Daniah M Trabzuni

List of Publications by Year
in descending order

70 papers	17,654 citations	66343 42 h-index	88630 70 g-index
76 all docs	76 docs citations	76 times ranked	26199 citing authors

#	Article	IF	Citations
1	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
2	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
3	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. Nature, 2021, 594, 117-123.	27.8	29
4	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
5	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787.	7.6	50
6	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	12.8	22
7	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	3.9	57
8	Comparison Between Expression Microarrays and RNA-Sequencing Using UKBEC Dataset Identified a -eQTL Associated with Gene in Substantia Nigra. , 2020, 1, 100001.		0
9	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.	10.2	1,414
10	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.	3.9	47
11	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	3.9	66
12	Transcriptomic and genetic analyses reveal potential causal drivers for intractable partial epilepsy. Brain, 2019, 142, 1616-1630.	7.6	47
13	Genome-wide human brain eQTLs: In-depth analysis and insights using the UKBEC dataset. Scientific Reports, 2019, 9, 19201.	3.3	15
14	Insights into the Influence of Specific Splicing Events on the Structural Organization of LRRK2. International Journal of Molecular Sciences, 2018, 19, 2784.	4.1	2
15	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
16	Major Shifts in Glial Regional Identity Are a Transcriptional Hallmark of Human Brain Aging. Cell Reports, 2017, 18, 557-570.	6.4	326
17	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.	1.4	57
18	693. Identification, Regulation and Characterisation of Transcribed Intergenic Regions in Human Substantia Nigra and Putamen. Biological Psychiatry, 2017, 81, S281.	1.3	0

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19	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
20	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
21	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.	6.2	333
22	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
23	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
24	Recursive splicing in long vertebrate genes. Nature, 2015, 521, 371-375.	27.8	128
25	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 657-665.	6.2	151
26	Loss of GPR3 reduces the amyloid plaque burden and improves memory in Alzheimerâ€™s disease mouse models. Science Translational Medicine, 2015, 7, 309ra164.	12.4	61
27	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.	7.1	342
28	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 486-492.	1.9	35
29	Genomewide association study in cervical dystonia demonstrates possible association with sodium leak channel. Movement Disorders, 2014, 29, 245-251.	3.9	43
30	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.	4.1	22
31	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
32	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. Brain Imaging and Behavior, 2014, 8, 153-182.	2.1	696
33	Genetic variability in the regulation of gene expression in ten regions of the human brain. Nature Neuroscience, 2014, 17, 1418-1428.	14.8	620
34	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.	3.1	33
35	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.	2.0	44
36	Genome-wide association study of Tourette's syndrome. Molecular Psychiatry, 2013, 18, 721-728.	7.9	161

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37	Insights into TREM2 biology by network analysis of human brain gene expression data. Neurobiology of Aging, 2013, 34, 2699-2714.	3.1	145
38	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	27.0	2,385
39	Age-associated changes in gene expression in human brain and isolated neurons. Neurobiology of Aging, 2013, 34, 1199-1209.	3.1	65
40	Genome-wide association study of obsessive-compulsive disorder. Molecular Psychiatry, 2013, 18, 788-798.	7.9	312
41	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	21.4	338
42	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
43	Widespread sex differences in gene expression and splicing in the adult human brain. Nature Communications, 2013, 4, 2771.	12.8	255
44	Serum Iron Levels and the Risk of Parkinson Disease: A Mendelian Randomization Study. PLoS Medicine, 2013, 10, e1001462.	8.4	116
45	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	2.9	122
46	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. Nucleic Acids Research, 2013, 41, e88-e88.	14.5	39
47	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.8	41
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.	1.9	43
49	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	5.3	148
50	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	2.5	45
51	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176
52	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	21.4	212
53	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. Human Mutation, 2012, 33, 1708-1718.	2.5	42
54	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594

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55	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. Human Molecular Genetics, 2012, 21, 4094-4103.	2.9	191
56	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.	3.9	48
57	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	6.2	224
58	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.	3.9	4
59	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.	4.4	121
60	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
61	Pathogenic LRRK2 Mutations Do Not Alter Gene Expression in Cell Model Systems or Human Brain Tissue. PLoS ONE, 2011, 6, e22489.	2.5	27
62	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.	3.9	214
63	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
64	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	3.5	247
65	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
66	Whole genome expression as a quantitative trait. Biochemical Society Transactions, 2009, 37, 1276-1277.	3.4	9
67	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.	2.9	229
68	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.	3.2	100
69	Upregulation of Bcl-2 proteins during the transition to pressure overload-induced heart failure. International Journal of Cardiology, 2007, 116, 27-33.	1.7	25
70	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.	2.8	36