

# Daniah M Trabzuni

## List of Publications by Year in descending order

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Version: 2024-02-01

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	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	8.1	3,833
2	<i>TREM2</i> Variants in Alzheimer's Disease. <i>New England Journal of Medicine</i> , 2013, 368, 117-127.	27.0	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. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	10.2	1,414
4	Common genetic variants influence human subcortical brain structures. <i>Nature</i> , 2015, 520, 224-229.	27.8	772
5	The ENIGMA Consortium: large-scale collaborative analyses of neuroimaging and genetic data. <i>Brain Imaging and Behavior</i> , 2014, 8, 153-182.	2.1	696
6	Genetic variability in the regulation of gene expression in ten regions of the human brain. <i>Nature Neuroscience</i> , 2014, 17, 1418-1428.	14.8	620
7	Identification of common variants associated with human hippocampal and intracranial volumes. <i>Nature Genetics</i> , 2012, 44, 552-561.	21.4	594
8	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014, 505, 550-554.	27.8	425
9	Unbiased screen for interactors of leucine-rich repeat kinase 2 supports a common pathway for sporadic and familial Parkinson disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 2626-2631.	7.1	342
10	Genome-wide meta-analysis identifies new susceptibility loci for migraine. <i>Nature Genetics</i> , 2013, 45, 912-917.	21.4	338
11	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	6.2	333
12	Major Shifts in Glial Regional Identity Are a Transcriptional Hallmark of Human Brain Aging. <i>Cell Reports</i> , 2017, 18, 557-570.	6.4	326
13	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	7.6	323
14	Genome-wide association study of obsessive-compulsive disorder. <i>Molecular Psychiatry</i> , 2013, 18, 788-798.	7.9	312
15	Widespread sex differences in gene expression and splicing in the adult human brain. <i>Nature Communications</i> , 2013, 4, 2771.	12.8	255
16	Novel genetic loci associated with hippocampal volume. <i>Nature Communications</i> , 2017, 8, 13624.	12.8	250
17	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. <i>PLoS Genetics</i> , 2011, 7, e1002142.	3.5	247
18	The Friedreich ataxia GAA repeat expansion mutation induces comparable epigenetic changes in human and transgenic mouse brain and heart tissues. <i>Human Molecular Genetics</i> , 2007, 17, 735-746.	2.9	229

#	ARTICLE	IF	CITATIONS
19	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. <i>American Journal of Human Genetics</i> , 2012, 91, 1041-1050.	6.2	224
20	Quality control parameters on a large dataset of regionally dissected human control brains for whole genome expression studies. <i>Journal of Neurochemistry</i> , 2011, 119, 275-282.	3.9	214
21	Novel genetic loci underlying human intracranial volume identified through genome-wide association. <i>Nature Neuroscience</i> , 2016, 19, 1569-1582.	14.8	213
22	Common variants at 12q14 and 12q24 are associated with hippocampal volume. <i>Nature Genetics</i> , 2012, 44, 545-551.	21.4	212
23	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. <i>Human Molecular Genetics</i> , 2012, 21, 4094-4103.	2.9	191
24	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012, 21, 4996-5009.	2.9	176
25	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. <i>Brain</i> , 2013, 136, 3140-3150.	7.6	168
26	Genome-wide association study of Tourette's syndrome. <i>Molecular Psychiatry</i> , 2013, 18, 721-728.	7.9	161
27	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 657-665.	6.2	151
28	Mutations in the autoregulatory domain of $\beta$ -tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , 2013, 73, 546-553.	5.3	148
29	Insights into TREM2 biology by network analysis of human brain gene expression data. <i>Neurobiology of Aging</i> , 2013, 34, 2699-2714.	3.1	145
30	Recursive splicing in long vertebrate genes. <i>Nature</i> , 2015, 521, 371-375.	27.8	128
31	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , 2013, 22, 1039-1049.	2.9	122
32	Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. <i>Neurobiology of Disease</i> , 2012, 47, 20-28.	4.4	121
33	Serum Iron Levels and the Risk of Parkinson Disease: A Mendelian Randomization Study. <i>PLoS Medicine</i> , 2013, 10, e1001462.	8.4	116
34	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 938-947.	6.2	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. <i>Journal of Medical Genetics</i> , 2007, 44, e89-e89.	3.2	100
36	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	9.0	95

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37	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	3.9	66
38	Age-associated changes in gene expression in human brain and isolated neurons. <i>Neurobiology of Aging</i> , 2013, 34, 1199-1209.	3.1	65
39	Loss of GPR3 reduces the amyloid plaque burden and improves memory in Alzheimer's disease mouse models. <i>Science Translational Medicine</i> , 2015, 7, 309ra164.	12.4	61
40	Increased brain expression of GPNMB is associated with genome wide significant risk for Parkinson's disease on chromosome 7p15.3. <i>Neurogenetics</i> , 2017, 18, 121-133.	1.4	57
41	Penetrance of Parkinson's Disease in <i>LRRK2</i> p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020, 35, 774-780.	3.9	57
42	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. <i>Brain</i> , 2020, 143, 2771-2787.	7.6	50
43	Investigating the utility of human embryonic stem cell-derived neurons to model ageing and neurodegenerative disease using whole-genome gene expression and splicing analysis. <i>Journal of Neurochemistry</i> , 2012, 122, 738-751.	3.9	48
44	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	3.9	47
45	Transcriptomic and genetic analyses reveal potential causal drivers for intractable partial epilepsy. <i>Brain</i> , 2019, 142, 1616-1630.	7.6	47
46	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. <i>PLoS ONE</i> , 2013, 8, e70724.	2.5	45
47	Genetic evidence for a pathogenic role for the vitamin D3 metabolizing enzyme CYP24A1 in multiple sclerosis. <i>Multiple Sclerosis and Related Disorders</i> , 2014, 3, 211-219.	2.0	44
48	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 666-673.	1.9	43
49	Genomewide association study in cervical dystonia demonstrates possible association with sodium leak channel. <i>Movement Disorders</i> , 2014, 29, 245-251.	3.9	43
50	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. <i>Human Mutation</i> , 2012, 33, 1708-1718.	2.5	42
51	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. <i>Annals of Human Genetics</i> , 2013, 77, 85-105.	0.8	41
52	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. <i>Nucleic Acids Research</i> , 2013, 41, e88-e88.	14.5	39
53	Activation of Apoptotic Caspase Cascade During the Transition to Pressure Overload-Induced Heart Failure. <i>Journal of the American College of Cardiology</i> , 2006, 48, 1451-1458.	2.8	36
54	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 486-492.	1.9	35

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55	Assessment of common variability and expression quantitative trait loci for genome-wide associations for progressive supranuclear palsy. <i>Neurobiology of Aging</i> , 2014, 35, 1514.e1-1514.e12.	3.1	33
56	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. <i>Brain</i> , 2021, 144, 769-780.	7.6	33
57	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. <i>Nature</i> , 2021, 594, 117-123.	27.8	29
58	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021, 90, 35-42.	5.3	29
59	Pathogenic LRRK2 Mutations Do Not Alter Gene Expression in Cell Model Systems or Human Brain Tissue. <i>PLoS ONE</i> , 2011, 6, e22489.	2.5	27
60	Upregulation of Bcl-2 proteins during the transition to pressure overload-induced heart failure. <i>International Journal of Cardiology</i> , 2007, 116, 27-33.	1.7	25
61	A comprehensive introduction to the genetic basis of non-syndromic hearing loss in the Saudi Arabian population. <i>BMC Medical Genetics</i> , 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. <i>Bioinformatics</i> , 2014, 30, 1555-1561.	4.1	22
63	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. <i>Nature Communications</i> , 2020, 11, 1041.	12.8	22
64	Genome-wide human brain eQTLs: In-depth analysis and insights using the UKBEC dataset. <i>Scientific Reports</i> , 2019, 9, 19201.	3.3	15
65	Whole genome expression as a quantitative trait. <i>Biochemical Society Transactions</i> , 2009, 37, 1276-1277.	3.4	9
66	Genotypic analysis of gene expression in the dissection of the aetiology of complex neurological and psychiatric diseases. <i>Briefings in Functional Genomics &amp; Proteomics</i> , 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. <i>Journal of Neurochemistry</i> , 2012, 120, 473-473.	3.9	4
68	Insights into the Influence of Specific Splicing Events on the Structural Organization of LRRK2. <i>International Journal of Molecular Sciences</i> , 2018, 19, 2784.	4.1	2
69	693. Identification, Regulation and Characterisation of Transcribed Intergenic Regions in Human Substantia Nigra and Putamen. <i>Biological Psychiatry</i> , 2017, 81, S281.	1.3	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