Joseph D Buxbaum

List of Publications by Citations

371 papers	45,877 citations	101 h-index	210 g-index
449 ext. papers	57,041 ext. citations	9.9 avg, IF	6.69 L-index

#	Paper	IF	Citations
371	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
370	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
369	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
368	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
367	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. *Nature Genetics*, **2011**, 43, 436-41	36.3	1367
366	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
365	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
364	Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. *Nature*, **2009**, 459, 569-73	50.4	1075
363	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates Aβ, tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
362	Intraneuronal Abeta42 accumulation in human brain. *American Journal of Pathology*, **2000**, 156, 15-20	5.8	827
361	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
360	Common genetic variants on 5p14.1 associate with autism spectrum disorders. *Nature*, **2009**, 459, 528-33	50.4	760
359	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746
358	Evidence that tumor necrosis factor alpha converting enzyme is involved in regulated alpha-secretase cleavage of the Alzheimer amyloid protein precursor. *Journal of Biological Chemistry*, **1998**, 273, 27765-7	5.4	744
357	Most genetic risk for autism resides with common variation. *Nature Genetics*, **2014**, 46, 881-5	36.3	734
356	White matter changes in schizophrenia: evidence for myelin-related dysfunction. *Archives of General Psychiatry*, **2003**, 60, 443-56		662
355	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656

354	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
353	Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nature Neuroscience*, **2016**, 19, 1442-1453	25.5	622
352	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
351	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
350	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. *Nature Genetics*, **2017**, 49, 27-35	36.3	530
349	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
348	Estrogen reduces neuronal generation of Alzheimer beta-amyloid peptides. *Nature Medicine*, **1998**, 4, 447-51	50.5	479
347	Sequencing of the sea lamprey (Petromyzon marinus) genome provides insights into vertebrate evolution. *Nature Genetics*, **2013**, 45, 415-21, 421e1-2	36.3	465
346	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443
345	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
344	Haploinsufficiency of the autism-associated Shank3 gene leads to deficits in synaptic function, social interaction, and social communication. *Molecular Autism*, **2010**, 1, 15	6.5	399
343	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. *Cell*, **2018**, 173, 1705-1715.e16	56.2	360
342	Meta-analysis confirms CR1, CLU, and PICALM as alzheimer disease risk loci and reveals interactions with APOE genotypes. *Archives of Neurology*, **2010**, 67, 1473-84		330
341	A spectral approach integrating functional genomic annotations for coding and noncoding variants. *Nature Genetics*, **2016**, 48, 214-20	36.3	327
340	Neuregulin 1-erbB signaling and the molecular/cellular basis of schizophrenia. *Nature Neuroscience*, **2004**, 7, 575-80	25.5	326
339	Calsenilin: a calcium-binding protein that interacts with the presenilins and regulates the levels of a presenilin fragment. *Nature Medicine*, **1998**, 4, 1177-81	50.5	306
338	Genome-wide analyses of exonic copy number variants in a family-based study point to novel autism susceptibility genes. *PLoS Genetics*, **2009**, 5, e1000536	6	305
337	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, 9643-8	11.5	305

336	Sequence kernel association tests for the combined effect of rare and common variants. *American Journal of Human Genetics*, **2013**, 92, 841-53	11	300
335	Abeta localization in abnormal endosomes: association with earliest Abeta elevations in AD and Down syndrome. *Neurobiology of Aging*, **2004**, 25, 1263-72	5.6	281
334	Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. *Nature Genetics*, **2011**, 44, 78-84	36.3	279
333	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
332	Variants in the ATP-binding cassette transporter (ABCA7), apolipoprotein E ?4,and the risk of late-onset Alzheimer disease in African Americans. *JAMA - Journal of the American Medical Association*, **2013**, 309, 1483-92	27.4	275
331	Reduced excitatory neurotransmission and mild autism-relevant phenotypes in adolescent Shank3 null mutant mice. *Journal of Neuroscience*, **2012**, 32, 6525-41	6.6	274
330	Evidence for a susceptibility gene for autism on chromosome 2 and for genetic heterogeneity. *American Journal of Human Genetics*, **2001**, 68, 1514-20	11	273
329	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, 1788-97	6.6	270
328	PGC-1alpha expression decreases in the Alzheimer disease brain as a function of dementia. *Archives of Neurology*, **2009**, 66, 352-61		250
327	Tumor necrosis factor-alpha-converting enzyme is required for cleavage of erbB4/HER4. *Journal of Biological Chemistry*, **2000**, 275, 10379-87	5.4	244
326	Alzheimer amyloid protein precursor in the rat hippocampus: transport and processing through the perforant path. *Journal of Neuroscience*, **1998**, 18, 9629-37	6.6	234
325	Gene expression patterns associated with posttraumatic stress disorder following exposure to the World Trade Center attacks. *Biological Psychiatry*, **2009**, 66, 708-11	7.9	232
324	The Sac1 domain of SYNJ1 identified mutated in a family with early-onset progressive Parkinsonism with generalized seizures. *Human Mutation*, **2013**, 34, 1200-7	4.7	228
323	Genome-wide association meta-analysis of neuropathologic features of Alzheimer's disease and related dementias. *PLoS Genetics*, **2014**, 10, e1004606	6	219
322	Epigenetic Biomarkers as Predictors and Correlates of Symptom Improvement Following Psychotherapy in Combat Veterans with PTSD. *Frontiers in Psychiatry*, **2013**, 4, 118	5	218
321	The emerging role of synaptic cell-adhesion pathways in the pathogenesis of autism spectrum disorders. *Trends in Neurosciences*, **2009**, 32, 402-12	13.3	215
320	HDAC2 regulates atypical antipsychotic responses through the modulation of mGlu2 promoter activity. *Nature Neuroscience*, **2012**, 15, 1245-54	25.5	208
319	Mutation screening of the PTEN gene in patients with autism spectrum disorders and macrocephaly. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2007**, 144B, 484-91	3.5	204

318	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. *Nature Genetics*, **2017**, 49, 504-510	36.3	203
317	Linking oligodendrocyte and myelin dysfunction to neurocircuitry abnormalities in schizophrenia. *Progress in Neurobiology*, **2011**, 93, 13-24	10.9	194
316	Autism spectrum disorder: neuropathology and animal models. *Acta Neuropathologica*, **2017**, 134, 537-566	14.3	191
315	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
314	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	7.2	187
313	The Alzheimer amyloid precursor protein (APP) and FE65, an APP-binding protein, regulate cell movement. *Journal of Cell Biology*, **2001**, 153, 1403-14	7.3	187
312	Prospective investigation of autism and genotype-phenotype correlations in 22q13 deletion syndrome and SHANK3 deficiency. *Molecular Autism*, **2013**, 4, 18	6.5	170
311	Autism-like Deficits in Shank3-Deficient Mice Are Rescued by Targeting Actin Regulators. *Cell Reports*, **2015**, 11, 1400-1413	10.6	170
310	Regulation of beta-amyloid secretion by FE65, an amyloid protein precursor-binding protein. *Journal of Biological Chemistry*, **1999**, 274, 7952-7	5.4	170
309	Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes. *PLoS Genetics*, **2013**, 9, e1003671	6	168
308	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, 10584-9	11.5	165
307	Linkage and association of the mitochondrial aspartate/glutamate carrier SLC25A12 gene with autism. *American Journal of Psychiatry*, **2004**, 161, 662-9	11.9	158
306	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. *Nature Genetics*, **2018**, 50, 727-736	36.3	156
305	Molecular and cellular evidence for an oligodendrocyte abnormality in schizophrenia. *Neurochemical Research*, **2002**, 27, 1193-200	4.6	152
304	Association of Genetic and Environmental Factors With Autism in a 5-Country Cohort. *JAMA Psychiatry*, **2019**, 76, 1035-1043	14.5	151
303	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
302	Putative biological mechanisms for the association between early life adversity and the subsequent development of PTSD. *Psychopharmacology*, **2010**, 212, 405-17	4.7	149
301	The increasing prevalence of reported diagnoses of childhood psychiatric disorders: a descriptive multinational comparison. *European Child and Adolescent Psychiatry*, **2015**, 24, 173-83	5.5	147

300	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
299	The amyloid precursor protein and its regulatory protein, FE65, in growth cones and synapses in vitro and in vivo. *Journal of Neuroscience*, **2003**, 23, 5407-15	6.6	145
298	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. *Nature Neuroscience*, **2017**, 20, 1217-1224	25.5	144
297	The Mount Sinai cohort of large-scale genomic, transcriptomic and proteomic data in Alzheimer's disease. *Scientific Data*, **2018**, 5, 180185	8.2	144
296	Characterization of New Polyclonal Antibodies Specific for 40 and 42 Amino Acid-Long Amyloid Peptides: Their Use to Examine the Cell Biology of Presenilins and the Immunohistochemistry of Sporadic Alzheimer Disease and Cerebral Amyloid Angiopathy Cases. *Molecular Medicine*, **1997**, 3, 695-707	6.2	137
295	Integrative network analysis of nineteen brain regions identifies molecular signatures and networks underlying selective regional vulnerability to Alzheimer's disease. *Genome Medicine*, **2016**, 8, 104	14.4	135
294	cGAS drives noncanonical-inflammasome activation in age-related macular degeneration. *Nature Medicine*, **2018**, 24, 50-61	50.5	134
293	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science*, **2018**, 362,	33.3	134
292	Whole-exome sequencing links a variant in DHDDS to retinitis pigmentosa. *American Journal of Human Genetics*, **2011**, 88, 201-6	11	130
291	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. *JAMA Neurology*, **2014**, 71, 1394-404	17.2	129
290	Interaction of the phosphotyrosine interaction/phosphotyrosine binding-related domains of Fe65 with wild-type and mutant Alzheimer's beta-amyloid precursor proteins. *Journal of Biological Chemistry*, **1997**, 272, 6399-405	5.4	126
289	Genetics and genomics of autism spectrum disorder: embracing complexity. *Human Molecular Genetics*, **2015**, 24, R24-31	5.6	125
288	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron*, **2012**, 76, 1052-6	13.9	124
287	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. *Neurology*, **2012**, 79, 221-8	6.5	124
286	A new testing strategy to identify rare variants with either risk or protective effect on disease. *PLoS Genetics*, **2011**, 7, e1001289	6	121
285	Identification of small exonic CNV from whole-exome sequence data and application to autism spectrum disorder. *American Journal of Human Genetics*, **2013**, 93, 607-19	11	120
284	Assessment of the genetic variance of late-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 41, 200.e13-200.e20	5.6	119
283	A critical role for the protein tyrosine phosphatase receptor type Z in functional recovery from demyelinating lesions. *Nature Genetics*, **2002**, 32, 411-4	36.3	116

282	Insulin-like growth factor-1 rescues synaptic and motor deficits in a mouse model of autism and developmental delay. *Molecular Autism*, **2013**, 4, 9	6.5	115
281	Exceptionally low likelihood of Alzheimer's dementia in APOE2 homozygotes from a 5,000-person neuropathological study. *Nature Communications*, **2020**, 11, 667	17.4	113
280	SHANK3 haploinsufficiency: a "common" but underdiagnosed highly penetrant monogenic cause of autism spectrum disorders. *Molecular Autism*, **2013**, 4, 17	6.5	111
279	Dementia revealed: novel chromosome 6 locus for late-onset Alzheimer disease provides genetic evidence for folate-pathway abnormalities. *PLoS Genetics*, **2010**, 6, e1001130	6	111
278	Genetic Markers for PTSD Risk and Resilience Among Survivors of the World Trade Center Attacks. *Disease Markers*, **2011**, 30, 101-110	3.2	109
277	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
276	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. *Alzheimers and Dementia*, **2017**, 13, 727-738	1.2	106
275	Symptom domains in autism and related conditions: evidence for familiality. *American Journal of Medical Genetics Part A*, **2002**, 114, 64-73		105
274	Calcium-regulated DNA binding and oligomerization of the neuronal calcium-sensing protein, calsenilin/DREAM/KChIP3. *Journal of Biological Chemistry*, **2001**, 276, 41005-13	5.4	104
273	Convergent evidence for 2',3'-cyclic nucleotide 3'-phosphodiesterase as a possible susceptibility gene for schizophrenia. *Archives of General Psychiatry*, **2006**, 63, 18-24		102
272	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, 12469-74	11.5	101
271	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, 824-30	5.6	98
270	Generation and regulation of beta-amyloid peptide variants by neurons. *Journal of Neurochemistry*, **1998**, 71, 1920-5	6	97
269	Altered Abeta formation and long-term potentiation in a calsenilin knock-out. *Journal of Neuroscience*, **2003**, 23, 9097-106	6.6	96
268	The carboxyl-terminus of BACE contains a sorting signal that regulates BACE trafficking but not the formation of total A(beta). *Molecular and Cellular Neurosciences*, **2002**, 19, 175-85	4.8	95
267	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, 7974-81	11.5	94
266	Correlation between Abetax-40-, Abetax-42-, and Abetax-43-containing amyloid plaques and cognitive decline. *Archives of Neurology*, **2001**, 58, 2025-32		90
265	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90

264	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. *Molecular Autism*, **2014**, 5, 22	6.5	89
263	Multiple rare variants in the etiology of autism spectrum disorders. *Dialogues in Clinical Neuroscience*, **2009**, 11, 35-43	5.7	89
262	Canonical Inflammasomes Drive IFN-β to Prime Caspase-11 in Defense against a Cytosol-Invasive Bacterium. *Cell Host and Microbe*, **2015**, 18, 320-32	23.4	88
261	Regulation of secretion of Alzheimer amyloid precursor protein by the mitogen-activated protein kinase cascade. *Journal of Neurochemistry*, **1998**, 70, 524-30	6	86
260	Regulatory consequences of neuronal ELAV-like protein binding to coding and non-coding RNAs in human brain. *ELife*, **2016**, 5,	8.9	86
259	Two rare AKAP9 variants are associated with Alzheimer's disease in African Americans. *Alzheimers and Dementia*, **2014**, 10, 609-618.e11	1.2	83
258	A pilot controlled trial of insulin-like growth factor-1 in children with Phelan-McDermid syndrome. *Molecular Autism*, **2014**, 5, 54	6.5	83
257	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, 13529-34	11.5	82
256	Phelan-McDermid syndrome: a review of the literature and practice parameters for medical assessment and monitoring. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 39	4.6	82
255	In vivo 1H-magnetic resonance spectroscopy study of the attentional networks in autism. *Brain Research*, **2011**, 1380, 198-205	3.7	81
254	Haploinsufficiency of Cyfip1 produces fragile X-like phenotypes in mice. *PLoS ONE*, **2012**, 7, e42422	3.7	79
253	Haploinsufficiency of Gtf2i, a gene deleted in Williams Syndrome, leads to increases in social interactions. *Autism Research*, **2011**, 4, 28-39	5.1	79
252	A critical role for human caspase-4 in endotoxin sensitivity. *Journal of Immunology*, **2014**, 193, 335-43	5.3	78
251	A genome-wide study reveals copy number variants exclusive to childhood obesity cases. *American Journal of Human Genetics*, **2010**, 87, 661-6	11	78
250	Mosaic epigenetic dysregulation of ectodermal cells in autism spectrum disorder. *PLoS Genetics*, **2014**, 10, e1004402	6	76
249	Oxytocin improves behavioral and electrophysiological deficits in a novel Shank3-deficient rat. *ELife* , **2017**, 6,	8.9	75
248	Treatment with controlled-release lovastatin decreases serum concentrations of human beta-amyloid (A beta) peptide. *International Journal of Neuropsychopharmacology*, **2001**, 4, 127-30	5.8	74
247	Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS. *American Journal of Human Genetics*, **2018**, 102, 1169-1184	11	73

246	Understanding autism in the light of sex/gender. *Molecular Autism*, **2015**, 6, 24	6.5	72
245	Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by point mutations. *Molecular Autism*, **2018**, 9, 31	6.5	70
244	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, 343-8	11.5	69
243	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	3.7	68
242	Family-based association tests for sequence data, and comparisons with population-based association tests. *European Journal of Human Genetics*, **2013**, 21, 1158-62	5.3	67
241	Lysosomal dysfunction in a mouse model of Sandhoff disease leads to accumulation of ganglioside-bound amyloid-peptide. *Journal of Neuroscience*, **2012**, 32, 5223-36	6.6	66
240	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, 31783-6	5.4	66
239	Advancing paternal age is associated with deficits in social and exploratory behaviors in the offspring: a mouse model. *PLoS ONE*, **2009**, 4, e8456	3.7	65
238	PTSD Blood Transcriptome Mega-Analysis: Shared Inflammatory Pathways across Biological Sex and Modes of Trauma. *Neuropsychopharmacology*, **2018**, 43, 469-481	8.7	61
237	Linking white and grey matter in schizophrenia: oligodendrocyte and neuron pathology in the prefrontal cortex. *Frontiers in Neuroanatomy*, **2009**, 3, 9	3.6	59
236	Elevated polygenic burden for autism is associated with differential DNA methylation at birth. *Genome Medicine*, **2018**, 10, 19	14.4	58
235	Atorvastatin-induced activation of Alzheimer's alpha secretase is resistant to standard inhibitors of protein phosphorylation-regulated ectodomain shedding. *Journal of Neurochemistry*, **2004**, 90, 1005-10	6	58
234	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, 19197-204	5.4	58
233	Amyloid beta protein-induced zinc sequestration leads to synaptic loss via dysregulation of the ProSAP2/Shank3 scaffold. *Molecular Neurodegeneration*, **2011**, 6, 65	19	57
232	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	2.3	57
231	Recessive gene disruptions in autism spectrum disorder. *Nature Genetics*, **2019**, 51, 1092-1098	36.3	56
230	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, 238-49	6.9	56
229	New translational perspectives for blood-based biomarkers of PTSD: From glucocorticoid to immune mediators of stress susceptibility. *Experimental Neurology*, **2016**, 284, 133-140	5.7	56

228	Genetic markers for PTSD risk and resilience among survivors of the World Trade Center attacks. *Disease Markers*, **2011**, 30, 101-10	3.2	54
227	CommonMind Consortium provides transcriptomic and epigenomic data for Schizophrenia and Bipolar Disorder. *Scientific Data*, **2019**, 6, 180	8.2	52
226	Cholesterol depletion with physiological concentrations of a statin decreases the formation of the Alzheimer amyloid Abeta peptide. *Journal of Alzheimer's Disease*, **2001**, 3, 221-229	4.3	52
225	Developmental social communication deficits in the Shank3 rat model of phelan-mcdermid syndrome and autism spectrum disorder. *Autism Research*, **2018**, 11, 587-601	5.1	51
224	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	10.6	51
223	Association analysis of the NrCAM gene in autism and in subsets of families with severe obsessive-compulsive or self-stimulatory behaviors. *Psychiatric Genetics*, **2006**, 16, 251-7	2.9	50
222	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	11	49
221	Large-Scale Identification of Common Trait and Disease Variants Affecting Gene Expression. *American Journal of Human Genetics*, **2017**, 100, 885-894	11	48
220	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. *Genome Medicine*, **2017**, 9, 114	14.4	48
219	PLXNA4 is associated with Alzheimer disease and modulates tau phosphorylation. *Annals of Neurology*, **2014**, 76, 379-92	9.4	48
218	Two novel loci, COBL and SLC10A2, for Alzheimer's disease in African Americans. *Alzheimer's and Dementia*, **2017**, 13, 119-129	1.2	48
217	Familial clustering of tic disorders and obsessive-compulsive disorder. *JAMA Psychiatry*, **2015**, 72, 359-66	14.5	48
216	SLITRK1 binds 14-3-3 and regulates neurite outgrowth in a phosphorylation-dependent manner. *Biological Psychiatry*, **2009**, 66, 918-25	7.9	48
215	Network topologies and convergent aetiologies arising from deletions and duplications observed in individuals with autism. *PLoS Genetics*, **2013**, 9, e1003523	6	47
214	Recent advances in the genetics of autism spectrum disorder. *Current Neurology and Neuroscience Reports*, **2015**, 15, 36	6.6	46
213	Phenotypic and functional analysis of SHANK3 stop mutations identified in individuals with ASD and/or ID. *Molecular Autism*, **2015**, 6, 23	6.5	45
212	Identification of rare de novo epigenetic variations in congenital disorders. *Nature Communications*, **2018**, 9, 2064	17.4	45
211	Scan-statistic approach identifies clusters of rare disease variants in LRP2, a gene linked and associated with autism spectrum disorders, in three datasets. *American Journal of Human Genetics*, **2012**, 90, 1002-13	11	44

210	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, 4006-23	5.6	43
209	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. *Cell Reports*, **2020**, 31, 107489	10.6	43
208	An analysis of candidate autism loci on chromosome 2q24-q33: evidence for association to the STK39 gene. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1152-8	3.5	43
207	Lack of evidence for association of the serotonin transporter gene SLC6A4 with autism. *Biological Psychiatry*, **2006**, 60, 186-91	7.9	43
206	De novo SCN2A splice site mutation in a boy with Autism spectrum disorder. *BMC Medical Genetics*, **2014**, 15, 35	2.1	42
205	Pepsin pretreatment allows collagen IV immunostaining of blood vessels in adult mouse brain. *Journal of Neuroscience Methods*, **2007**, 163, 76-82	3	42
204	Autism-related routines and rituals associated with a mitochondrial aspartate/glutamate carrier SLC25A12 polymorphism. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147, 408-10	3.5	42
203	Familial symptom domains in monozygotic siblings with autism. *American Journal of Medical Genetics Part A*, **2004**, 129B, 76-81		42
202	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. *Annals of Neurology*, **2016**, 79, 739-747	9.4	42
201	Highly conserved molecular pathways, including Wnt signaling, promote functional recovery from spinal cord injury in lampreys. *Scientific Reports*, **2018**, 8, 742	4.9	41
200	Amyloid beta peptide formation in cell-free preparations. Regulation by protein kinase C, calmodulin, and calcineurin. *Journal of Biological Chemistry*, **1996**, 271, 24670-4	5.4	41
199	Evidence for Genetic Overlap Between Schizophrenia and Age at First Birth in Women. *JAMA Psychiatry*, **2016**, 73, 497-505	14.5	40
198	Neuropathology of the anterior midcingulate cortex in young children with autism. *Journal of Neuropathology and Experimental Neurology*, **2014**, 73, 891-902	3.1	40
197	Finding disease variants in Mendelian disorders by using sequence data: methods and applications. *American Journal of Human Genetics*, **2011**, 89, 701-12	11	40
196	AnnTools: a comprehensive and versatile annotation toolkit for genomic variants. *Bioinformatics*, **2012**, 28, 724-5	7.2	40
195	Presence of BACE1 and BACE2 in muscle fibres of patients with sporadic inclusion-body myositis. *Lancet, The*, **2001**, 358, 1962-4	40	40
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