James S. Sutcliffe

List of Publications by Year
in descending order

134 papers	39,899 citations	12303 69 h-index	13338 130 g-index
137 all docs	137 docs citations	137 times ranked	30024 citing authors

#	Article	IF	Citations
1	Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. Cell, 1991, 65, 905-914.	13.5	3,285
2	Strong Association of De Novo Copy Number Mutations with Autism. Science, 2007, 316, 445-449.	6.0	2,497
3	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
4	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
5	Variation of the CGG repeat at the fragile X site results in genetic instability: Resolution of the Sherman paradox. Cell, 1991, 67, 1047-1058.	13.5	2,007
6	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
7	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
8	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
9	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	9.4	1,272
10	Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. Nature, 2009, 459, 569-573.	13.7	1,270
11	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
12	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
13	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
14	Common genetic variants on 5p14.1 associate with autism spectrum disorders. Nature, 2009, 459, 528-533.	13.7	912
15	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
16	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. Nature Genetics, 1997, 15, 74-77.	9.4	801
17	DNA methylation represses FMR-1 transcription in fragile X syndrome. Human Molecular Genetics, 1992, 1, 397-400.	1.4	674
18	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663

#	Article	IF	Citations
19	Microduplications of 16p11.2 are associated with schizophrenia. Nature Genetics, 2009, 41, 1223-1227.	9.4	646
20	Contribution of SHANK3 Mutations to Autism Spectrum Disorder. American Journal of Human Genetics, 2007, 81, 1289-1297.	2.6	604
21	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	13.7	570
22	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
23	Imprinted expression of the murine Angelman syndrome gene, Ube3a, in hippocampal and Purkinje neurons. Nature Genetics, 1997, 17, 75-78.	9.4	466
24	Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. Nature Genetics, 1994, 8, 52-58.	9.4	418
25	A genetic variant that disrupts MET transcription is associated with autism. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 16834-16839.	3.3	389
26	A Multisite Study of the Clinical Diagnosis of Different Autism Spectrum Disorders. Archives of General Psychiatry, 2012, 69, 306.	13.8	385
27	Allelic Heterogeneity at the Serotonin Transporter Locus (SLC6A4) Confers Susceptibility to Autism and Rigid-Compulsive Behaviors. American Journal of Human Genetics, 2005, 77, 265-279.	2.6	378
28	Genome-Wide Analyses of Exonic Copy Number Variants in a Family-Based Study Point to Novel Autism Susceptibility Genes. PLoS Genetics, 2009, 5, e1000536.	1.5	374
29	Tissue specific expression of FMRâ€“1 provides evidence for a functional role in fragile X syndrome. Nature Genetics, 1993, 3, 36-43.	9.4	358
30	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	2.6	357
31	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	1.4	334
32	Autism gene variant causes hyperserotonemia, serotonin receptor hypersensitivity, social impairment and repetitive behavior. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 5469-5474.	3.3	278
33	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	3.8	265
34	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	1.5	253
35	Human and murine FMR-1: alternative splicing and translational initiation downstream of the CGGâ€“repeat. Nature Genetics, 1993, 4, 244-251.	9.4	247
36	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242

#	Article	IF	Citations
37	Partial and generalized epilepsy with febrile seizures plus and a novel SCN1A mutation. Neurology, 2001, 57, 2265-2272.	1.5	193
38	Analysis of the RELN gene as a genetic risk factor for autism. Molecular Psychiatry, 2005, 10, 563-571.	4.1	181
39	De novo mutation in the dopamine transporter gene associates dopamine dysfunction with autism spectrum disorder. Molecular Psychiatry, 2013, 18, 1315-1323.	4.1	181
40	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	1.8	180
41	Human serotonin transporter variants display altered sensitivity to protein kinase G and p38 mitogen-activated protein kinase. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 11545-11550.	3.3	167
42	Linkage Disequilibrium at the Angelman Syndrome Gene UBE3A in Autism Families. Genomics, 2001, 77, 105-113.	1.3	154
43	The spectrum of mutations in UBE3A causing Angelman syndrome. Human Molecular Genetics, 1999, 8, 129-135.	1.4	150
44	Association of oxytocin receptor (OXTR) gene variants with multiple phenotype domains of autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2011, 3, 101-112.	1.5	148
45	Genetic evidence implicating multiple genes in the MET receptor tyrosine kinase pathway in autism spectrum disorder. Autism Research, 2008, 1, 159-168.	2.1	143
46	Distinct Genetic Risk Based on Association of MET in Families With Co-occurring Autism and Gastrointestinal Conditions. Pediatrics, 2009, 123, 1018-1024.	1.0	141
47	A linkage disequilibrium map of the 1-Mb 15q12 GABAAreceptor subunit cluster and association to autism. American Journal of Medical Genetics Part A, 2004, 131B, 51-59.	2.4	135
48	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
49	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	0.7	133
50	Genome-wide and Ordered-Subset linkage analyses provide support for autism loci on 17q and 19p with evidence of phenotypic and interlocus genetic correlates. BMC Medical Genetics, 2005, 6, 1.	2.1	130
51	Imprinting analysis of three genes in the Prader â€" Willi/Angelman region: SNRPN, E6-associated protein, and PAR-2 (D15S225E). Human Molecular Genetics, 1994, 3, 309-315.	1.4	125
52	Enhanced activity of human serotonin transporter variants associated with autism. Philosophical Transactions of the Royal Society B: Biological Sciences, 2009, 364, 163-173.	1.8	120
53	Linkage and association analysis at the serotonin transporter (SLC6A4) locus in a rigid-compulsive subset of autism. American Journal of Medical Genetics Part A, 2004, 127B, 104-112.	2.4	118
54	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118

#	Article	IF	Citations
55	A Bayesian framework that integrates multi-omics data and gene networks predicts risk genes from schizophrenia GWAS data. Nature Neuroscience, 2019, 22, 691-699.	7.1	118
56	Exploratory Subsetting of Autism Families Based on Savant Skills Improves Evidence of Genetic Linkage to 15q11-q13. Journal of the American Academy of Child and Adolescent Psychiatry, 2003, 42, 856-863.	0.3	112
57	Autism and 15q11-q13 disorders: Behavioral, genetic, and pathophysiological issues. Mental Retardation and Developmental Disabilities Research Reviews, 2004, 10, 284-291.	3.5	112
58	Shorter sleep duration is associated with social impairment and comorbidities in ASD. Autism Research, 2017, 10, 1221-1238.	2.1	109
59	SLC6A3 coding variant Ala559Val found in two autism probands alters dopamine transporter function and trafficking. Translational Psychiatry, 2014, 4, e464-e464.	2.4	108
60	The E6â€“AP Ubiquitinâ€“Protein Ligase (UBE3A) Gene Is Localized within a Narrowed Angelman Syndrome Criticalâ€‰Region. Genome Research, 1997, 7, 368-377.	2.4	106
61	Maternal transmission of a rare GABRB3 signal peptide variant is associated with autism. Molecular Psychiatry, 2011, 16, 86-96.	4.1	106
62	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	1.4	101
63	A complete YAC contig of the Prader-Willi/Angelman chromosome region (15q11â€“q13) and refined localization of the SNRPN gene. Genomics, 1993, 18, 546-552.	1.3	100
64	Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. Journal of Medical Genetics, 2011, 48, 48-54.	1.5	94
65	Validation studies of SNRPN methylation as a diagnostic test for Prader-Willi syndrome. , 1996, 66, 77-80.		87
66	Mouse/human sequence divergence in a region with a paternal-specific methylation imprint at the human H19 locus. Human Molecular Genetics, 1996, 5, 1155-1161.	1.4	85
67	A Novel HumanCAMK2AMutation Disrupts Dendritic Morphology and Synaptic Transmission, and Causes ASD-Related Behaviors. Journal of Neuroscience, 2017, 37, 2216-2233.	1.7	83
68	Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. American Journal of Medical Genetics Part A, 1992, 43, 237-243.	2.4	82
69	Variation in ITGB3 is associated with whole-blood serotonin level and autism susceptibility. European Journal of Human Genetics, 2006, 14, 923-931.	1.4	82
70	Association and Mutation Analyses of 16p11.2 Autism Candidate Genes. PLoS ONE, 2009, 4, e4582.	1.1	80
71	Appetitive behavior, compulsivity, and neurochemistry in Prader-Willi syndrome. Mental Retardation and Developmental Disabilities Research Reviews, 2000, 6, 125-130.	3.5	75
72	Integrated YAC Contig Map of the Praderâ€“Willi/Angelman Region on Chromosome 15q11â€“q13 with Average STS Spacing of 35â€‰kb. Genome Research, 1998, 8, 146-157.	2.4	72

#	Article	IF	Citations
73	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	0.7	70
74	Rare Autism-Associated Variants Implicate Syntaxin 1 (STX1 R26Q) Phosphorylation and the Dopamine Transporter (hDAT R51W) in Dopamine Neurotransmission and Behaviors. EBioMedicine, 2015, 2, 135-146.	2.7	70
75	Tissue-specific and allele-specific replication timing control in the imprinted human Prader-Willi syndrome region.. Genes and Development, 1995, 9, 808-820.	2.7	67
76	Intellectual Disability Is Associated with Increased Runs of Homozygosity in Simplex Autism. American Journal of Human Genetics, 2013, 93, 103-109.	2.6	63
77	Dense linkage disequilibrium mapping in the 15q11â€“q13 maternal expression domain yields evidence for association in autism. Molecular Psychiatry, 2003, 8, 624-634.	4.1	60
78	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15<scp>q</scp>11.2, Specifically Breakpoints 1 to 2. Autism Research, 2014, 7, 355-362.	2.1	59
79	Molecular Genetics of the Platelet Serotonin System in First-Degree Relatives of Patients with Autism. Neuropsychopharmacology, 2008, 33, 353-360.	2.8	57
80	Examination of NRCAM, LRRN3, KIAA0716, and LAMB1as autism candidate genes. BMC Medical Genetics, 2004, 5, 12.	2.1	56
81	Neuronally-expressed necdin gene: an imprinted candidate gene in Prader-Willi syndrome. Lancet, The, 1997, 350, 1520-1521.	6.3	53
82	Insights into the Pathogenesis of Autism. Science, 2008, 321, 208-209.	6.0	53
83	Genetics of Childhood Disorders: XLVII. Autism, Part 6: Duplication and Inherited Susceptibility of Chromosome 15q11-q13 Genes in Autism. Journal of the American Academy of Child and Adolescent Psychiatry, 2003, 42, 253-256.	0.3	51
84	Genetic analysis of biological pathway data through genomic randomization. Human Genetics, 2011, 129, 563-571.	1.8	50
85	Leveraging blood serotonin as an endophenotype to identify de novo and rare variants involved in autism. Molecular Autism, 2017, 8, 14.	2.6	50
86	Association of MET with social and communication phenotypes in individuals with autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 438-446.	1.1	49
87	Modeling rare gene variation to gain insight into the oldest biomarker in autism: construction of the serotonin transporter Gly56Ala knock-in mouse. Journal of Neurodevelopmental Disorders, 2009, 1, 158-171.	1.5	43
88	Parent-of-origin effects of the serotonin transporter gene associated with autism. , 2011, 156, 139-144.		41
89	Analysis of CHRNA7 rare variants in autism spectrum disorder susceptibility. American Journal of Medical Genetics, Part A, 2015, 167, 715-723.	0.7	41
90	Autosomal Dominant Lateral Temporal Epilepsy: Two Families with Novel Mutations in the LGI1 Gene. Epilepsia, 2004, 45, 218-222.	2.6	39

#	Article	IF	Citations
91	Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. European Journal of Human Genetics, 2011, 19, 1082-1089.	1.4	39
92	Loci nominally associated with autism from genome-wide analysis show enrichment of brain expression quantitative trait loci but not lymphoblastoid cell line expression quantitative trait loci. Molecular Autism, 2012, 3, 3.	2.6	38
93	Lack of Association Between Autism andSLC25A12. American Journal of Psychiatry, 2006, 163, 929-931.	4.0	36
94	Genetic background modulates phenotypes of serotonin transporter Ala56 knock-in mice. Molecular Autism, 2013, 4, 35.	2.6	35
95	Structural, functional, and behavioral insights of dopamine dysfunction revealed by a deletion in SLC6A3. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 3853-3862.	3.3	35
96	Defining the autism minimum candidate gene region on chromosome 7. American Journal of Medical Genetics Part A, 2003, 117B, 90-96.	2.4	32
97	Analysis of the autism chromosome 2 linkage region: GAD1 and other candidate genes. Neuroscience Letters, 2004, 372, 209-214.	1.0	32
98	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. Molecular Autism, 2014, 5, 34.	2.6	31
99	Colocalization and Regulated Physical Association of Presynaptic Serotonin Transporters with A₃Adenosine Receptors. Molecular Pharmacology, 2011, 80, 458-465.	1.0	30
100	Integrin Î²3 Haploinsufficiency Modulates Serotonin Transport and Antidepressant-Sensitive Behavior in Mice. Neuropsychopharmacology, 2015, 40, 2015-2024.	2.8	26
101	Is there sexual dimorphism of hyperserotonemia in autism spectrum disorder?. Autism Research, 2017, 10, 1417-1423.	2.1	24
102	Maternal Serotonin Levels Are Associated With Cognitive Ability and Core Symptoms in Autism Spectrum Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2018, 57, 867-875.	0.3	24
103	Identification of a novel paternally expressed transcript adjacent to snRPN in the Prader-Willi syndrome critical region.. Genome Research, 1996, 6, 742-746.	2.4	23
104	Rare coding variants of the adenosine A3 receptor are increased in autism: on the trail of the serotonin transporter regulome. Molecular Autism, 2013, 4, 28.	2.6	23
105	Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. Journal of Neurodevelopmental Disorders, 2011, 3, 113-123.	1.5	22
106	The Gain-of-Function Integrin Î²3 Pro33 Variant Alters the Serotonin System in the Mouse Brain. Journal of Neuroscience, 2017, 37, 11271-11284.	1.7	22
107	Partial duplication of the APBA2 gene in chromosome 15q13 corresponds to duplicon structures. BMC Genomics, 2003, 4, 15.	1.2	20
108	Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. Human Genetics, 2015, 134, 191-201.	1.8	20

#	Article	IF	Citations
109	Zn2+ reverses functional deficits in a de novo dopamine transporter variant associated with autism spectrum disorder. Molecular Autism, 2015, 6, 8.	2.6	19
110	Possible dosage effect of maternally expressed genes on visual recognition memory in Prader-Willi syndrome. American Journal of Medical Genetics Part A, 2001, 105, 71-75.	2.4	18
111	Consensus Genotyper for Exome Sequencing (CGES): improving the quality of exome variant genotypes. Bioinformatics, 2015, 31, 187-193.	1.8	18
112	The GABBR1 locus and the G1465A variant is not associated with temporal lobe epilepsy preceded by febrile seizures. BMC Medical Genetics, 2005, 6, 13.	2.1	17
113	Accuracy of phenotyping children with autism based on parent report: what specifically do we gain phenotyping â€œrapidlyâ€?. Autism Research, 2012, 5, 31-38.	2.1	17
114	Sequencing and Functional Analysis of theSNRPNPromoter: In Vitro Methylation Abolishes Promoterâ€‰Activity. Genome Research, 1997, 7, 642-648.	2.4	16
115	â€˜Severeâ€™ Prader-Willi syndrome with a large deletion of chromosomeâ€ƒ15 due to an unbalanced t(15,22)(q14;q11.2) translocation. Clinical Genetics, 2003, 63, 79-81.	1.0	14
116	Mutations in GABRA1, GABRA5, GABRG2 and GABRD receptor genes are not a major factor in the pathogenesis of familial focal epilepsy preceded by febrile seizures. Neuroscience Letters, 2006, 394, 74-78.	1.0	14
117	A haplotype-based framework for group-wise transmission/disequilibrium tests for rare variant association analysis. Bioinformatics, 2015, 31, 1452-1459.	1.8	14
118	Detection of imprinting mutations in Angelman syndrome using a probe for exon Î± ofSNRPN. American Journal of Medical Genetics Part A, 1996, 63, 414-415.	2.4	13
119	Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. Genomics, 2013, 102, 270-277.	1.3	13
120	Strategy for molecular cloning of the fragile X site DNA. American Journal of Medical Genetics Part A, 1988, 30, 613-623.	2.4	12
121	Fine mapping and association studies in a candidate region for autism on chromosome 2q31â€“q32. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 535-544.	1.1	12
122	The impact of genotype calling errors on family-based studies. Scientific Reports, 2016, 6, 28323.	1.6	12
123	PCR amplification and analysis of yeast artificial chromosomes. Genomics, 1992, 13, 1303-1306.	1.3	11
124	The Drosophila Gene Sulfateless Modulates Autism-Like Behaviors. Frontiers in Genetics, 2019, 10, 574.	1.1	11
125	Drosophila melanogaster: a novel animal model for the behavioral characterization of autism-associated mutations in the dopamine transporter gene. Molecular Psychiatry, 2013, 18, 1235-1235.	4.1	9
126	Pro32Pro33 Mutations in the IntegrinÎ²₃PSI Domain Result inÎ±IIbÎ²₃Priming and Enhanced Adhesion: Reversal of the Hypercoagulability Phenotype by the Src Inhibitor SKI-606. Molecular Pharmacology, 2014, 85, 921-931.	1.0	7

#	Article	IF	Citations
127	Psychometric validation and refinement of the Interoception Sensory Questionnaire (ISQ) in adolescents and adults on the autism spectrum. Molecular Autism, 2021, 12, 42.	2.6	6
128	Angelman syndrome in an inbred family. Human Genetics, 1996, 97, 294-298.	1.8	5
129	Our vision for Autism Research. Autism Research, 2008, 1, 71-72.	2.1	3
130	Heterogeneity and the design of genetic studies in autism. Autism Research, 2008, 1, 205-206.	2.1	3
131	Affiliative Behaviors and Beyond: It's the Phenotype, Stupid. Biological Psychiatry, 2008, 63, 909-910.	0.7	2
132	Advantages of RT-PCR and denaturing gradient gel electrophoresis for analysis of genomic imprinting: Detection of new mouse and human expressed polymorphisms. , 1996, 7, 144-148.		1
133	An Automated Functional Annotation Pipeline That Rapidly Prioritizes Clinically Relevant Genes for Autism Spectrum Disorder. International Journal of Molecular Sciences, 2020, 21, 9029.	1.8	1
134	Calculating genetic risk for dysfunction in pleiotropic biological processes using whole exome sequencing data. Journal of Neurodevelopmental Disorders, 2022, 14, .	1.5	0