Douglas M Ruderfer

List of Publications by Citations

145 papers	48,707 citations	71 h-index	162 g-index
162 ext. papers	60,189 ext. citations	17.9 avg, IF	8.31 L-index

#	Paper	IF	Citations
145	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
144	Biological insights from 108 schizophrenia-associated genetic loci. *Nature*, **2014**, 511, 421-7	50.4	5249
143	Common polygenic variation contributes to risk of schizophrenia and bipolar disorder. *Nature*, **2009**, 460, 748-52	50.4	3568
142	Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis. *Lancet, The*, **2013**, 381, 1371-1379	40	2112
141	LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. *Nature Genetics*, **2015**, 47, 291-5	36.3	2096
140	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
139	Genome-wide association study identifies five new schizophrenia loci. *Nature Genetics*, **2011**, 43, 969-76	36.3	1508
138	Schizophrenia risk from complex variation of complement component 4. *Nature*, **2016**, 530, 177-83	50.4	1352
137	Rare chromosomal deletions and duplications increase risk of schizophrenia. *Nature*, **2008**, 455, 237-41	50.4	1251
136	Association between microdeletion and microduplication at 16p11.2 and autism. *New England Journal of Medicine*, **2008**, 358, 667-75	59.2	1249
135	De novo mutations in schizophrenia implicate synaptic networks. *Nature*, **2014**, 506, 179-84	50.4	1163
134	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nature Genetics*, **2013**, 45, 1150-9	36.3	1153
133	Large-scale genome-wide association analysis of bipolar disorder identifies a new susceptibility locus near ODZ4. *Nature Genetics*, **2011**, 43, 977-83	36.3	1094
132	A polygenic burden of rare disruptive mutations in schizophrenia. *Nature*, **2014**, 506, 185-90	50.4	1059
131	Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. *Nature Genetics*, **2008**, 40, 1056-8	36.3	949
130	Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. *Nature Genetics*, **2018**, 50, 381-389	36.3	787
129	Genetic basis of proteome variation in yeast. *Nature Genetics*, **2007**, 39, 1369-75	36.3	722

128	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
127	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. *American Journal of Human Genetics*, **2015**, 97, 576-92	11	649
126	Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nature Neuroscience*, **2016**, 19, 1442-1453	25.5	622
125	De novo CNV analysis implicates specific abnormalities of postsynaptic signalling complexes in the pathogenesis of schizophrenia. *Molecular Psychiatry*, **2012**, 17, 142-53	15.1	611
124	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. *Nature Neuroscience*, **2015**, 18, 199-209	25.5	572
123	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
122	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
121	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
120	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	8.9	414
119	Partitioning heritability of regulatory and cell-type-specific variants across 11 common diseases. *American Journal of Human Genetics*, **2014**, 95, 535-52	11	411
118	Discovery and statistical genotyping of copy-number variation from whole-exome sequencing depth. *American Journal of Human Genetics*, **2012**, 91, 597-607	11	391
117	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. *Cell*, **2018**, 173, 1705-1715.e16	56.2	360
116	Comprehensive polymorphism survey elucidates population structure of Saccharomyces cerevisiae. *Nature*, **2009**, 458, 342-5	50.4	355
115	The ExAC browser: displaying reference data information from over 60 000 exomes. *Nucleic Acids Research*, **2017**, 45, D840-D845	20.1	348
114	Contrasting genetic architectures of schizophrenia and other complex diseases using fast variance-components analysis. *Nature Genetics*, **2015**, 47, 1385-92	36.3	299
113	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
112	Improved detection of common variants associated with schizophrenia by leveraging pleiotropy with cardiovascular-disease risk factors. *American Journal of Human Genetics*, **2013**, 92, 197-209	11	293
111	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. *Nature Genetics*, **2015**, 47, 1415-25	36.3	292

110	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. *Nature Neuroscience*, **2016**, 19, 1433-1441	25.5	291
109	Polygenic dissection of diagnosis and clinical dimensions of bipolar disorder and schizophrenia. *Molecular Psychiatry*, **2014**, 19, 1017-1024	15.1	258
108	Genome-wide detection of polymorphisms at nucleotide resolution with a single DNA microarray. *Science*, **2006**, 311, 1932-6	33.3	229
107	All SNPs are not created equal: genome-wide association studies reveal a consistent pattern of enrichment among functionally annotated SNPs. *PLoS Genetics*, **2013**, 9, e1003449	6	209
106	Quantifying prion disease penetrance using large population control cohorts. *Science Translational Medicine*, **2016**, 8, 322ra9	17.5	205
105	Genome-wide association study in a Swedish population yields support for greater CNV and MHC involvement in schizophrenia compared with bipolar disorder. *Molecular Psychiatry*, **2012**, 17, 880-6	15.1	196
104	Copy number variation in schizophrenia in Sweden. *Molecular Psychiatry*, **2014**, 19, 762-73	15.1	191
103	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
102	Molecular pathways involved in neuronal cell adhesion and membrane scaffolding contribute to schizophrenia and bipolar disorder susceptibility. *Molecular Psychiatry*, **2011**, 16, 286-92	15.1	175
101	A role for noncoding variation in schizophrenia. *Cell Reports*, **2014**, 9, 1417-29	10.6	174
100	Population genomic analysis of outcrossing and recombination in yeast. *Nature Genetics*, **2006**, 38, 1077-81	33.3	173
99	Identification of pathways for bipolar disorder: a meta-analysis. *JAMA Psychiatry*, **2014**, 71, 657-64	14.5	172
98	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
97	Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. *American Journal of Human Genetics*, **2014**, 94, 233-45	11	170
96	Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. *American Journal of Human Genetics*, **2011**, 89, 551-63	11	166
95	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. *Nature Neuroscience*, **2016**, 19, 420-431	25.5	163
94	Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. *American Journal of Human Genetics*, **2015**, 96, 283-94	11	161
93	Telomere length as a quantitative trait: genome-wide survey and genetic mapping of telomere length-control genes in yeast. *PLoS Genetics*, **2006**, 2, e35	6	144

92	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. *Nature Genetics*, **2016**, 48, 1107-11	36.3	135
91	Accurately assessing the risk of schizophrenia conferred by rare copy-number variation affecting genes with brain function. *PLoS Genetics*, **2010**, 6, e1001097	6	118
90	Schizophrenia susceptibility alleles are enriched for alleles that affect gene expression in adult human brain. *Molecular Psychiatry*, **2012**, 17, 193-201	15.1	104
89	Phenotype risk scores identify patients with unrecognized Mendelian disease patterns. *Science*, **2018**, 359, 1233-1239	33.3	101
88	Gene expression imputation across multiple brain regions provides insights into schizophrenia risk. *Nature Genetics*, **2019**, 51, 659-674	36.3	99
87	Genetic basis of individual differences in the response to small-molecule drugs in yeast. *Nature Genetics*, **2007**, 39, 496-502	36.3	93
86	Transcriptional signatures of schizophrenia in hiPSC-derived NPCs and neurons are concordant with post-mortem adult brains. *Nature Communications*, **2017**, 8, 2225	17.4	92
85	Genome-wide analysis of nucleotide-level variation in commonly used Saccharomyces cerevisiae strains. *PLoS ONE*, **2007**, 2, e322	3.7	88
84	Network-assisted investigation of combined causal signals from genome-wide association studies in schizophrenia. *PLoS Computational Biology*, **2012**, 8, e1002587	5	86
83	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. *Nature Genetics*, **2021**, 53, 817-829	36.3	83
82	Dysregulation of miRNA-9 in a Subset of Schizophrenia Patient-Derived Neural Progenitor Cells. *Cell Reports*, **2016**, 15, 1024-1036	10.6	82
81	Analysis of protein-coding genetic variation in 60,706 humans		81
80	Validation of electronic health record phenotyping of bipolar disorder cases and controls. *American Journal of Psychiatry*, **2015**, 172, 363-72	11.9	79
79	20.4 MODELING THE CONTRIBUTION OF COMMON VARIANTS TO SCHIZOPHRENIA RISK. *Schizophrenia Bulletin*, **2018**, 44, S34-S34	1.3	78
78	Polygenic overlap between schizophrenia risk and antipsychotic response: a genomic medicine approach. *Lancet Psychiatry,the*, **2016**, 3, 350-7	23.3	77
77	Improving genetic prediction by leveraging genetic correlations among human diseases and traits. *Nature Communications*, **2018**, 9, 989	17.4	76
76	Penetrance and Pleiotropy of Polygenic Risk Scores for Schizophrenia in 106,160 Patients Across Four Health Care Systems. *American Journal of Psychiatry*, **2019**, 176, 846-855	11.9	73
75	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

74	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
73	Prediction of human population responses to toxic compounds by a collaborative competition. *Nature Biotechnology*, **2015**, 33, 933-40	44.5	70
72	A bias-reducing pathway enrichment analysis of genome-wide association data confirmed association of the MHC region with schizophrenia. *Journal of Medical Genetics*, **2012**, 49, 96-103	5.8	64
71	Cis-acting regulation of brain-specific ANK3 gene expression by a genetic variant associated with bipolar disorder. *Molecular Psychiatry*, **2013**, 18, 922-9	15.1	58
70	GWA study data mining and independent replication identify cardiomyopathy-associated 5 (CMYA5) as a risk gene for schizophrenia. *Molecular Psychiatry*, **2011**, 16, 1117-29	15.1	58
69	The Genetics of the Mood Disorder Spectrum: Genome-wide Association Analyses of More Than 185,000 Cases and 439,000 Controls. *Biological Psychiatry*, **2020**, 88, 169-184	7.9	57
68	Implication of a rare deletion at distal 16p11.2 in schizophrenia. *JAMA Psychiatry*, **2013**, 70, 253-60	14.5	56
67	Estimation of Genetic Correlation via Linkage Disequilibrium Score Regression and Genomic Restricted Maximum Likelihood. *American Journal of Human Genetics*, **2018**, 102, 1185-1194	11	55
66	Rare copy number variation in treatment-resistant major depressive disorder. *Biological Psychiatry*, **2014**, 76, 536-41	7.9	54
65	Disruption of a large intergenic noncoding RNA in subjects with neurodevelopmental disabilities. *American Journal of Human Genetics*, **2012**, 91, 1128-34	11	53
64	Investigation of the genetic association between quantitative measures of psychosis and schizophrenia: a polygenic risk score analysis. *PLoS ONE*, **2012**, 7, e37852	3.7	53
63	Genetic modifiers and subtypes in schizophrenia: investigations of age at onset, severity, sex and family history. *Schizophrenia Research*, **2014**, 154, 48-53	3.6	49
62	Large-Scale Identification of Common Trait and Disease Variants Affecting Gene Expression. *American Journal of Human Genetics*, **2017**, 100, 885-894	11	48
61	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. *Genome Medicine*, **2017**, 9, 114	14.4	48
60	Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. *Human Molecular Genetics*, **2013**, 22, 2529-38	5.6	48
59	Genome-wide association study identifies SESTD1 as a novel risk gene for lithium-responsive bipolar disorder. *Molecular Psychiatry*, **2016**, 21, 1290-7	15.1	45
58	Genetic identification of a common collagen disease in puerto ricans via identity-by-descent mapping in a health system. *ELife*, **2017**, 6,	8.9	44
57	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. *International Journal of Epidemiology*, **2015**, 44, 1706-21	7.8	43

56	Translocations disrupting PHF21A in the Potocki-Shaffer-syndrome region are associated with intellectual disability and craniofacial anomalies. *American Journal of Human Genetics*, **2012**, 91, 56-72	11	42
55	Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide. *Molecular Psychiatry*, **2020**, 25, 2422-2430	15.1	36
54	Schizophrenia genetic variants are not associated with intelligence. *Psychological Medicine*, **2013**, 43, 2563-70	6.9	34
53	Revealing complex traits with small molecules and naturally recombinant yeast strains. *Chemistry and Biology*, **2006**, 13, 319-27		34
52	and Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 981-994	12.7	30
51	Genome-wide association study identifies 30 Loci Associated with Bipolar Disorder		28
50	A family-based study of common polygenic variation and risk of schizophrenia. *Molecular Psychiatry*, **2011**, 16, 887-8	15.1	26
49	Analysis of exome sequence in 604 trios for recessive genotypes in schizophrenia. *Translational Psychiatry*, **2015**, 5, e607	8.6	25
48	Genetic variants and functional pathways associated with resilience to Alzheimer disease. *Brain*, **2020**, 143, 2561-2575	11.2	25
47	Variability in working memory performance explained by epistasis vs polygenic scores in the ZNF804A pathway. *JAMA Psychiatry*, **2014**, 71, 778-785	14.5	24
46	Genome-wide association study reveals greater polygenic loading for schizophrenia in cases with a family history of illness. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2016** , 171B, 276-89	3.5	23
45	Copy number variation in subjects with major depressive disorder who attempted suicide. *PLoS ONE*, **2012**, 7, e46315	3.7	22
44	Analysis of Genetically Regulated Gene Expression Identifies a Prefrontal PTSD Gene, SNRNP35, Specific to Military Cohorts. *Cell Reports*, **2020**, 31, 107716	10.6	21
43	Contribution of Rare Copy Number Variants to Bipolar Disorder Risk Is Limited to Schizoaffective Cases. *Biological Psychiatry*, **2019**, 86, 110-119	7.9	20
42	Two non-synonymous markers in PTPN21, identified by genome-wide association study data-mining and replication, are associated with schizophrenia. *Schizophrenia Research*, **2011**, 131, 43-51	3.6	19
41	Functional annotation of rare structural variation in the human brain. *Nature Communications*, **2020**, 11, 2990	17.4	18
40	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. *European Journal of Human Genetics*, **2015**, 23, 555-7	5.3	17
39	A Comparison of Ten Polygenic Score Methods for Psychiatric Disorders Applied Across Multiple Cohorts. *Biological Psychiatry*, **2021**, 90, 611-620	7.9	17

38	Highly penetrant alterations of a critical region including BDNF in human psychopathology and obesity. *Archives of General Psychiatry*, **2012**, 69, 1238-46		16
37	Fine-mapping reveals novel alternative splicing of the dopamine transporter. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1434-47	3.5	16
36	No Reliable Association between Runs of Homozygosity and Schizophrenia in a Well-Powered Replication Study. *PLoS Genetics*, **2016**, 12, e1006343	6	15
35	Genetic validation of bipolar disorder identified by automated phenotyping using electronic health records. *Translational Psychiatry*, **2018**, 8, 86	8.6	14
34	Genetic classification of populations using supervised learning. *PLoS ONE*, **2011**, 6, e14802	3.7	14
33	Using expression and genotype to predict drug response in yeast. *PLoS ONE*, **2009**, 4, e6907	3.7	14
32	Family-based genetic risk prediction of multifactorial disease. *Genome Medicine*, **2010**, 2, 2	14.4	13
31	Vaccine-preventable outbreaks: still with us after all these years. *Pediatric Annals*, **2015**, 44, e76-81	1.3	12
30	Genome-wide association study of over 40,000 bipolar disorder cases provides new insights into the underlying biology		11
29	Clinical laboratory test-wide association scan of polygenic scores identifies biomarkers of complex disease. *Genome Medicine*, **2021**, 13, 6	14.4	11
28	Mosaic copy number variation in schizophrenia. *European Journal of Human Genetics*, **2013**, 21, 1007-11	5.3	10
27	Transcriptomic Imputation of Bipolar Disorder and Bipolar subtypes reveals 29 novel associated genes		9
26	Genetic risk for major depressive disorder and loneliness in sex-specific associations with coronary artery disease. *Molecular Psychiatry*, **2021**, 26, 4254-4264	15.1	9
25	Genetic risk for schizophrenia and autism, social impairment and developmental pathways to psychosis. *Translational Psychiatry*, **2018**, 8, 204	8.6	9
24	Characterization of Single Gene Copy Number Variants in Schizophrenia. *Biological Psychiatry*, **2020**, 87, 736-744	7.9	8
23	Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide		7
22	APOE ε4-specific associations of VEGF gene family expression with cognitive aging and Alzheimer's disease. *Neurobiology of Aging*, **2020**, 87, 18-25	5.6	7
21	Co-localization of Conditional eQTL and GWAS Signatures in Schizophrenia		6

20	Gene expression imputation across multiple brain regions reveals schizophrenia risk throughout development		6
19	Bipolar disorder and a history of suicide attempts with a duplication in 5HTR1A. *American Journal of Psychiatry*, **2012**, 169, 1213-4	11.9	3
18	Learning Opportunities for Drug Repositioning via GWAS and PheWAS Findings. *AMIA Summits on Translational Science Proceedings*, **2018**, 2017, 237-246	1.1	3
17	Gene Expression Elucidates Functional Impact of Polygenic Risk for Schizophrenia		3
16	Functional annotation of rare structural variation in the human brain		3
15	The ExAC Browser: Displaying reference data information from over 60,000 exomes		3
14	Phenotypic signatures in clinical data enable systematic identification of patients for genetic testing. *Nature Medicine*, **2021**, 27, 1097-1104	50.5	3
13	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. *Molecular Psychiatry*, **2021**, 26, 5239-5250	15.1	3
12	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2019**, 180, 223-231	3.5	2
11	Deep phenotyping predicts Huntington's genotype. *Nature Biotechnology*, **2016**, 34, 823-4	44.5	2
10	Characterisation of age and polarity at onset in bipolar disorder.. *British Journal of Psychiatry*, **2021**, 219, 659-669	5.4	2
9	Characterization of single gene copy number variants in schizophrenia		1
8	Identifying tissues implicated in Anorexia Nervosa using Transcriptomic Imputation		1
7	Analysis of Genetically Regulated Gene Expression identifies a trauma type specific PTSD gene, SNRNP35		1
6	Characterization of Age and Polarity at Onset in Bipolar Disorder		1
5	Expanding cultural and ancestral representation in psychiatric genetic studies. *Neuropsychopharmacology*, **2020**, 45, 1593-1594	8.7	1
4	Genomics-driven screening for causal determinants of suicide attempt.. *Australian and New Zealand Journal of Psychiatry*, **2022**, 48674221091499	2.6	0
3	[S4.3]: Large-scale genetic studies of rare and common variation in schizophrenia risk. *International Journal of Developmental Neuroscience*, **2010**, 28, 647-647	2.7	

2	VEGF-family brain protein abundance: Associations with Alzheimer's disease pathology and cognitive decline.. *Alzheimer's and Dementia*, **2021**, 17 Suppl 3, e052984	1.2
1	Sex differences in the genetic architecture underlying resilience in AD.. *Alzheimer's and Dementia*, **2021**, 17 Suppl 3, e055010	1.2