth factor-1 in children with Phelan-McDermid syndrome. Molecular Autism, 2014, 5, 54.	2.6	109
118	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	9.4	109
119	Understanding autism in the light of sex/gender. Molecular Autism, 2015, 6, 24.	2.6	107
120	The Carboxyl-Terminus of BACE Contains a Sorting Signal That Regulates BACE Trafficking but Not the Formation of Total AÎ². Molecular and Cellular Neurosciences, 2002, 19, 175-185.	1.0	106
121	Altered AÎ² Formation and Long-Term Potentiation in a Calsenilin Knock-Out. Journal of Neuroscience, 2003, 23, 9097-9106.	1.7	103
122	Regulation of Secretion of Alzheimer Amyloid Precursor Protein by the Mitogenâ€Activated Protein Kinase Cascade. Journal of Neurochemistry, 1998, 70, 524-530.	2.1	102
123	Canonical Inflammasomes Drive IFN-Î³ to Prime Caspase-11 in Defense against a Cytosol-Invasive Bacterium. Cell Host and Microbe, 2015, 18, 320-332.	5.1	101
124	In vivo 1H-magnetic resonance spectroscopy study of the attentional networks in autism. Brain Research, 2011, 1380, 198-205.	1.1	98
125	Correlation Between AÎ²x-40â€“, AÎ²x-42â€“, and AÎ²x-43â€“Containing Amyloid Plaques and Cognitive Decline. Archives of Neurology, 2001, 58, 2025.	4.9	95
126	Haploinsufficiency of Cyfip1 Produces Fragile X-Like Phenotypes in Mice. PLoS ONE, 2012, 7, e42422.	1.1	95

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127	A Critical Role for Human Caspase-4 in Endotoxin Sensitivity. Journal of Immunology, 2014, 193, 335-343.	0.4	95
128	Two rare AKAP9 variants are associated with Alzheimer's disease in African Americans. Alzheimer's and Dementia, 2014, 10, 609.	0.4	94
129	Mosaic Epigenetic Dysregulation of Ectodermal Cells in Autism Spectrum Disorder. PLoS Genetics, 2014, 10, e1004402.	1.5	93
130	PTSD Blood Transcriptome Mega-Analysis: Shared Inflammatory Pathways across Biological Sex and Modes of Trauma. Neuropsychopharmacology, 2018, 43, 469-481.	2.8	92
131	A Genome-wide Study Reveals Copy Number Variants Exclusive to Childhood Obesity Cases. American Journal of Human Genetics, 2010, 87, 661-666.	2.6	91
132	Large-Scale Identification of Common Trait and Disease Variants Affecting Gene Expression. American Journal of Human Genetics, 2017, 100, 885-894.	2.6	91
133	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	2.9	91
134	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. Cell Reports, 2020, 31, 107489.	2.9	91
135	Elevated polygenic burden for autism is associated with differential DNA methylation at birth. Genome Medicine, 2018, 10, 19.	3.6	88
136	Treatment with controlled-release lovastatin decreases serum concentrations of human Î²-amyloid (AÎ²) peptide. International Journal of Neuropsychopharmacology, 2001, 4, 127-30.	1.0	87
137	Two novel loci, COBL and SLC10A2, for Alzheimer's disease in African Americans. Alzheimer's and Dementia, 2017, 13, 119-129.	0.4	87
138	Scan statistic-based analysis of exome sequencing data identifies FAN1 at 15q13.3 as a susceptibility gene for schizophrenia and autism. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 343-348.	3.3	86
139	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. Genome Medicine, 2017, 9, 114.	3.6	86
140	Lysosomal Dysfunction in a Mouse Model of Sandhoff Disease Leads to Accumulation of Ganglioside-Bound Amyloid-Î² Peptide. Journal of Neuroscience, 2012, 32, 5223-5236.	1.7	84
141	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	7.7	83
142	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
143	Family-based association tests for sequence data, and comparisons with population-based association tests. European Journal of Human Genetics, 2013, 21, 1158-1162.	1.4	81
144	Behavioral Phenotyping of an Improved Mouse Model of Phelanâ€“McDermid Syndrome with a Complete Deletion of theShank3Gene. ENeuro, 2018, 5, ENEURO.0046-18.2018.	0.9	79

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145	New translational perspectives for blood-based biomarkers of PTSD: From glucocorticoid to immune mediators of stress susceptibility. Experimental Neurology, 2016, 284, 133-140.	2.0	78
146	Developmental social communication deficits in the Shank3 rat model of phelanâ€mcdermid syndrome and autism spectrum disorder. Autism Research, 2018, 11, 587-601.	2.1	78
147	Alzheimer Amyloid Protein Precursor Is Localized in Nerve Terminal Preparations to Rab5-containing Vesicular Organelles Distinct from Those Implicated in the Synaptic Vesicle Pathway. Journal of Biological Chemistry, 1996, 271, 31783-31786.	1.6	77
148	Advancing Paternal Age Is Associated with Deficits in Social and Exploratory Behaviors in the Offspring: A Mouse Model. PLoS ONE, 2009, 4, e8456.	1.1	77
149	FUN-LDA: A Latent Dirichlet Allocation Model for Predicting Tissue-Specific Functional Effects of Noncoding Variation: Methods and Applications. American Journal of Human Genetics, 2018, 102, 920-942.	2.6	75
150	Multiplex ligation-dependent probe amplification for genetic screening in autism spectrum disorders: Efficient identification of known microduplications and identification of a novel microduplication in ASMT. BMC Medical Genomics, 2008, 1, 50.	0.7	74
151	Atorvastatin-induced activation of Alzheimer's alpha secretase is resistant to standard inhibitors of protein phosphorylation-regulated ectodomain shedding. Journal of Neurochemistry, 2004, 90, 1005-1010.	2.1	69
152	Formamidines interact withDrosophila octopamine receptors, alter the flies' behavior and reduce their learning ability. Journal of Comparative Physiology A: Neuroethology, Sensory, Neural, and Behavioral Physiology, 1987, 161, 739-746.	0.7	68
153	Phenotypic and functional analysis of SHANK3 stop mutations identified in individuals with ASD and/or ID. Molecular Autism, 2015, 6, 23.	2.6	68
154	Linking white and grey matter in schizophrenia: Oligodendrocyte and neuron pathology in the prefrontal cortex. Frontiers in Neuroanatomy, 2009, 3, 9.	0.9	67
155	Familial Clustering of Tic Disorders and Obsessive-Compulsive Disorder. JAMA Psychiatry, 2015, 72, 359.	6.0	67
156	Two knockdown models of the autism genes SYNGAP1 and SHANK3 in zebrafish produce similar behavioral phenotypes associated with embryonic disruptions of brain morphogenesis. Human Molecular Genetics, 2015, 24, 4006-4023.	1.4	67
157	Amyloid beta protein-induced zinc sequestration leads to synaptic loss via dysregulation of the ProSAP2/Shank3 scaffold. Molecular Neurodegeneration, 2011, 6, 65.	4.4	66
158	Prospective investigation of FOXP1 syndrome. Molecular Autism, 2017, 8, 57.	2.6	65
159	Genetic markers for PTSD risk and resilience among survivors of the World Trade Center attacks. Disease Markers, 2011, 30, 101-10.	0.6	65
160	Cholesterol depletion with physiological concentrations of a statin decreases the formation of the Alzheimer amyloid Aî² peptide. Journal of Alzheimer's Disease, 2001, 3, 221-229.	1.2	65
161	Global landscape and genetic regulation of RNA editing in cortical samples from individuals with schizophrenia. Nature Neuroscience, 2019, 22, 1402-1412.	7.1	63
162	Phelan McDermid Syndrome. Journal of Child Neurology, 2015, 30, 1861-1870.	0.7	62

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163	Highly conserved molecular pathways, including Wnt signaling, promote functional recovery from spinal cord injury in lampreys. Scientific Reports, 2018, 8, 742.	1.6	62
164	A high proportion of polymorphisms in the promoters of brain expressed genes influences transcriptional activity. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2004, 1690, 238-249.	1.8	61
165	Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders. Biological Psychiatry, 2022, 91, 102-117.	0.7	61
166	Association analysis of the NrCAM gene in autism and in subsets of families with severe obsessiveâ€“compulsive or self-stimulatory behaviors. Psychiatric Genetics, 2006, 16, 251-257.	0.6	60
167	<scp>PLXNA</scp>4 is associated with <scp>A</scp>lzheimer disease and modulates tau phosphorylation. Annals of Neurology, 2014, 76, 379-392.	2.8	60
168	Calsenilin Is a Substrate for Caspase-3 That Preferentially Interacts with the Familial Alzheimer's Disease-associated C-terminal Fragment of Presenilin 2. Journal of Biological Chemistry, 2001, 276, 19197-19204.	1.6	59
169	A clinicianâ€administered observation and corresponding caregiver interview capturing DSMâ€5 sensory reactivity symptoms in children with ASD. Autism Research, 2017, 10, 1133-1140.	2.1	59
170	GJA1 (connexin43) is a key regulator of Alzheimerâ€™s disease pathogenesis. Acta Neuropathologica Communications, 2018, 6, 144.	2.4	59
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