Kathryn M Roeder

List of Publications by Citations

156 papers	26,395 citations	69 h-index	162 g-index
178 ext. papers	32,223 ext. citations	10.9 avg, IF	6.51 L-index

#	Paper	IF	Citations
156	Genomic control for association studies. *Biometrics*, **1999**, 55, 997-1004	1.8	2262
155	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
154	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
153	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. *Nature*, **2012**, 485, 237-41	50.4	1470
152	A SAS Procedure Based on Mixture Models for Estimating Developmental Trajectories. *Sociological Methods and Research*, **2001**, 29, 374-393	2.9	1466
151	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
150	Multiple recurrent de novo CNVs, including duplications of the 7q11.23 Williams syndrome region, are strongly associated with autism. *Neuron*, **2011**, 70, 863-85	13.9	932
149	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
148	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746
147	Most genetic risk for autism resides with common variation. *Nature Genetics*, **2014**, 46, 881-5	36.3	734
146	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
145	Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nature Neuroscience*, **2016**, 19, 1442-1453	25.5	622
144	Coexpression networks implicate human midfetal deep cortical projection neurons in the pathogenesis of autism. *Cell*, **2013**, 155, 997-1007	56.2	591
143	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
142	Genome-wide association identifies multiple ulcerative colitis susceptibility loci. *Nature Genetics*, **2010**, 42, 332-7	36.3	491
141	Testing for an unusual distribution of rare variants. *PLoS Genetics*, **2011**, 7, e1001322	6	465
140	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443

139	Genomic control, a new approach to genetic-based association studies. *Theoretical Population Biology*, **2001**, 60, 155-66	1.2	403
138	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
137	The heritability of IQ. *Nature*, **1997**, 388, 468-71	50.4	369
136	Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. *Nature Genetics*, **2009**, 41, 216-20	36.3	325
135	Practical Bayesian Density Estimation Using Mixtures of Normals. *Journal of the American Statistical Association*, **1997**, 92, 894-902	2.8	303
134	Common genetic variants, acting additively, are a major source of risk for autism. *Molecular Autism*, **2012**, 3, 9	6.5	294
133	The power of genomic control. *American Journal of Human Genetics*, **2000**, 66, 1933-44	11	294
132	Modeling Uncertainty in Latent Class Membership: A Case Study in Criminology. *Journal of the American Statistical Association*, **1999**, 94, 766-776	2.8	284
131	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
130	HIGH DIMENSIONAL VARIABLE SELECTION. *Annals of Statistics*, **2009**, 37, 2178-2201	3.2	260
129	Common genetic variants in the CLDN2 and PRSS1-PRSS2 loci alter risk for alcohol-related and sporadic pancreatitis. *Nature Genetics*, **2012**, 44, 1349-54	36.3	223
128	The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. *Nature Communications*, **2015**, 6, 6404	17.4	218
127	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
126	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
125	False discovery control with p-value weighting. *Biometrika*, **2006**, 93, 509-524	2	174
124	Using linkage genome scans to improve power of association in genome scans. *American Journal of Human Genetics*, **2006**, 78, 243-52	11	173
123	Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes. *PLoS Genetics*, **2013**, 9, e1003671	6	168
122	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

121	Vitamin D insufficiency and severe asthma exacerbations in Puerto Rican children. *American Journal of Respiratory and Critical Care Medicine*, **2012**, 186, 140-6	10.2	154
120	Density Estimation with Confidence Sets Exemplified by Superclusters and Voids in the Galaxies. *Journal of the American Statistical Association*, **1990**, 85, 617-624	2.8	146
119	The huge Package for High-dimensional Undirected Graph Estimation in R. *Journal of Machine Learning Research*, **2012**, 13, 1059-1062	28.6	141
118	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134
117	Genomic Control to the extreme. *Nature Genetics*, **2004**, 36, 1129-30; author reply 1131	36.3	130
116	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron*, **2012**, 76, 1052-6	13.9	124
115	On the identification of disease mutations by the analysis of haplotype similarity and goodness of fit. *American Journal of Human Genetics*, **2003**, 72, 891-902	11	119
114	Unbiased methods for population-based association studies. *Genetic Epidemiology*, **2001**, 21, 273-84	2.6	119
113	De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. *Cell Reports*, **2014**, 9, 16-23	10.6	118
112	SNP-based analysis of genetic substructure in the German population. *Human Heredity*, **2006**, 62, 20-9	1.1	109
111	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
110	On the use of general control samples for genome-wide association studies: genetic matching highlights causal variants. *American Journal of Human Genetics*, **2008**, 82, 453-63	11	106
109	Pleiotropy and principal components of heritability combine to increase power for association analysis. *Genetic Epidemiology*, **2008**, 32, 9-19	2.6	105
108	Evolutionary-based association analysis using haplotype data. *Genetic Epidemiology*, **2003**, 25, 48-58	2.6	100
107	Stability Approach to Regularization Selection (StARS) for High Dimensional Graphical Models. *Advances in Neural Information Processing Systems*, **2010**, 24, 1432-1440	2.2	98
106	Association studies for quantitative traits in structured populations. *Genetic Epidemiology*, **2002**, 22, 78-93	2.6	96
105	Characterization of multilocus linkage disequilibrium. *Genetic Epidemiology*, **2005**, 28, 193-206	2.6	95
104	Transmission/disequilibrium test meets measured haplotype analysis: family-based association analysis guided by evolution of haplotypes. *American Journal of Human Genetics*, **2001**, 68, 1250-63	11	90

103	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. *Molecular Autism*, **2014**, 5, 22	6.5	89
102	Practical Bayesian Density Estimation Using Mixtures of Normals		89
101	Analysis of single-locus tests to detect gene/disease associations. *Genetic Epidemiology*, **2005**, 28, 207-19	2.6	88
100	Screen and clean: a tool for identifying interactions in genome-wide association studies. *Genetic Epidemiology*, **2010**, 34, 275-85	2.6	87
99	Flexible parametric measurement error models. *Biometrics*, **1999**, 55, 44-54	1.8	82
98	Disequilibrium mapping: composite likelihood for pairwise disequilibrium. *Genomics*, **1996**, 36, 1-16	4.3	82
97	Do common variants play a role in risk for autism? Evidence and theoretical musings. *Brain Research* , **2011**, 1380, 78-84	3.7	80
96	Transcriptional consequences of 16p11.2 deletion and duplication in mouse cortex and multiplex autism families. *American Journal of Human Genetics*, **2014**, 94, 870-83	11	78
95	A Graphical Technique for Determining the Number of Components in a Mixture of Normals. *Journal of the American Statistical Association*, **1994**, 89, 487-495	2.8	77
94	Improving power in genome-wide association studies: weights tip the scale. *Genetic Epidemiology*, **2007**, 31, 741-7	2.6	76
93	Discovering genetic ancestry using spectral graph theory. *Genetic Epidemiology*, **2010**, 34, 51-9	2.6	74
92	Genome-Wide Significance Levels and Weighted Hypothesis Testing. *Statistical Science*, **2009**, 24, 398-413	2.4	74
91	African ancestry and lung function in Puerto Rican children. *Journal of Allergy and Clinical Immunology*, **2012**, 129, 1484-90.e6	11.5	73
90	A Semiparametric Mixture Approach to Case-Control Studies with Errors in Covariables. *Journal of the American Statistical Association*, **1996**, 91, 722-732	2.8	73
89	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
88	Application of Maximum Likelihood Methods to Population Genetic Data for the Estimation of Individual Fertilities. *Biometrics*, **1989**, 45, 363	1.8	70
87	Global spectral clustering in dynamic networks. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 927-932	11.5	65
86	Analysis of multilocus models of association. *Genetic Epidemiology*, **2003**, 25, 36-47	2.6	65

85	A statistical model for locating regulatory regions in genomic DNA. *Journal of Molecular Biology*, **1997**, 268, 8-14	6.5	61
84	A Bayesian semiparametric model for case-control studies with errors in variables. *Biometrika*, **1997**, 84, 523-537	2	57
83	High-density association study of 383 candidate genes for volumetric BMD at the femoral neck and lumbar spine among older men. *Journal of Bone and Mineral Research*, **2009**, 24, 2039-49	6.3	54
82	Haplotype fine mapping by evolutionary trees. *American Journal of Human Genetics*, **2000**, 66, 659-73	11	53
81	Modeling Uncertainty in Latent Class Membership: A Case Study in Criminology		53
80	Residual Diagnostics for Mixture Models. *Journal of the American Statistical Association*, **1992**, 87, 785-794	2.8	49
79	DNA Fingerprinting: A Review of the Controversy. *Statistical Science*, **1994**, 9,	2.4	46
78	An interactome perturbation framework prioritizes damaging missense mutations for developmental disorders. *Nature Genetics*, **2018**, 50, 1032-1040	36.3	44
77	Candidate gene analysis of femoral neck trabecular and cortical volumetric bone mineral density in older men. *Journal of Bone and Mineral Research*, **2010**, 25, 330-8	6.3	44
76	Mixture models for linkage analysis of affected sibling pairs and covariates. *Genetic Epidemiology*, **2002**, 22, 52-65	2.6	44
75	Genomic control for association studies: a semiparametric test to detect excess-haplotype sharing. *Biostatistics*, **2000**, 1, 369-87	3.7	44
74	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. *Cell Reports*, **2020**, 31, 107489	10.6	43
73	A UNIFIED STATISTICAL FRAMEWORK FOR SINGLE CELL AND BULK RNA SEQUENCING DATA. *Annals of Applied Statistics*, **2018**, 12, 609-632	2.1	42
72	Forensic Inference from DNA Fingerprints. *Journal of the American Statistical Association*, **1992**, 87, 337-350	2.8	38
71	Semisoft clustering of single-cell data. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 466-471	11.5	36
70	Common risk variants identified in autism spectrum disorder		32
69	The Yin and Yang of Autism Genetics: How Rare De Novo and Common Variations Affect Liability. *Annual Review of Genomics and Human Genetics*, **2017**, 18, 167-187	9.7	31
68	APOE and TREM2 regulate amyloid-responsive microglia in Alzheimer's disease. *Acta Neuropathologica*, **2020**, 140, 477-493	14.3	31

67	Depression and mental health help-seeking behaviors in a predominantly African American population of children and adolescents with epilepsy. *Epilepsia*, **2009**, 50, 1943-52	6.4	30
66	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
65	TOMM40 poly-T repeat lengths, age of onset and psychosis risk in Alzheimer disease. *Neurobiology of Aging*, **2011**, 32, 2328.e1-9	5.6	28
64	Genome-wide distribution of linkage disequilibrium in the population of Palau and its implications for gene flow in Remote Oceania. *Human Genetics*, **2001**, 108, 521-8	6.3	28
63	A Unified Treatment of Integer Parameter Models. *Journal of the American Statistical Association*, **1987**, 82, 758-764	2.8	28
62	A Graphical Technique for Determining the Number of Components in a Mixture of Normals		28
61	Uniqueness of estimation and identifiability in mixture models. *Canadian Journal of Statistics*, **1993**, 21, 139-147	0.4	25
60	NETWORK ASSISTED ANALYSIS TO REVEAL THE GENETIC BASIS OF AUTISM. *Annals of Applied Statistics*, **2015**, 9, 1571-1600	2.1	24
59	Copy number variants for schizophrenia and related psychotic disorders in Oceanic Palau: risk and transmission in extended pedigrees. *Biological Psychiatry*, **2011**, 70, 1115-21	7.9	23
58	Outlier Detection and False Discovery Rates for Whole-Genome DNA Matching. *Journal of the American Statistical Association*, **2003**, 98, 236-246	2.8	23
57	Integration of association statistics over genomic regions using Bayesian adaptive regression splines. *Human Genomics*, **2003**, 1, 20-9	6.8	22
56	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21
55	Differential activity of transcribed enhancers in the prefrontal cortex of 537 cases with schizophrenia and controls. *Molecular Psychiatry*, **2019**, 24, 1685-1695	15.1	20
54	Positive Semidefinite Rank-based Correlation Matrix Estimation with Application to Semiparametric Graph Estimation. *Journal of Computational and Graphical Statistics*, **2014**, 23, 895-922	1.4	20
53	A Semiparametric Mixture Approach to Case-Control Studies with Errors in Covariables		20
52	A SPECTRAL GRAPH APPROACH TO DISCOVERING GENETIC ANCESTRY. *Annals of Applied Statistics*, **2010**, 4, 179-202	2.1	19
51	Residual Diagnostics for Mixture Models		19
50	A Bayesian hierarchical model for allele frequencies. *Genetic Epidemiology*, **2001**, 20, 17-33	2.6	18

49	Overdispersion Diagnostics for Generalized Linear Models. *Journal of the American Statistical Association*, **1995**, 90, 1225-1236	2.8	17
48	Searching for disease susceptibility variants in structured populations. *Genomics*, **2009**, 93, 1-4	4.3	14
47	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. *American Journal of Human Genetics*, **2016**, 98, 857-868	11	14
46	Using ancestry matching to combine family-based and unrelated samples for genome-wide association studies. *Statistics in Medicine*, **2010**, 29, 2932-45	2.3	13
45	Using multiple measurements of tissue to estimate subject- and cell-type-specific gene expression. *Bioinformatics*, **2020**, 36, 782-788	7.2	13
44	Functional connectome fingerprinting accuracy in youths and adults is similar when examined on the same day and 1.5-years apart. *Human Brain Mapping*, **2020**, 41, 4187-4199	5.9	11
43	Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. *Genomics*, **2013**, 102, 270-7	4.3	11
42	Comments on the Statistical Aspects of the NRC's Report on DNA Typing. *Journal of Forensic Sciences*, **1994**, 39, 13568J	1.8	11
41	Forensic Inference from DNA Fingerprints		11
40	TESTING HIGH-DIMENSIONAL COVARIANCE MATRICES, WITH APPLICATION TO DETECTING SCHIZOPHRENIA RISK GENES. *Annals of Applied Statistics*, **2017**, 11, 1810-1831	2.1	10
39	Genetic liability to schizophrenia in Oceanic Palau: a search in the affected and maternal generation. *Human Genetics*, **2007**, 121, 675-84	6.3	10
38	Overdispersion Diagnostics for Generalized Linear Models		9
37	REFINING GENETICALLY INFERRED RELATIONSHIPS USING TREELET COVARIANCE SMOOTHING. *Annals of Applied Statistics*, **2013**, 7, 669-690	2.1	8
36	De novo missense variants disrupting protein-protein interactions affect risk for autism through gene co-expression and protein networks in neuronal cell types. *Molecular Autism*, **2020**, 11, 76	6.5	8
35	H-MAGMA, inheriting a shaky statistical foundation, yields excess false positives. *Annals of Human Genetics*, **2021**, 85, 97-100	2.2	7
34	Integration and transfer learning of single-cell transcriptomes via cFIT. *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	7
33	Clustering and alignment of polymorphic sequences for HLA-DRB1 genotyping. *PLoS ONE*, **2013**, 8, e59835	3.7	6
32	Limited contribution of rare, noncoding variation to autism spectrum disorder from sequencing of 2,076 genomes in quartet families		6

31	Not All Autism Genes Are Created Equal: A Response to Myers et al. *American Journal of Human Genetics*, **2020**, 107, 1000-1003	11	5
30	STRUCTURED, SPARSE REGRESSION WITH APPLICATION TO HIV DRUG RESISTANCE. *Annals of Applied Statistics*, **2011**, 5, 628-644	2.1	5
29	Refining the role of de novo protein truncating variants in neurodevelopmental disorders using population reference samples		5
28	Conditional resampling improves calibration and sensitivity in single-cell CRISPR screen analysis		5
27	Functional connectome fingerprinting accuracy in youths and adults is similar when examined on the same day and 1.5 years apart		5
26	Discovery of target genes and pathways of blood trait loci using pooled CRISPR screens and single cell RNA sequencing		5
25	A selective inference approach for false discovery rate control using multiomics covariates yields insights into disease risk. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 15028-15035	11.5	4
24	Characterizing runs of homozygosity and their impact on risk for psychosis in a population isolate. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2014**, 165B, 521-30	3.5	4
23	Rare coding variation illuminates the allelic architecture, risk genes, cellular expression patterns, and phenotypic context of autism		4
22	Prevalence and phenotypic impact of rare potentially damaging variants in autism spectrum disorder. *Molecular Autism*, **2021**, 12, 65	6.5	4
21	ESCO: single cell expression simulation incorporating gene co-expression. *Bioinformatics*, **2021**,	7.2	4
20	MIRA: mutual information-based reporter algorithm for metabolic networks. *Bioinformatics*, **2014**, 30, i175-84	7.2	3
19	H-MAGMA, inheriting a shaky statistical foundation, yields excess false positives		3
18	Resting-State Functional Network Organization Is Stable Across Adolescent Development for Typical and Psychosis Spectrum Youth. *Schizophrenia Bulletin*, **2020**, 46, 395-407	1.3	2
17	Binning clones by hybridization with complex probes: statistical refinement of an inner product mapping method. *Genomics*, **1997**, 41, 141-54	4.3	2
16	Reply to Olson *Genetic Epidemiology*, **2002**, 23, 449-455	2.6	2
15	Moment-based oscillation properties of mixture models. *Annals of Statistics*, **1997**, 25,	3.2	2
14	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

13	How rare and common risk variation jointly affect liability for autism spectrum disorder. *Molecular Autism*, **2021**, 12, 66	6.5	2
12	Bayesian estimation of cell-type-specific gene expression per bulk sample with prior derived from single-cell data		2
11	Exponential-Family Embedding With Application to Cell Developmental Trajectories for Single-Cell RNA-Seq Data. *Journal of the American Statistical Association*, **2021**, 116, 457-470	2.8	2
10	SCEPTRE improves calibration and sensitivity in single-cell CRISPR screen analysis.. *Genome Biology*, **2021**, 22, 344	18.3	2
9	Analysis of Shared Haplotypes amongst Palauans Maps Loci for Psychotic Disorders to 4q28 and 5q23-q31. *Molecular Neuropsychiatry*, **2017**, 2, 173-184	4.9	1
8	How rare and common risk variation jointly affect liability for autism spectrum disorder		1
7	New mutations, old statistical challenges		1
6	A United Statistical Framework for Single Cell and Bulk Sequencing Data		1
5	Cell type hierarchy reconstruction via reconciliation of multi-resolution cluster tree. *Nucleic Acids Research*, **2021**, 49, e91	20.1	1
4	Covariance-based sample selection for heterogeneous data: Applications to gene expression and autism risk gene detection. *Journal of the American Statistical Association*, **2021**, 116, 54-67	2.8	1
3	The genetic architecture of obsessive-compulsive disorder: alleles across the frequency spectrum contribute liability to OCD		1
2	Bayesian estimation of cell type-specific gene expression with prior derived from single-cell data. *Genome Research*, **2021**, 31, 1807-1818	9.7	0
1	Rejoinder for Exponential-Family Embedding With Application to Cell Developmental Trajectories for Single-Cell RNA-Seq Data *Journal of the American Statistical Association*, **2021**, 116, 478-480	2.8	