Daniah M Trabzuni

List of Publications by Year
in descending order

70 papers	17,654 citations	66234 42 h-index	88477 70 g-index
76 all docs	76 docs citations	76 times ranked	26199 citing authors

#	Article	IF	Citations
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	3.8	3,833
2	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	13.9	2,385
3	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	4.9	1,414
4	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	13.7	772
5	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	1.1	696
6	Genetic variability in the regulation of gene expression in ten regions of the human brain. Nature Neuroscience, 2014, 17, 1418-1428.	7.1	620
7	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	9.4	594
8	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	13.7	425
9	Unbiased screen for interactors of leucine-rich repeat kinase 2 supports a common pathway for sporadic and familial Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 2626-2631.	3.3	342
10	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	9.4	338
11	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	2.6	333
12	Major Shifts in Glial Regional Identity Are a Transcriptional Hallmark of Human Brain Aging. Cell Reports, 2017, 18, 557-570.	2.9	326
13	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
14	Genome-wide association study of obsessive-compulsive disorder. Molecular Psychiatry, 2013, 18, 788-798.	4.1	312
15	Widespread sex differences in gene expression and splicing in the adult human brain. Nature Communications, 2013, 4, 2771.	5.8	255
16	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	5.8	250
17	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	1.5	247
18	The Friedreich ataxia GAA repeat expansion mutation induces comparable epigenetic changes in human and transgenic mouse brain and heart tissues. Human Molecular Genetics, 2007, 17, 735-746.	1.4	229

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19	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	2.6	224
20	Quality control parameters on a large dataset of regionally dissected human control brains for whole genome expression studies. Journal of Neurochemistry, 2011, 119, 275-282.	2.1	214
21	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	7.1	213
22	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	9.4	212
23	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. Human Molecular Genetics, 2012, 21, 4094-4103.	1.4	191
24	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
25	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	3.7	168
26	Genome-wide association study of Tourette's syndrome. Molecular Psychiatry, 2013, 18, 721-728.	4.1	161
27	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 657-665.	2.6	151
28	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	2.8	148
29	Insights into TREM2 biology by network analysis of human brain gene expression data. Neurobiology of Aging, 2013, 34, 2699-2714.	1.5	145
30	Recursive splicing in long vertebrate genes. Nature, 2015, 521, 371-375.	13.7	128
31	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
32	Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. Neurobiology of Disease, 2012, 47, 20-28.	2.1	121
33	Serum Iron Levels and the Risk of Parkinson Disease: A Mendelian Randomization Study. PLoS Medicine, 2013, 10, e1001462.	3.9	116
34	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
35	The T/G 13915 variant upstream of the lactase gene (LCT) is the founder allele of lactase persistence in an urban Saudi population. Journal of Medical Genetics, 2007, 44, e89-e89.	1.5	100
36	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	4.5	95

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37	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	2.2	66
38	Age-associated changes in gene expression in human brain and isolated neurons. Neurobiology of Aging, 2013, 34, 1199-1209.	1.5	65
39	Loss of GPR3 reduces the amyloid plaque burden and improves memory in Alzheimerâ€™s disease mouse models. Science Translational Medicine, 2015, 7, 309ra164.	5.8	61
40	Increased brain expression of GPNMB is associated with genome wide significant risk for Parkinsonâ€™s disease on chromosome 7p15.3. Neurogenetics, 2017, 18, 121-133.	0.7	57
41	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	2.2	57
42	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787.	3.7	50
43	Investigating the utility of human embryonic stem cellâ€derived neurons to model ageing and neurodegenerative disease using wholeâ€genome gene expression and splicing analysis. Journal of Neurochemistry, 2012, 122, 738-751.	2.1	48
44	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	2.2	47
45	Transcriptomic and genetic analyses reveal potential causal drivers for intractable partial epilepsy. Brain, 2019, 142, 1616-1630.	3.7	47
46	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	1.1	45
47	Genetic evidence for a pathogenic role for the vitamin D3 metabolizing enzyme CYP24A1 in multiple sclerosis. Multiple Sclerosis and Related Disorders, 2014, 3, 211-219.	0.9	44
48	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 666-673.	0.9	43
49	Genomewide association study in cervical dystonia demonstrates possible association with sodium leak channel. Movement Disorders, 2014, 29, 245-251.	2.2	43
50	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. Human Mutation, 2012, 33, 1708-1718.	1.1	42
51	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.3	41
52	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. Nucleic Acids Research, 2013, 41, e88-e88.	6.5	39
53	Activation of Apoptotic Caspase Cascade During the Transition to Pressure Overload-Induced Heart Failure. Journal of the American College of Cardiology, 2006, 48, 1451-1458.	1.2	36
54	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 486-492.	0.9	35

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55	Assessment of common variability and expression quantitative trait loci for genome-wide associations for progressive supranuclear palsy. Neurobiology of Aging, 2014, 35, 1514.e1-1514.e12.	1.5	33
56	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	3.7	33
57	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. Nature, 2021, 594, 117-123.	13.7	29
58	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	2.8	29
59	Pathogenic LRRK2 Mutations Do Not Alter Gene Expression in Cell Model Systems or Human Brain Tissue. PLoS ONE, 2011, 6, e22489.	1.1	27
60	Upregulation of Bcl-2 proteins during the transition to pressure overload-induced heart failure. International Journal of Cardiology, 2007, 116, 27-33.	0.8	25
61	A comprehensive introduction to the genetic basis of non-syndromic hearing loss in the Saudi Arabian population. BMC Medical Genetics, 2011, 12, 91.	2.1	23
62	Analysis of gene expression data using a linear mixed model/finite mixture model approach: application to regional differences in the human brain. Bioinformatics, 2014, 30, 1555-1561.	1.8	22
63	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	5.8	22
64	Genome-wide human brain eQTLs: In-depth analysis and insights using the UKBEC dataset. Scientific Reports, 2019, 9, 19201.	1.6	15
65	Whole genome expression as a quantitative trait. Biochemical Society Transactions, 2009, 37, 1276-1277.	1.6	9
66	Genotypic analysis of gene expression in the dissection of the aetiology of complex neurological and psychiatric diseases. Briefings in Functional Genomics & Proteomics, 2009, 8, 194-198.	3.8	4
67	Quality control parameters on a large dataset of regionally dissected human control brains for whole genome expression studies. Journal of Neurochemistry, 2012, 120, 473-473.	2.1	4
68	Insights into the Influence of Specific Splicing Events on the Structural Organization of LRRK2. International Journal of Molecular Sciences, 2018, 19, 2784.	1.8	2
69	693. Identification, Regulation and Characterisation of Transcribed Intergenic Regions in Human Substantia Nigra and Putamen. Biological Psychiatry, 2017, 81, S281.	0.7	0
70	Comparison Between Expression Microarrays and RNA-Sequencing Using UKBEC Dataset Identified a -eQTL Associated with Gene in Substantia Nigra. , 2020, 1, 100001.		0