James S. Sutcliffe

List of Publications by Citations

131 papers	32,005 citations	67 h-index	137 g-index
137 ext. papers	36,918 ext. citations	13.1 avg, IF	5.7 L-index

#	Paper	IF	Citations
131	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, 905-14	56.2	2848
130	Strong association of de novo copy number mutations with autism. *Science*, **2007**, 316, 445-9	33.3	2126
129	Variation of the CGG repeat at the fragile X site results in genetic instability: resolution of the Sherman paradox. *Cell*, **1991**, 67, 1047-58	56.2	1786
128	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
127	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
126	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
125	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
124	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
123	Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. *Nature*, **2009**, 459, 569-73	50.4	1075
122	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
121	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
120	Common genetic variants on 5p14.1 associate with autism spectrum disorders. *Nature*, **2009**, 459, 528-33	50.4	760
119	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. *Nature Genetics*, **1997**, 15, 74-7	36.3	677
118	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
117	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
116	DNA methylation represses FMR-1 transcription in fragile X syndrome. *Human Molecular Genetics*, **1992**, 1, 397-400	5.6	607
115	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587

114	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
113	Microduplications of 16p11.2 are associated with schizophrenia. *Nature Genetics*, **2009**, 41, 1223-7	36.3	550
112	Contribution of SHANK3 mutations to autism spectrum disorder. *American Journal of Human Genetics*, **2007**, 81, 1289-97	11	492
111	A genome-wide linkage and association scan reveals novel loci for autism. *Nature*, **2009**, 461, 802-8	50.4	474
110	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443
109	Imprinted expression of the murine Angelman syndrome gene, Ube3a, in hippocampal and Purkinje neurons. *Nature Genetics*, **1997**, 17, 75-8	36.3	415
108	Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. *Nature Genetics*, **1994**, 8, 52-8	36.3	388
107	Allelic heterogeneity at the serotonin transporter locus (SLC6A4) confers susceptibility to autism and rigid-compulsive behaviors. *American Journal of Human Genetics*, **2005**, 77, 265-79	11	332
106	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, 16834-9	11.5	331
105	Tissue specific expression of FMR-1 provides evidence for a functional role in fragile X syndrome. *Nature Genetics*, **1993**, 3, 36-43	36.3	320
104	Genome-wide analyses of exonic copy number variants in a family-based study point to novel autism susceptibility genes. *PLoS Genetics*, **2009**, 5, e1000536	6	305
103	Common genetic variants, acting additively, are a major source of risk for autism. *Molecular Autism*, **2012**, 3, 9	6.5	294
102	A multisite study of the clinical diagnosis of different autism spectrum disorders. *Archives of General Psychiatry*, **2012**, 69, 306-13		289
101	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
100	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, 5469-74	11.5	225
99	Human and murine FMR-1: alternative splicing and translational initiation downstream of the CGG-repeat. *Nature Genetics*, **1993**, 4, 244-51	36.3	223
98	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
97	Partial and generalized epilepsy with febrile seizures plus and a novel SCN1A mutation. *Neurology*, **2001**, 57, 2265-72	6.5	169

96	Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes. *PLoS Genetics*, **2013**, 9, e1003671	6	168
95	Analysis of the RELN gene as a genetic risk factor for autism. *Molecular Psychiatry*, **2005**, 10, 563-71	15.1	154
94	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
93	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, 11545-50	11.5	148
92	Linkage disequilibrium at the Angelman syndrome gene UBE3A in autism families. *Genomics*, **2001**, 77, 105-13	4.3	145
91	De novo mutation in the dopamine transporter gene associates dopamine dysfunction with autism spectrum disorder. *Molecular Psychiatry*, **2013**, 18, 1315-23	15.1	127
90	Genetic evidence implicating multiple genes in the MET receptor tyrosine kinase pathway in autism spectrum disorder. *Autism Research*, **2008**, 1, 159-68	5.1	123
89	Association of oxytocin receptor (OXTR) gene variants with multiple phenotype domains of autism spectrum disorder. *Journal of Neurodevelopmental Disorders*, **2011**, 3, 101-12	4.6	119
88	A linkage disequilibrium map of the 1-Mb 15q12 GABA(A) receptor subunit cluster and association to autism. *American Journal of Medical Genetics Part A*, **2004**, 131B, 51-9		119
87	Imprinting analysis of three genes in the Prader-Willi/Angelman region: SNRPN, E6-associated protein, and PAR-2 (D15S225E). *Human Molecular Genetics*, **1994**, 3, 309-15	5.6	116
86	Distinct genetic risk based on association of MET in families with co-occurring autism and gastrointestinal conditions. *Pediatrics*, **2009**, 123, 1018-24	7.4	115
85	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, 775-84	7.9	113
84	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. *Neuron*, **2018**, 97, 488-493	13.9	112
83	The spectrum of mutations in UBE3A causing Angelman syndrome. *Human Molecular Genetics*, **1999**, 8, 129-35	5.6	112
82	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	110
81	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, 104-12		109
80	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
79	Autism and 15q11-q13 disorders: behavioral, genetic, and pathophysiological issues. *Mental Retardation and Developmental Disabilities Research Reviews*, **2004**, 10, 284-91		101

78	Enhanced activity of human serotonin transporter variants associated with autism. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2009**, 364, 163-73	5.8	97
77	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, 7974-81	11.5	94
76	Maternal transmission of a rare GABRB3 signal peptide variant is associated with autism. *Molecular Psychiatry*, **2011**, 16, 86-96	15.1	88
75	The E6-Ap ubiquitin-protein ligase (UBE3A) gene is localized within a narrowed Angelman syndrome critical region. *Genome Research*, **1997**, 7, 368-77	9.7	88
74	A complete YAC contig of the Prader-Willi/Angelman chromosome region (15q11-q13) and refined localization of the SNRPN gene. *Genomics*, **1993**, 18, 546-52	4.3	88
73	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, 856-63	7.3	87
72	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, 4360-70	5.6	84
71	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	5.8	81
70	Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. *American Journal of Medical Genetics Part A*, **1992**, 43, 237-43		75
69	Variation in ITGB3 is associated with whole-blood serotonin level and autism susceptibility. *European Journal of Human Genetics*, **2006**, 14, 923-31	5.3	73
68	Validation studies of SNRPN methylation as a diagnostic test for Prader-Willi syndrome. *American Journal of Medical Genetics Part A*, **1996**, 66, 77-80		71
67	Mouse/human sequence divergence in a region with a paternal-specific methylation imprint at the human H19 locus. *Human Molecular Genetics*, **1996**, 5, 1155-61	5.6	71
66	Association and mutation analyses of 16p11.2 autism candidate genes. *PLoS ONE*, **2009**, 4, e4582	3.7	68
65	Appetitive behavior, compulsivity, and neurochemistry in Prader-Willi syndrome. *Mental Retardation and Developmental Disabilities Research Reviews*, **2000**, 6, 125-30		67
64	SLC6A3 coding variant Ala559Val found in two autism probands alters dopamine transporter function and trafficking. *Translational Psychiatry*, **2014**, 4, e464	8.6	65
63	Integrated YAC contig map of the Prader-Willi/Angelman region on chromosome 15q11-q13 with average STS spacing of 35 kb. *Genome Research*, **1998**, 8, 146-57	9.7	64
62	A Bayesian framework that integrates multi-omics data and gene networks predicts risk genes from schizophrenia GWAS data. *Nature Neuroscience*, **2019**, 22, 691-699	25.5	62
61	Adjusting head circumference for covariates in autism: clinical correlates of a highly heritable continuous trait. *Biological Psychiatry*, **2013**, 74, 576-84	7.9	59

60	Shorter sleep duration is associated with social impairment and comorbidities in ASD. *Autism Research*, **2017**, 10, 1221-1238	5.1	55
59	Molecular genetics of the platelet serotonin system in first-degree relatives of patients with autism. *Neuropsychopharmacology*, **2008**, 33, 353-60	8.7	52
58	Dense linkage disequilibrium mapping in the 15q11-q13 maternal expression domain yields evidence for association in autism. *Molecular Psychiatry*, **2003**, 8, 624-34, 570	15.1	52
57	Intellectual disability is associated with increased runs of homozygosity in simplex autism. *American Journal of Human Genetics*, **2013**, 93, 103-9	11	51
56	Tissue-specific and allele-specific replication timing control in the imprinted human Prader-Willi syndrome region. *Genes and Development*, **1995**, 9, 808-20	12.6	51
55	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	8.8	50
54	A Novel Human Mutation Disrupts Dendritic Morphology and Synaptic Transmission, and Causes ASD-Related Behaviors. *Journal of Neuroscience*, **2017**, 37, 2216-2233	6.6	49
53	Modest impact on risk for autism spectrum disorder of rare copy number variants at 15q11.2, specifically breakpoints 1 to 2. *Autism Research*, **2014**, 7, 355-62	5.1	49
52	Genetic analysis of biological pathway data through genomic randomization. *Human Genetics*, **2011**, 129, 563-71	6.3	48
51	Examination of NRCAM, LRRN3, KIAA0716, and LAMB1 as autism candidate genes. *BMC Medical Genetics*, **2004**, 5, 12	2.1	48
50	Neuronally-expressed necdin gene: an imprinted candidate gene in Prader-Willi syndrome. *Lancet, The*, **1997**, 350, 1520-1	40	47
49	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, 253-6	7.2	46
48	Genetics. Insights into the pathogenesis of autism. *Science*, **2008**, 321, 208-9	33.3	44
47	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, 158-71	4.6	39
46	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	3.5	39
45	Autosomal dominant lateral temporal epilepsy: two families with novel mutations in the LGI1 gene. *Epilepsia*, **2004**, 45, 218-22	6.4	36
44	Parent-of-origin effects of the serotonin transporter gene associated with autism. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156, 139-44	3.5	34
43	Genetic background modulates phenotypes of serotonin transporter Ala56 knock-in mice. *Molecular Autism*, **2013**, 4, 35	6.5	33

42	Leveraging blood serotonin as an endophenotype to identify de novo and rare variants involved in autism. *Molecular Autism*, **2017**, 8, 14	6.5	30
41	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, 1082-9	5.3	30
40	Lack of association between autism and SLC25A12. *American Journal of Psychiatry*, **2006**, 163, 929-31	11.9	30
39	Analysis of the autism chromosome 2 linkage region: GAD1 and other candidate genes. *Neuroscience Letters*, **2004**, 372, 209-14	3.3	30
38	Analysis of CHRNA7 rare variants in autism spectrum disorder susceptibility. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 715-23	2.5	29
37	Colocalization and regulated physical association of presynaptic serotonin transporters with A adenosine receptors. *Molecular Pharmacology*, **2011**, 80, 458-65	4.3	28
36	Defining the autism minimum candidate gene region on chromosome 7. *American Journal of Medical Genetics Part A*, **2003**, 117B, 90-6		28
35	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	6.5	27
34	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. *Molecular Autism*, **2014**, 5, 34	6.5	25
33	Structural, functional, and behavioral insights of dopamine dysfunction revealed by a deletion in. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 3853-3862	11.5	21
32	Integrin Haploinsufficiency Modulates Serotonin Transport and Antidepressant-Sensitive Behavior in Mice. *Neuropsychopharmacology*, **2015**, 40, 2015-24	8.7	21
31	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21
30	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, 113-23	4.6	20
29	Identification of a novel paternally expressed transcript adjacent to snRPN in the Prader-Willi syndrome critical region. *Genome Research*, **1996**, 6, 742-6	9.7	19
28	The Gain-of-Function Integrin Pro33 Variant Alters the Serotonin System in the Mouse Brain. *Journal of Neuroscience*, **2017**, 37, 11271-11284	6.6	17
27	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	6.5	17
26	Zn(2+) reverses functional deficits in a de novo dopamine transporter variant associated with autism spectrum disorder. *Molecular Autism*, **2015**, 6, 8	6.5	16
25	Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. *Human Genetics*, **2015**, 134, 191-201	6.3	16

24	Partial duplication of the APBA2 gene in chromosome 15q13 corresponds to duplicon structures. *BMC Genomics*, **2003**, 4, 15	4.5	16
23	Sequencing and functional analysis of the SNRPN promoter: in vitro methylation abolishes promoter activity. *Genome Research*, **1997**, 7, 642-8	9.7	15
22	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		15
21	Is there sexual dimorphism of hyperserotonemia in autism spectrum disorder?. *Autism Research*, **2017**, 10, 1417-1423	5.1	14
20	Accuracy of phenotyping children with autism based on parent report: what specifically do we gain phenotyping "rapidly"?. *Autism Research*, **2012**, 5, 31-8	5.1	14
19	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	14
18	Consensus Genotyper for Exome Sequencing (CGES): improving the quality of exome variant genotypes. *Bioinformatics*, **2015**, 31, 187-93	7.2	13
17	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, 74-8	3.3	13
16	Severe Prader-Willi syndrome with a large deletion of chromosome 15 due to an unbalanced t(15,22)(q14;q11.2) translocation. *Clinical Genetics*, **2003**, 63, 79-81	4	13
15	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	7.2	13
14	A haplotype-based framework for group-wise transmission/disequilibrium tests for rare variant association analysis. *Bioinformatics*, **2015**, 31, 1452-9	7.2	12
13	Detection of imprinting mutations in Angelman syndrome using a probe for exon alpha of SNRPN. *American Journal of Medical Genetics Part A*, **1996**, 63, 414-5		12
12	Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. *Genomics*, **2013**, 102, 270-7	4.3	11
11	Fine mapping and association studies in a candidate region for autism on chromosome 2q31-q32. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2009**, 150B, 535-44	3.5	10
10	PCR amplification and analysis of yeast artificial chromosomes. *Genomics*, **1992**, 13, 1303-6	4.3	9
9	Strategy for molecular cloning of the fragile X site DNA. *American Journal of Medical Genetics Part A* , **1988**, 30, 613-23		9
8	The impact of genotype calling errors on family-based studies. *Scientific Reports*, **2016**, 6, 28323	4.9	8
7	Drosophila melanogaster: a novel animal model for the behavioral characterization of autism-associated mutations in the dopamine transporter gene. *Molecular Psychiatry*, **2013**, 18, 1235	15.1	8

6	The Drosophila Gene Modulates Autism-Like Behaviors. *Frontiers in Genetics*, **2019**, 10, 574	4.5	6
5	Pro32Pro33 mutations in the integrin β PSI domain result in αIIbβ priming and enhanced adhesion: reversal of the hypercoagulability phenotype by the Src inhibitor SKI-606. *Molecular Pharmacology*, **2014**, 85, 921-31	4.3	5
4	Angelman syndrome in an inbred family. *Human Genetics*, **1996**, 97, 294-8	6.3	5
3	Affiliative behaviors and beyond: it's the phenotype, stupid. *Biological Psychiatry*, **2008**, 63, 909-10	7.9	2
2	Advantages of RT-PCR and denaturing gradient gel electrophoresis for analysis of genomic imprinting: detection of new mouse and human expressed polymorphisms. *Human Mutation*, **1996**, 7, 144-8	4.7	1
1	Psychometric validation and refinement of the Interoception Sensory Questionnaire (ISQ) in adolescents and adults on the autism spectrum. *Molecular Autism*, **2021**, 12, 42	6.5	1