Joseph D Buxbaum

List of Publications by Year in Descending Order

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	Psychometric properties of the Swedish translation of the Obsessive-Compulsive Inventory-Revised and the population characteristics of the symptom dimensions of OCD.. *Social Psychiatry and Psychiatric Epidemiology*, **2022**, 1	4.5	
370	A proof-of-concept study of growth hormone in children with Phelan-McDermid syndrome.. *Molecular Autism*, **2022**, 13, 6	6.5	2
369	Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders. *Biological Psychiatry*, **2022**, 91, 102-117	7.9	11
368	Mapping genomic loci implicates genes and synaptic biology in schizophrenia.. *Nature*, **2022**,	50.4	35
367	Clinical trial of insulin-like growth factor-1 in Phelan-McDermid syndrome.. *Molecular Autism*, **2022**, 13, 17	6.5	2
366	Manifestations of Alzheimer's disease genetic risk in the blood are evident in a multiomic analysis in healthy adults aged 18 to 90.. *Scientific Reports*, **2022**, 12, 6117	4.9	1
365	Neural Markers of Auditory Response and Habituation in Phelan-McDermid Syndrome.. *Frontiers in Neuroscience*, **2022**, 16, 815933	5.1	0
364	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 53	4.6	0
363	Powerful gene-based testing by integrating long-range chromatin interactions and knockoff genotypes. *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	1
362	The Genetic Architecture of Obsessive-Compulsive Disorder: Contribution of Liability to OCD From Alleles Across the Frequency Spectrum. *American Journal of Psychiatry*, **2021**, appiajp202121010101	11.9	2
361	How rare and common risk variation jointly affect liability for autism spectrum disorder. *Molecular Autism*, **2021**, 12, 66	6.5	2
360	Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. *Molecular Autism*, **2021**, 12, 65	6.5	4
359	Maternal health around pregnancy and autism risk: a diagnosis-wide, population-based study. *Psychological Medicine*, **2021**, 1-9	6.9	0
358	Shifted phase of EEG cross-frequency coupling in individuals with Phelan-McDermid syndrome. *Molecular Autism*, **2021**, 12, 29	6.5	1
357	FOXP1 syndrome: a review of the literature and practice parameters for medical assessment and monitoring. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 18	4.6	2
356	Information Avoidance and Information Seeking Among Parents of Children With ASD. *American Journal on Intellectual and Developmental Disabilities*, **2021**, 126, 249-259	2.2	2
355	Prospective and detailed behavioral phenotyping in DDX3X syndrome. *Molecular Autism*, **2021**, 12, 36	6.5	3

354	Reduced engagement of visual attention in children with autism spectrum disorder. *Autism*, **2021**, 25, 2064-2073	6.6	1
353	Developmental and Behavioral Phenotypes in a Mouse Model of DDX3X Syndrome. *Biological Psychiatry*, **2021**, 90, 742-755	7.9	2
352	Sensory Reactivity Phenotype in Phelan-McDermid Syndrome Is Distinct from Idiopathic ASD. *Genes* , **2021**, 12,	4.2	3
351	Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
350	Visual Evoked Potential Abnormalities in Phelan-McDermid Syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2021**,	7.2	1
349	Systematic review and meta-analysis: relationships between attention-deficit/hyperactivity disorder and urinary symptoms in children. *European Child and Adolescent Psychiatry*, **2021**, 1	5.5	3
348	Sensory Reactivity Symptoms Are a Core Feature of ADNP Syndrome Irrespective of Autism Diagnosis. *Genes*, **2021**, 12,	4.2	2
347	Clinical signs associated with earlier diagnosis of children with autism Spectrum disorder. *BMC Pediatrics*, **2021**, 21, 96	2.6	5
346	Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1712-1720	2.5	1
345	Reduced brain volume and white matter alterations in Shank3-deficient rats. *Autism Research*, **2021**, 14, 1837-1842	5.1	0
344	The promise of precision medicine in autism. *Neuron*, **2021**, 109, 2212-2215	13.9	1
343	Systematic review and meta-analysis identify significant relationships between clinical anxiety and lower urinary tract symptoms. *Brain and Behavior*, **2021**, 11, e2268	3.4	3
342	Strong evidence for genotype-phenotype correlations in Phelan-McDermid syndrome: Results from the developmental Synaptopathies consortium. *Human Molecular Genetics*, **2021**,	5.6	2
341	A randomized controlled trial of intranasal oxytocin in Phelan-McDermid syndrome. *Molecular Autism*, **2021**, 12, 62	6.5	0
340	Individuals with FOXP1 syndrome present with a complex neurobehavioral profile with high rates of ADHD, anxiety, repetitive behaviors, and sensory symptoms. *Molecular Autism*, **2021**, 12, 61	6.5	2
339	Social visual attentional engagement and memory in Phelan-McDermid syndrome and autism spectrum disorder: a pilot eye tracking study. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 58	4.6	0
338	Not All Autism Genes Are Created Equal: A Response to Myers et al. *American Journal of Human Genetics*, **2020**, 107, 1000-1003	11	5
337	Psychometric Study of the Social Responsiveness Scale in Phelan-McDermid Syndrome. *Autism Research*, **2020**, 13, 1383-1396	5.1	4

336	Co-localization between Sequence Constraint and Epigenomic Information Improves Interpretation of Whole-Genome Sequencing Data. *American Journal of Human Genetics*, **2020**, 106, 513-524	11	0
335	mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. *Nature Communications*, **2020**, 11, 2929	17.4	2
334	Functional annotation of rare structural variation in the human brain. *Nature Communications*, **2020**, 11, 2990	17.4	18
333	Transcriptional signatures of participant-derived neural progenitor cells and neurons implicate altered Wnt signaling in Phelan-McDermid syndrome and autism. *Molecular Autism*, **2020**, 11, 53	6.5	11
332	Gene constraint and genotype-phenotype correlations in neurodevelopmental disorders. *Current Opinion in Genetics and Development*, **2020**, 65, 69-75	4.9	3
331	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
330	Maternal Effects as Causes of Risk for Obsessive-Compulsive Disorder. *Biological Psychiatry*, **2020**, 87, 1045-1051	7.9	8
329	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. *Pediatric Neurology*, **2020**, 106, 24-31	2.9	4
328	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
327	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. *Cell Reports*, **2020**, 31, 107489	10.6	43
326	Cohort profile: Epidemiology and Genetics of Obsessive-compulsive disorder and chronic tic disorders in Sweden (EGOS). *Social Psychiatry and Psychiatric Epidemiology*, **2020**, 55, 1383-1393	4.5	8
325	Making Sense of Antisense: Getting From a Locus to a Gene. *Biological Psychiatry*, **2020**, 87, 95-97	7.9	
324	The Immersive Theater Experience for Individuals with Autism Spectrum Disorder. *Journal of Autism and Developmental Disorders*, **2020**, 50, 1073-1080	4.6	
323	Altered synaptic ultrastructure in the prefrontal cortex of Shank3-deficient rats. *Molecular Autism*, **2020**, 11, 89	6.5	5
322	Analysis of Genetically Regulated Gene Expression Identifies a Prefrontal PTSD Gene, SNRNP35, Specific to Military Cohorts. *Cell Reports*, **2020**, 31, 107716	10.6	21
321	Reduced axonal caliber and structural changes in a rat model of Fragile X syndrome with a deletion of a K-Homology domain of Fmr1. *Translational Psychiatry*, **2020**, 10, 280	8.6	2
320	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. *Journal of Medical Genetics*, **2020**,	5.8	2
319	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. *American Journal of Human Genetics*, **2020**, 107, 555-563	11	6

318	A framework for an evidence-based gene list relevant to autism spectrum disorder. *Nature Reviews Genetics*, **2020**, 21, 367-376	30.1	30
317	Global landscape and genetic regulation of RNA editing in cortical samples from individuals with schizophrenia. *Nature Neuroscience*, **2019**, 22, 1402-1412	25.5	32
316	Differential transcriptional response following glucocorticoid activation in cultured blood immune cells: a novel approach to PTSD biomarker development. *Translational Psychiatry*, **2019**, 9, 201	8.6	12
315	CommonMind Consortium provides transcriptomic and epigenomic data for Schizophrenia and Bipolar Disorder. *Scientific Data*, **2019**, 6, 180	8.2	52
314	Recessive gene disruptions in autism spectrum disorder. *Nature Genetics*, **2019**, 51, 1092-1098	36.3	56
313	Deletion of the KH1 Domain of Fmr1 Leads to Transcriptional Alterations and Attentional Deficits in Rats. *Cerebral Cortex*, **2019**, 29, 2228-2244	5.1	13
312	Recurrence Risk of Autism in Siblings and Cousins: A Multinational, Population-Based Study. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2019**, 58, 866-875	7.2	17
311	Variable DNA methylation in neonates mediates the association between prenatal smoking and birth weight. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2019**, 374, 20180120	5.8	28
310	Intestinal dysmotility in a zebrafish () mutant model of autism. *Molecular Autism*, **2019**, 10, 3	6.5	23
309	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746
308	Association of Genetic and Environmental Factors With Autism in a 5-Country Cohort. *JAMA Psychiatry*, **2019**, 76, 1035-1043	14.5	151
307	A genome-wide scan statistic framework for whole-genome sequence data analysis. *Nature Communications*, **2019**, 10, 3018	17.4	14
306	Parental Age and Differential Estimates of Risk for Neuropsychiatric Disorders: Findings From the Danish Birth Cohort. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2019**, 58, 618-627	7.2	15
305	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
304	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
303	Neuropsychiatric decompensation in adolescents and adults with Phelan-McDermid syndrome: a systematic review of the literature. *Molecular Autism*, **2019**, 10, 50	6.5	17
302	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. *Pediatric Neurology*, **2019**, 90, 37-43	2.9	14
301	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

300	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
299	Highly conserved molecular pathways, including Wnt signaling, promote functional recovery from spinal cord injury in lampreys. *Scientific Reports*, **2018**, 8, 742	4.9	41
298	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
297	Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by point mutations. *Molecular Autism*, **2018**, 9, 31	6.5	70
296	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
295	Heritable Variation, With Little or No Maternal Effect, Accounts for Recurrence Risk to Autism Spectrum Disorder in Sweden. *Biological Psychiatry*, **2018**, 83, 589-597	7.9	28
294	Shank3-deficient rats exhibit degraded cortical responses to sound. *Autism Research*, **2018**, 11, 59-68	5.1	13
293	Grandma knows best: Family structure and age of diagnosis of autism spectrum disorder. *Autism*, **2018**, 22, 368-376	6.6	15
292	PTSD Blood Transcriptome Mega-Analysis: Shared Inflammatory Pathways across Biological Sex and Modes of Trauma. *Neuropsychopharmacology*, **2018**, 43, 469-481	8.7	61
291	Calsenilin, a Presenilin Interactor, Regulates RhoA Signaling and Neurite Outgrowth. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	4
290	Elevated polygenic burden for autism is associated with differential DNA methylation at birth. *Genome Medicine*, **2018**, 10, 19	14.4	58
289	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. *Cell*, **2018**, 173, 1705-1715.e16	56.2	360
288	The Mount Sinai cohort of large-scale genomic, transcriptomic and proteomic data in Alzheimer's disease. *Scientific Data*, **2018**, 5, 180185	8.2	144
287	Disrupted circuits in mouse models of autism spectrum disorder and intellectual disability. *Current Opinion in Neurobiology*, **2018**, 48, 106-112	7.6	31
286	cGAS drives noncanonical-inflammasome activation in age-related macular degeneration. *Nature Medicine*, **2018**, 24, 50-61	50.5	134
285	Behavioral Phenotyping of an Improved Mouse Model of Phelan-McDermid Syndrome with a Complete Deletion of the Gene. *ENeuro*, **2018**, 5,	3.9	36
284	GJA1 (connexin43) is a key regulator of Alzheimer's disease pathogenesis. *Acta Neuropathologica Communications*, **2018**, 6, 144	7.3	37
283	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134

282	Temporal proteomic profiling of postnatal human cortical development. *Translational Psychiatry*, **2018**, 8, 267	8.6	10
281	Genetic risk for schizophrenia and autism, social impairment and developmental pathways to psychosis. *Translational Psychiatry*, **2018**, 8, 204	8.6	9
280	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
279	Association of Autism Spectrum Disorder With Prenatal Exposure to Medication Affecting Neurotransmitter Systems. *JAMA Psychiatry*, **2018**, 75, 1217-1224	14.5	18
278	Identification of rare de novo epigenetic variations in congenital disorders. *Nature Communications*, **2018**, 9, 2064	17.4	45
277	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
276	Gene expression in cord blood links genetic risk for neurodevelopmental disorders with maternal psychological distress and adverse childhood outcomes. *Brain, Behavior, and Immunity*, **2018**, 73, 320-330	16.6	16
275	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. *Alzheimers and Dementia*, **2017**, 13, 727-738	1.2	106
274	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
273	Autism spectrum disorder: neuropathology and animal models. *Acta Neuropathologica*, **2017**, 134, 537-566	14.3	191
272	Large-Scale Identification of Common Trait and Disease Variants Affecting Gene Expression. *American Journal of Human Genetics*, **2017**, 100, 885-894	11	48
271	A clinician-administered observation and corresponding caregiver interview capturing DSM-5 sensory reactivity symptoms in children with ASD. *Autism Research*, **2017**, 10, 1133-1140	5.1	30
270	Language ENvironment Analysis (LENA) in Phelan-McDermid Syndrome: Validity and Suggestions for Use in Minimally Verbal Children with Autism Spectrum Disorder. *Journal of Autism and Developmental Disorders*, **2017**, 47, 1605-1617	4.6	8
269	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. *Genome Medicine*, **2017**, 9, 114	14.4	48
268	Prospective investigation of FOXP1 syndrome. *Molecular Autism*, **2017**, 8, 57	6.5	35
267	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
266	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. *Nature Neuroscience*, **2017**, 20, 1217-1224	25.5	144
265	Two novel loci, COBL and SLC10A2, for Alzheimer's disease in African Americans. *Alzheimers and Dementia*, **2017**, 13, 119-129	1.2	48

264	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
263	Oxytocin improves behavioral and electrophysiological deficits in a novel Shank3-deficient rat. *ELife* , **2017**, 6,	8.9	75
262	Synaptic Interactome Mining Reveals p140Cap as a New Hub for PSD Proteins Involved in Psychiatric and Neurological Disorders. *Frontiers in Molecular Neuroscience*, **2017**, 10, 212	6.1	16
261	Global and local ancestry in African-Americans: Implications for Alzheimer's disease risk. *Alzheimer's and Dementia*, **2016**, 12, 233-43	1.2	27
260	Prenatal Maternal Smoking and Increased Risk for Tourette Syndrome and Chronic Tic Disorders. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2016**, 55, 784-91	7.2	22
259	The human-specific CASP4 gene product contributes to Alzheimer-related synaptic and behavioural deficits. *Human Molecular Genetics*, **2016**, 25, 4315-4327	5.6	10
258	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
257	Neural selectivity for communicative auditory signals in Phelan-McDermid syndrome. *Journal of Neurodevelopmental Disorders*, **2016**, 8, 5	4.6	13
256	Evidence for Genetic Overlap Between Schizophrenia and Age at First Birth in Women. *JAMA Psychiatry*, **2016**, 73, 497-505	14.5	40
255	A spectral approach integrating functional genomic annotations for coding and noncoding variants. *Nature Genetics*, **2016**, 48, 214-20	36.3	327
254	Measuring Sensory Reactivity in Autism Spectrum Disorder: Application and Simplification of a Clinician-Administered Sensory Observation Scale. *Journal of Autism and Developmental Disorders*, **2016**, 46, 287-293	4.6	35
253	Rapid and Objective Assessment of Neural Function in Autism Spectrum Disorder Using Transient Visual Evoked Potentials. *PLoS ONE*, **2016**, 11, e0164422	3.7	15
252	Cyfip1 Regulates Presynaptic Activity during Development. *Journal of Neuroscience*, **2016**, 36, 1564-76	6.6	38
251	Regulatory consequences of neuronal ELAV-like protein binding to coding and non-coding RNAs in human brain. *ELife*, **2016**, 5,	8.9	86
250	A Simplified Diagnostic Observational Assessment of Autism Spectrum Disorder in Early Childhood. *Autism Research*, **2016**, 9, 443-9	5.1	9
249	Altered tactile processing in children with autism spectrum disorder. *Autism Research*, **2016**, 9, 616-20	5.1	34
248	Assessment of the genetic variance of late-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 41, 200.e13-200.e20	5.6	119
247	Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nature Neuroscience*, **2016**, 19, 1442-1453	25.5	622

246	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
245	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. *Annals of Neurology*, **2016**, 79, 739-747	9.4	42
244	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
243	Genetics and genomics of autism spectrum disorder: embracing complexity. *Human Molecular Genetics*, **2015**, 24, R24-31	5.6	125
242	DSM-5 and psychiatric genetics - round hole, meet square peg. *Biological Psychiatry*, **2015**, 77, 766-8	7.9	8
241	Recent advances in the genetics of autism spectrum disorder. *Current Neurology and Neuroscience Reports*, **2015**, 15, 36	6.6	46
240	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
239	Phelan McDermid Syndrome: From Genetic Discoveries to Animal Models and Treatment. *Journal of Child Neurology*, **2015**, 30, 1861-70	2.5	37
238	Canonical Inflammasomes Drive IFN-□ to Prime Caspase-11 in Defense against a Cytosol-Invasive Bacterium. *Cell Host and Microbe*, **2015**, 18, 320-32	23.4	88
237	Association of Long Runs of Homozygosity With Alzheimer Disease Among African American Individuals. *JAMA Neurology*, **2015**, 72, 1313-23	17.2	27
236	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
235	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
234	Ultrastructural analyses in the hippocampus CA1 field in Shank3-deficient mice. *Molecular Autism*, **2015**, 6, 41	6.5	21
233	Understanding autism in the light of sex/gender. *Molecular Autism*, **2015**, 6, 24	6.5	72
232	Rarity of the Alzheimer disease-protective APP A673T variant in the United States. *JAMA Neurology* , **2015**, 72, 209-16	17.2	31
231	Familial clustering of tic disorders and obsessive-compulsive disorder. *JAMA Psychiatry*, **2015**, 72, 359-66	14.5	48
230	Autism-like Deficits in Shank3-Deficient Mice Are Rescued by Targeting Actin Regulators. *Cell Reports*, **2015**, 11, 1400-1413	10.6	170
229	Phenotypic and functional analysis of SHANK3 stop mutations identified in individuals with ASD and/or ID. *Molecular Autism*, **2015**, 6, 23	6.5	45

228	Randomized comparative trial of a social cognitive skills group for children with autism spectrum disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2015**, 54, 208-216.e1	7.2	34
227	PARK10 is a major locus for sporadic neuropathologically confirmed Parkinson disease. *Neurology*, **2015**, 84, 972-80	6.5	38
226	The autism mental status exam: sensitivity and specificity using DSM-5 criteria for autism spectrum disorder in verbally fluent adults. *Journal of Autism and Developmental Disorders*, **2014**, 44, 609-14	4.6	15
225	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
224	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
223	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
222	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
221	PLXNA4 is associated with Alzheimer disease and modulates tau phosphorylation. *Annals of Neurology*, **2014**, 76, 379-92	9.4	48
220	Most genetic risk for autism resides with common variation. *Nature Genetics*, **2014**, 46, 881-5	36.3	734
219	Neuropathology of the posteroinferior occipitotemporal gyrus in children with autism. *Molecular Autism*, **2014**, 5, 17	6.5	14
218	De novo SCN2A splice site mutation in a boy with Autism spectrum disorder. *BMC Medical Genetics*, **2014**, 15, 35	2.1	42
217	Two rare AKAP9 variants are associated with Alzheimer's disease in African Americans. *Alzheimers and Dementia*, **2014**, 10, 609-618.e11	1.2	83
216	Maturation of cortical circuits requires Semaphorin 7A. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 13978-83	11.5	23
215	Identification of rare causal variants in sequence-based studies: methods and applications to VPS13B, a gene involved in Cohen syndrome and autism. *PLoS Genetics*, **2014**, 10, e1004729	6	34
214	Mosaic epigenetic dysregulation of ectodermal cells in autism spectrum disorder. *PLoS Genetics*, **2014**, 10, e1004402	6	76
213	Genome-wide association meta-analysis of neuropathologic features of Alzheimer's disease and related dementias. *PLoS Genetics*, **2014**, 10, e1004606	6	219
212	Absence of strong strain effects in behavioral analyses of Shank3-deficient mice. *DMM Disease Models and Mechanisms*, **2014**, 7, 667-81	4.1	39
211	A critical role for human caspase-4 in endotoxin sensitivity. *Journal of Immunology*, **2014**, 193, 335-43	5.3	78

210	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
209	A pilot controlled trial of insulin-like growth factor-1 in children with Phelan-McDermid syndrome. *Molecular Autism*, **2014**, 5, 54	6.5	83
208	Neuropathology of the anterior midcingulate cortex in young children with autism. *Journal of Neuropathology and Experimental Neurology*, **2014**, 73, 891-902	3.1	40
207	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
206	Introduction of the human AVPR1A gene substantially alters brain receptor expression patterns and enhances aspects of social behavior in transgenic mice. *DMM Disease Models and Mechanisms*, **2014**, 7, 1013-22	4.1	13
205	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. *Molecular Autism*, **2014**, 5, 22	6.5	89
204	Transcriptomic changes in the frontal cortex associated with paternal age. *Molecular Autism*, **2014**, 5, 24	6.5	8
203	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. *Molecular Autism*, **2014**, 5, 34	6.5	25
202	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
201	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
200	Prospective investigation of autism and genotype-phenotype correlations in 22q13 deletion syndrome and SHANK3 deficiency. *Molecular Autism*, **2013**, 4, 18	6.5	170
199	SHANK3 haploinsufficiency: a "common" but underdiagnosed highly penetrant monogenic cause of autism spectrum disorders. *Molecular Autism*, **2013**, 4, 17	6.5	111
198	DSM-5: the debate continues. *Molecular Autism*, **2013**, 4, 11	6.5	11
197	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
196	Identification of COL6A2 mutations in progressive myoclonus epilepsy syndrome. *Human Genetics*, **2013**, 132, 275-83	6.3	17
195	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
194	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
193	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628

192	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
191	Next-Generation Sequencing For Gene and Pathway Discovery and Analysis in Autism Spectrum Disorders **2013**, 169-177		1
190	Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. *Genomics*, **2013**, 102, 270-7	4.3	11
189	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
188	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
187	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
186	Sequence kernel association tests for the combined effect of rare and common variants. *American Journal of Human Genetics*, **2013**, 92, 841-53	11	300
185	Sequencing of the sea lamprey (Petromyzon marinus) genome provides insights into vertebrate evolution. *Nature Genetics*, **2013**, 45, 415-21, 421e1-2	36.3	465
184	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. *Journal of Molecular Medicine*, **2013**, 91, 1399-406	5.5	38
183	Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes. *PLoS Genetics*, **2013**, 9, e1003671	6	168
182	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
181	Network topologies and convergent aetiologies arising from deletions and duplications observed in individuals with autism. *PLoS Genetics*, **2013**, 9, e1003523	6	47
180	Epigenetic Biomarkers as Predictors and Correlates of Symptom Improvement Following Psychotherapy in Combat Veterans with PTSD. *Frontiers in Psychiatry*, **2013**, 4, 118	5	218
179	SHANK2 and SHANK3 Mutations Implicate Glutamate Signaling Abnormalities in Autism Spectrum Disorders **2013**, 437-448		1
178	Capping four years of growth of Molecular Autism: impact factor coming in 2014. *Molecular Autism*, **2013**, 4, 50	6.5	
177	Copy number variations in alternative splicing gene networks impact lifespan. *PLoS ONE*, **2013**, 8, e53846	3.7	12
176	Characterization of SLITRK1 variation in obsessive-compulsive disorder. *PLoS ONE*, **2013**, 8, e70376	3.7	37
175	Getting from 1,000 Genes to a Triad of Symptoms **2013**, 461-471		

174	Optimizing the phenotyping of rodent ASD models: enrichment analysis of mouse and human neurobiological phenotypes associated with high-risk autism genes identifies morphological, electrophysiological, neurological, and behavioral features. *Molecular Autism*, **2012**, 3, 1	6.5	30
173	The effect of an autism-associated polymorphism in the STK39 gene on the autism symptom domains. *Journal of Autism and Developmental Disorders*, **2012**, 42, 319-20	4.6	1
172	Brief report: the Autism Mental Status Examination: development of a brief autism-focused exam. *Journal of Autism and Developmental Disorders*, **2012**, 42, 455-9	4.6	30
171	Exome sequencing identifies GCDH (glutaryl-CoA dehydrogenase) mutations as a cause of a progressive form of early-onset generalized dystonia. *Human Genetics*, **2012**, 131, 435-42	6.3	19
170	HDAC2 regulates atypical antipsychotic responses through the modulation of mGlu2 promoter activity. *Nature Neuroscience*, **2012**, 15, 1245-54	25.5	208
169	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron*, **2012**, 76, 1052-6	13.9	124
168	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
167	Evidence against a role for rare ADAM10 mutations in sporadic Alzheimer disease. *Neurobiology of Aging*, **2012**, 33, 416-417.e3	5.6	24
166	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. *Neurology*, **2012**, 79, 221-8	6.5	124
165	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
164	Haploinsufficiency of Cyfip1 produces fragile X-like phenotypes in mice. *PLoS ONE*, **2012**, 7, e42422	3.7	79
163	Complex autism spectrum disorder in a patient with a 17q12 microduplication. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1170-7	2.5	13
162	Network- and attribute-based classifiers can prioritize genes and pathways for autism spectrum disorders and intellectual disability. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2012**, 160C, 130-42	3.1	25
161	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
160	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
159	Advancing paternal age and simplex autism. *Autism*, **2012**, 16, 367-80	6.6	26
158	Reduced excitatory neurotransmission and mild autism-relevant phenotypes in adolescent Shank3 null mutant mice. *Journal of Neuroscience*, **2012**, 32, 6525-41	6.6	274
157	AnnTools: a comprehensive and versatile annotation toolkit for genomic variants. *Bioinformatics*, **2012**, 28, 724-5	7.2	40

156	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
155	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
154	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
153	Loss of function studies in mice and genetic association link receptor protein tyrosine phosphatase to schizophrenia. *Biological Psychiatry*, **2011**, 70, 626-35	7.9	17
152	Calsenilin is degraded by the ubiquitin-proteasome pathway. *Biochemical and Biophysical Research Communications*, **2011**, 405, 180-5	3.4	5
151	Human induced pluripotent stem cells: a new model for schizophrenia?. *Cell Stem Cell*, **2011**, 8, 461-2	18	3
150	Linking oligodendrocyte and myelin dysfunction to neurocircuitry abnormalities in schizophrenia. *Progress in Neurobiology*, **2011**, 93, 13-24	10.9	194
149	Genetic Markers for PTSD Risk and Resilience Among Survivors of the World Trade Center Attacks. *Disease Markers*, **2011**, 30, 101-110	3.2	109
148	Regeneration in the era of functional genomics and gene network analysis. *Biological Bulletin*, **2011**, 221, 18-34	1.5	19
147	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
146	Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. *European Journal of Human Genetics* , **2011**, 19, 1082-9	5.3	30
145	Analysis of a purported SHANK3 mutation in a boy with autism: clinical impact of rare variant research in neurodevelopmental disabilities. *Brain Research*, **2011**, 1380, 98-105	3.7	26
144	In vivo 1H-magnetic resonance spectroscopy study of the attentional networks in autism. *Brain Research*, **2011**, 1380, 198-205	3.7	81
143	No evidence for IL1RAPL1 involvement in selected high-risk autism pedigrees from the AGRE data set. *Autism Research*, **2011**, 4, 293-6	5.1	0
142	Calsenilin regulates presenilin 1/ secretase-mediated N-cadherin cleavage and catenin signaling. *FASEB Journal*, **2011**, 25, 4174-83	0.9	23
141	Whole-exome sequencing links a variant in DHDDS to retinitis pigmentosa. *American Journal of Human Genetics*, **2011**, 88, 201-6	11	130
140	Finding disease variants in Mendelian disorders by using sequence data: methods and applications. *American Journal of Human Genetics*, **2011**, 89, 701-12	11	40
139	Amyloid beta protein-induced zinc sequestration leads to synaptic loss via dysregulation of the ProSAP2/Shank3 scaffold. *Molecular Neurodegeneration*, **2011**, 6, 65	19	57

138	Haploinsufficiency of Gtf2i, a gene deleted in Williams Syndrome, leads to increases in social interactions. *Autism Research*, **2011**, 4, 28-39	5.1	79
137	A new testing strategy to identify rare variants with either risk or protective effect on disease. *PLoS Genetics*, **2011**, 7, e1001289	6	121
136	Genetic markers for PTSD risk and resilience among survivors of the World Trade Center attacks. *Disease Markers*, **2011**, 30, 101-10	3.2	54
135	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
134	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
133	APOE genotype results in differential effects on the peripheral clearance of amyloid-beta42 in APOE knock-in and knock-out mice. *Journal of Alzheimer's Disease*, **2010**, 21, 403-9	4.3	39
132	Profiling brain and plasma lipids in human APOE epsilon2, epsilon3, and epsilon4 knock-in mice using electrospray ionization mass spectrometry. *Journal of Alzheimer's Disease*, **2010**, 20, 105-11	4.3	25
131	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443
130	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
129	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
128	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
127	Slc25a12 disruption alters myelination and neurofilaments: a model for a hypomyelination syndrome and childhood neurodevelopmental disorders. *Biological Psychiatry*, **2010**, 67, 887-94	7.9	39
126	PCDH11X variation is not associated with late-onset Alzheimer disease susceptibility. *Psychiatric Genetics*, **2010**, 20, 321-4	2.9	15
125	PERIPHERAL MYELIN PROTEIN-22 IS EXPRESSED IN CNS MYELIN. *Translational Neuroscience*, **2010**, 1, 282-285	1.2	7
124	A large-scale survey of the novel 15q24 microdeletion syndrome in autism spectrum disorders identifies an atypical deletion that narrows the critical region. *Molecular Autism*, **2010**, 1, 5	6.5	38
123	A genome-wide study reveals copy number variants exclusive to childhood obesity cases. *American Journal of Human Genetics*, **2010**, 87, 661-6	11	78
122	Putative biological mechanisms for the association between early life adversity and the subsequent development of PTSD. *Psychopharmacology*, **2010**, 212, 405-17	4.7	149
121	Extensive proteomic screening identifies the obesity-related NYGGF4 protein as a novel LRP1-interactor, showing reduced expression in early Alzheimer's disease. *Molecular Neurodegeneration*, **2010**, 5, 1	19	28

120	Molecular Autism: accelerating and integrating research into neurodevelopmental conditions. *Molecular Autism*, **2010**, 1, 1	6.5	17
119	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
118	Genetics in psychiatry: common variant association studies. *Molecular Autism*, **2010**, 1, 6	6.5	17
117	FE65 binds Teashirt, inhibiting expression of the primate-specific caspase-4. *PLoS ONE*, **2009**, 4, e5071	3.7	27
116	Linking white and grey matter in schizophrenia: oligodendrocyte and neuron pathology in the prefrontal cortex. *Frontiers in Neuroanatomy*, **2009**, 3, 9	3.6	59
115	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
114	PGC-1alpha expression decreases in the Alzheimer disease brain as a function of dementia. *Archives of Neurology*, **2009**, 66, 352-61		250
113	Dietary composition modulates brain mass and solubilizable Abeta levels in a mouse model of aggressive Alzheimer's amyloid pathology. *Molecular Neurodegeneration*, **2009**, 4, 40	19	34
112	Novel cerebrovascular pathology in mice fed a high cholesterol diet. *Molecular Neurodegeneration*, **2009**, 4, 42	19	26
111	Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. *Nature*, **2009**, 459, 569-73	50.4	1075
110	Common genetic variants on 5p14.1 associate with autism spectrum disorders. *Nature*, **2009**, 459, 528-33	50.4	760
109	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
108	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
107	SLITRK1 binds 14-3-3 and regulates neurite outgrowth in a phosphorylation-dependent manner. *Biological Psychiatry*, **2009**, 66, 918-25	7.9	48
106	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
105	Multiple rare variants in the etiology of autism spectrum disorders. *Dialogues in Clinical Neuroscience*, **2009**, 11, 35-43	5.7	89
104	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
103	Increased locomotor activity in mice lacking the low-density lipoprotein receptor. *Behavioural Brain Research*, **2008**, 191, 256-65	3.4	29

102	A large-scale screen for coding variants in SERT/SLC6A4 in autism spectrum disorders. *Autism Research*, **2008**, 1, 251-7	5.1	16
101	Preclinical Animal Models of Autistic Spectrum Disorders (ASD) **2008**, 353-394		1
100	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
99	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
98	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
97	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
96	Pepsin pretreatment allows collagen IV immunostaining of blood vessels in adult mouse brain. *Journal of Neuroscience Methods*, **2007**, 163, 76-82	3	42
95	Mutation analysis of the NSD1 gene in patients with autism spectrum disorders and macrocephaly. *BMC Medical Genetics*, **2007**, 8, 68	2.1	18
94	Evidence against roles for phorbol binding protein Munc13-1, ADAM adaptor Eve-1, or vesicle trafficking phosphoproteins Munc18 or NSF as phospho-state-sensitive modulators of phorbol/PKC-activated Alzheimer APP ectodomain shedding. *Molecular Neurodegeneration*, **2007**, 2, 23	19	14
93	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
92	Elevated plasma cholesterol does not affect brain Abeta in mice lacking the low-density lipoprotein receptor. *Journal of Neurochemistry*, **2007**, 102, 1220-31	6	26
91	Transcriptional profiling of C57 and DBA strains of mice in the absence and presence of morphine. *BMC Genomics*, **2007**, 8, 76	4.5	31
90	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
89	A macromolecular complex involving the amyloid precursor protein (APP) and the cytosolic adapter FE65 is a negative regulator of axon branching. *Molecular and Cellular Neurosciences*, **2007**, 35, 57-63	4.8	35
88	Characterization of KIAA0513, a novel signaling molecule that interacts with modulators of neuroplasticity, apoptosis, and the cytoskeleton. *Brain Research*, **2006**, 1121, 1-11	3.7	18
87	Family-based association study of TPH1 and TPH2 polymorphisms in autism. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2006**, 141B, 861-7	3.5	25
86	Downstream regulatory element antagonistic modulator regulates islet prodynorphin expression. *American Journal of Physiology - Endocrinology and Metabolism*, **2006**, 291, E587-95	6	10
85	Receptor protein tyrosine phosphatase gamma is a marker for pyramidal cells and sensory neurons in the nervous system and is not necessary for normal development. *Molecular and Cellular Biology*, **2006**, 26, 5106-19	4.8	38

84	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
83	Lack of evidence for association of the serotonin transporter gene SLC6A4 with autism. *Biological Psychiatry*, **2006**, 60, 186-91	7.9	43
82	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
81	Autism and ultraconserved non-coding sequence on chromosome 7q. *Psychiatric Genetics*, **2006**, 16, 19-23	2.9	12
80	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
79	Increased Expression of Calsenilin in the Brains of Scrapie-infected Mice. *Annals of the New York Academy of Sciences*, **2006**, 928, 363-363	6.5	
78	Overexpression of Calsenilin in Sporadic Alzheimer's Disease Brain. *Annals of the New York Academy of Sciences*, **2006**, 928, 373-373	6.5	
77	Calsenilin interacts with transcriptional co-repressor C-terminal binding protein(s). *Journal of Neurochemistry*, **2006**, 98, 1290-301	6	18
76	The genetics of autism spectrum disorders. *NeuroMolecular Medicine*, **2006**, 8, 451-60	4.6	30
75	Fine mapping of the 5p13 locus linked to schizophrenia and schizotypal personality disorder in a Puerto Rican family. *Psychiatric Genetics*, **2005**, 15, 205-10	2.9	13
74	Expression of calsenilin in neurons and astrocytes in the Alzheimer's disease brain. *NeuroReport*, **2005**, 16, 451-5	1.7	21
73	Multiplexed variation scanning for 1,000 amplicons in hundreds of patients using mismatch repair detection (MRD) on tag arrays. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 14717-22	11.5	24
72	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
71	Allelic expression of APOE in human brain: effects of epsilon status and promoter haplotypes. *Human Molecular Genetics*, **2004**, 13, 2885-92	5.6	39
70	Linkage and association of the mitochondrial aspartate/glutamate carrier SLC25A12 gene with autism. *American Journal of Psychiatry*, **2004**, 161, 662-9	11.9	158
69	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
68	Neuregulin 1-erbB signaling and the molecular/cellular basis of schizophrenia. *Nature Neuroscience*, **2004**, 7, 575-80	25.5	326
67	Intracellular calcium modulates the nuclear translocation of calsenilin. *Journal of Neurochemistry*, **2004**, 89, 593-601	6	28

66	Familial symptom domains in monozygotic siblings with autism. *American Journal of Medical Genetics Part A*, **2004**, 129B, 76-81		42
65	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
64	A role for calsenilin and related proteins in multiple aspects of neuronal function. *Biochemical and Biophysical Research Communications*, **2004**, 322, 1140-4	3.4	37
63	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
62	Disease susceptibility genes for autism. *Annals of Medicine*, **2003**, 35, 274-81	1.5	30
61	White matter changes in schizophrenia: evidence for myelin-related dysfunction. *Archives of General Psychiatry*, **2003**, 60, 443-56		662
60	Molecular and cellular basis for anti-amyloid therapy in Alzheimer disease. *Alzheimer Disease and Associated Disorders*, **2003**, 17, 259-66	2.5	37
59	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
58	Neurobiologic Basis of Age-Related Dementing Disorders **2003**, 1095-1111		
57	Altered Abeta formation and long-term potentiation in a calsenilin knock-out. *Journal of Neuroscience*, **2003**, 23, 9097-106	6.6	96
56	BACE1 and BACE2 in pathologic and normal human muscle. *Experimental Neurology*, **2003**, 179, 150-8	5.7	35
55	Pharmacological concentrations of the HMG-CoA reductase inhibitor lovastatin decrease the formation of the Alzheimer beta-amyloid peptide in vitro and in patients. *Frontiers in Bioscience - Landmark*, **2002**, 7, a50-9	2.8	29
54	Symptom domains in autism and related conditions: evidence for familiality. *American Journal of Medical Genetics Part A*, **2002**, 114, 64-73		105
53	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
52	Molecular and cellular evidence for an oligodendrocyte abnormality in schizophrenia. *Neurochemical Research*, **2002**, 27, 1193-200	4.6	152
51	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
50	A role for APP in motility and transcription?. *Trends in Pharmacological Sciences*, **2002**, 23, 205-206	13.2	2
49	Calsenilin-Presenilin Interaction in Alzheimer's Disease. *Advances in Behavioral Biology*, **2002**, 105-110		

48	Correlation between Abetax-40-, Abetax-42-, and Abetax-43-containing amyloid plaques and cognitive decline. *Archives of Neurology*, **2001**, 58, 2025-32		90
47	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
46	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
45	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
44	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
43	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
42	Subcellular localization of presenilin 2 endoproteolytic C-terminal fragments. *Molecular Brain Research*, **2001**, 96, 14-20		10
41	Presence of BACE1 and BACE2 in muscle fibres of patients with sporadic inclusion-body myositis. *Lancet, The*, **2001**, 358, 1962-4	40	40
40	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
39	Intraneuronal Abeta42 accumulation in human brain. *American Journal of Pathology*, **2000**, 156, 15-20	5.8	827
38	Genomic structure, expression pattern, and chromosomal localization of the human calsenilin gene: no association between an exonic polymorphism and Alzheimer's disease. *Neuroscience Letters*, **2000**, 294, 135-8	3.3	4
37	Tumor necrosis factor-alpha-converting enzyme is required for cleavage of erbB4/HER4. *Journal of Biological Chemistry*, **2000**, 275, 10379-87	5.4	244
36	Regulation of beta-amyloid secretion by FE65, an amyloid protein precursor-binding protein. *Journal of Biological Chemistry*, **1999**, 274, 7952-7	5.4	170
35	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
34	Estrogen reduces neuronal generation of Alzheimer beta-amyloid peptides. *Nature Medicine*, **1998**, 4, 447-51	50.5	479
33	Regulation of secretion of Alzheimer amyloid precursor protein by the mitogen-activated protein kinase cascade. *Journal of Neurochemistry*, **1998**, 70, 524-30	6	86
32	Generation and regulation of beta-amyloid peptide variants by neurons. *Journal of Neurochemistry*, **1998**, 71, 1920-5	6	97
31	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

30	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
29	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
28	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
27	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
26	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
25	APP processing, A beta-amyloidogenesis, and the pathogenesis of Alzheimer's disease. *Neurobiology of Aging*, **1994**, 15, 253-6	5.6	37
24	Protein phosphorylation regulates relative utilization of processing pathways for Alzheimer beta/A4 amyloid precursor protein. *Annals of the New York Academy of Sciences*, **1993**, 695, 117-21	6.5	26
23	Characterization of alternative routes for processing of the Alzheimer beta/A4-amyloid precursor protein. Differential effects of phorbol esters and chloroquine. *Annals of the New York Academy of Sciences*, **1992**, 674, 203-17	6.5	14
22	The nature and metabolism of potentially amyloidogenic carboxyl-terminal fragments of the Alzheimer beta/A4-amyloid precursor protein: some technical notes. *Neurobiology of Aging*, **1992**, 13, 601-3	5.6	10
21	In vivo protein phosphorylation in Drosophila mutants defective in learning and memory. *Neuroscience Letters*, **1989**, 104, 351-5	3.3	4
20	A microtiter-based assay for protein kinase activity suitable for the analysis of large numbers of samples, and its application to the study of Drosophila learning mutants. *Analytical Biochemistry*, **1988**, 169, 209-15	3.1	8
19	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
18	In vitro protein phosphorylation in head preparations from normal and mutant Drosophila melanogaster. *Journal of Neurochemistry*, **1987**, 49, 1161-73	6	7
17	Rare schizophrenia risk variant burden is conserved in diverse human populations		1
16	Rare coding variation illuminates the allelic architecture, risk genes, cellular expression patterns, and phenotypic context of autism		4
15	How rare and common risk variation jointly affect liability for autism spectrum disorder		1
14	Unperturbed Expression Bias of Imprinted Genes in Schizophrenia		1
13	Gene Expression Elucidates Functional Impact of Polygenic Risk for Schizophrenia		3

12	Refining the role of de novo protein truncating variants in neurodevelopmental disorders using population reference samples	5
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10	New mutations, old statistical challenges	1
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