

James S. Sutcliffe

List of Publications by Year in descending order

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134
papers

39,899
citations

14124

69
h-index

15253

130
g-index

137
all docs

137
docs citations

137
times ranked

33195
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Calculating genetic risk for dysfunction in pleiotropic biological processes using whole exome sequencing data. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, . | 1.5 | 0 |
| 2 | Psychometric validation and refinement of the Interoception Sensory Questionnaire (ISQ) in adolescents and adults on the autism spectrum. <i>Molecular Autism</i> , 2021, 12, 42. | 2.6 | 6 |
| 3 | An Automated Functional Annotation Pipeline That Rapidly Prioritizes Clinically Relevant Genes for Autism Spectrum Disorder. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9029. | 1.8 | 1 |
| 4 | Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23. | 13.5 | 1,422 |
| 5 | The <i>Drosophila</i> Gene <i>Sulfateless</i> Modulates Autism-Like Behaviors. <i>Frontiers in Genetics</i> , 2019, 10, 574. | 1.1 | 11 |
| 6 | A Bayesian framework that integrates multi-omics data and gene networks predicts risk genes from schizophrenia GWAS data. <i>Nature Neuroscience</i> , 2019, 22, 691-699. | 7.1 | 118 |
| 7 | Structural, functional, and behavioral insights of dopamine dysfunction revealed by a deletion in <i>SLC6A3</i> . <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 3853-3862. | 3.3 | 35 |
| 8 | SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. <i>Neuron</i> , 2018, 97, 488-493. | 3.8 | 265 |
| 9 | Maternal Serotonin Levels Are Associated With Cognitive Ability and Core Symptoms in Autism Spectrum Disorder. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2018, 57, 867-875. | 0.3 | 24 |
| 10 | A Novel Human <i>CAMK2A</i> Mutation Disrupts Dendritic Morphology and Synaptic Transmission, and Causes ASD-Related Behaviors. <i>Journal of Neuroscience</i> , 2017, 37, 2216-2233. | 1.7 | 83 |
| 11 | Is there sexual dimorphism of hyperserotonemia in autism spectrum disorder?. <i>Autism Research</i> , 2017, 10, 1417-1423. | 2.1 | 24 |
| 12 | Leveraging blood serotonin as an endophenotype to identify de novo and rare variants involved in autism. <i>Molecular Autism</i> , 2017, 8, 14. | 2.6 | 50 |
| 13 | Shorter sleep duration is associated with social impairment and comorbidities in ASD. <i>Autism Research</i> , 2017, 10, 1221-1238. | 2.1 | 109 |
| 14 | The Gain-of-Function Integrin β 3 Pro33 Variant Alters the Serotonin System in the Mouse Brain. <i>Journal of Neuroscience</i> , 2017, 37, 11271-11284. | 1.7 | 22 |
| 15 | The impact of genotype calling errors on family-based studies. <i>Scientific Reports</i> , 2016, 6, 28323. | 1.6 | 12 |
| 16 | Integrin β 3 Haploinsufficiency Modulates Serotonin Transport and Antidepressant-Sensitive Behavior in Mice. <i>Neuropsychopharmacology</i> , 2015, 40, 2015-2024. | 2.8 | 26 |
| 17 | Rare Autism-Associated Variants Implicate Syntaxin 1 (STX1 R26Q) Phosphorylation and the Dopamine Transporter (hDAT R51W) in Dopamine Neurotransmission and Behaviors. <i>EBioMedicine</i> , 2015, 2, 135-146. | 2.7 | 70 |
| 18 | Consensus Genotyper for Exome Sequencing (CGES): improving the quality of exome variant genotypes. <i>Bioinformatics</i> , 2015, 31, 187-193. | 1.8 | 18 |

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|----|--|------|-----------|
| 19 | Zn ²⁺ reverses functional deficits in a de novo dopamine transporter variant associated with autism spectrum disorder. <i>Molecular Autism</i> , 2015, 6, 8. | 2.6 | 19 |
| 20 | A haplotype-based framework for group-wise transmission/disequilibrium tests for rare variant association analysis. <i>Bioinformatics</i> , 2015, 31, 1452-1459. | 1.8 | 14 |
| 21 | Analysis of <i>CHRNA7</i> rare variants in autism spectrum disorder susceptibility. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 715-723. | 0.7 | 41 |
| 22 | Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233. | 3.8 | 1,219 |
| 23 | Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. <i>Human Genetics</i> , 2015, 134, 191-201. | 1.8 | 20 |
| 24 | A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. <i>Biological Psychiatry</i> , 2015, 77, 775-784. | 0.7 | 133 |
| 25 | SLC6A3 coding variant Ala559Val found in two autism probands alters dopamine transporter function and trafficking. <i>Translational Psychiatry</i> , 2014, 4, e464-e464. | 2.4 | 108 |
| 26 | The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. <i>Molecular Autism</i> , 2014, 5, 34. | 2.6 | 31 |
| 27 | Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2014, 94, 677-694. | 2.6 | 819 |
| 28 | Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215. | 13.7 | 2,254 |
| 29 | Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15q11.2, Specifically Breakpoints 1 to 2. <i>Autism Research</i> , 2014, 7, 355-362. | 2.1 | 59 |
| 30 | Pro32Pro33 Mutations in the Integrin $\beta 3$ PSI Domain Result in $\beta 3$ Priming and Enhanced Adhesion: Reversal of the Hypercoagulability Phenotype by the Src Inhibitor SKI-606. <i>Molecular Pharmacology</i> , 2014, 85, 921-931. | 1.0 | 7 |
| 31 | A framework for the interpretation of de novo mutation in human disease. <i>Nature Genetics</i> , 2014, 46, 944-950. | 9.4 | 943 |
| 32 | Intellectual Disability Is Associated with Increased Runs of Homozygosity in Simplex Autism. <i>American Journal of Human Genetics</i> , 2013, 93, 103-109. | 2.6 | 63 |
| 33 | Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. <i>Biological Psychiatry</i> , 2013, 74, 576-584. | 0.7 | 70 |
| 34 | Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013, 45, 984-994. | 9.4 | 2,067 |
| 35 | Genetic background modulates phenotypes of serotonin transporter Ala56 knock-in mice. <i>Molecular Autism</i> , 2013, 4, 35. | 2.6 | 35 |
| 36 | Rare coding variants of the adenosine A3 receptor are increased in autism: on the trail of the serotonin transporter regulome. <i>Molecular Autism</i> , 2013, 4, 28. | 2.6 | 23 |

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|----|---|------|-----------|
| 37 | De novo mutation in the dopamine transporter gene associates dopamine dysfunction with autism spectrum disorder. <i>Molecular Psychiatry</i> , 2013, 18, 1315-1323. | 4.1 | 181 |
| 38 | Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. <i>Genomics</i> , 2013, 102, 270-277. | 1.3 | 13 |
| 39 | Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. <i>Neuron</i> , 2013, 77, 235-242. | 3.8 | 242 |
| 40 | Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. <i>PLoS Genetics</i> , 2013, 9, e1003671. | 1.5 | 253 |
| 41 | Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. <i>PLoS Genetics</i> , 2013, 9, e1003443. | 1.5 | 133 |
| 42 | <i>Drosophila melanogaster</i> : a novel animal model for the behavioral characterization of autism-associated mutations in the dopamine transporter gene. <i>Molecular Psychiatry</i> , 2013, 18, 1235-1235. | 4.1 | 9 |
| 43 | Individual common variants exert weak effects on the risk for autism spectrum disorders. <i>Human Molecular Genetics</i> , 2012, 21, 4781-4792. | 1.4 | 334 |
| 44 | A Multisite Study of the Clinical Diagnosis of Different Autism Spectrum Disorders. <i>Archives of General Psychiatry</i> , 2012, 69, 306. | 13.8 | 385 |
| 45 | A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 7974-7981. | 3.3 | 118 |
| 46 | A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. <i>Human Genetics</i> , 2012, 131, 565-579. | 1.8 | 180 |
| 47 | Autism gene variant causes hyperserotonemia, serotonin receptor hypersensitivity, social impairment and repetitive behavior. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 5469-5474. | 3.3 | 278 |
| 48 | 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. <i>Molecular Autism</i> , 2012, 3, 3. | 2.6 | 38 |
| 49 | Patterns and rates of exonic de novo mutations in autism spectrum disorders. <i>Nature</i> , 2012, 485, 242-245. | 13.7 | 1,597 |
| 50 | Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9. | 2.6 | 357 |
| 51 | Accuracy of phenotyping children with autism based on parent report: what specifically do we gain phenotyping "rapidly"? <i>Autism Research</i> , 2012, 5, 31-38. | 2.1 | 17 |
| 52 | Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. <i>Neuron</i> , 2011, 70, 863-885. | 3.8 | 1,146 |
| 53 | Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. <i>European Journal of Human Genetics</i> , 2011, 19, 1082-1089. | 1.4 | 39 |
| 54 | Maternal transmission of a rare GABRB3 signal peptide variant is associated with autism. <i>Molecular Psychiatry</i> , 2011, 16, 86-96. | 4.1 | 106 |

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|----|---|------|-----------|
| 55 | Genetic analysis of biological pathway data through genomic randomization. <i>Human Genetics</i> , 2011, 129, 563-571. | 1.8 | 50 |
| 56 | Association of oxytocin receptor (OXTR) gene variants with multiple phenotype domains of autism spectrum disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2011, 3, 101-112. | 1.5 | 148 |
| 57 | Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. <i>Journal of Neurodevelopmental Disorders</i> , 2011, 3, 113-123. | 1.5 | 22 |
| 58 | Parent-of-origin effects of the serotonin transporter gene associated with autism. , 2011, 156, 139-144. | | 41 |
| 59 | Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. <i>Human Molecular Genetics</i> , 2011, 20, 4360-4370. | 1.4 | 101 |
| 60 | Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. <i>Journal of Medical Genetics</i> , 2011, 48, 48-54. | 1.5 | 94 |
| 61 | Colocalization and Regulated Physical Association of Presynaptic Serotonin Transporters with Adenosine Receptors. <i>Molecular Pharmacology</i> , 2011, 80, 458-465. | 1.0 | 30 |
| 62 | Association of <i>MET</i> with social and communication phenotypes in individuals with autism spectrum disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 438-446. | 1.1 | 49 |
| 63 | Functional impact of global rare copy number variation in autism spectrum disorders. <i>Nature</i> , 2010, 466, 368-372. | 13.7 | 1,803 |
| 64 | A genome-wide scan for common alleles affecting risk for autism. <i>Human Molecular Genetics</i> , 2010, 19, 4072-4082. | 1.4 | 538 |
| 65 | Association and Mutation Analyses of 16p11.2 Autism Candidate Genes. <i>PLoS ONE</i> , 2009, 4, e4582. | 1.1 | 80 |
| 66 | Genome-Wide Analyses of Exonic Copy Number Variants in a Family-Based Study Point to Novel Autism Susceptibility Genes. <i>PLoS Genetics</i> , 2009, 5, e1000536. | 1.5 | 374 |
| 67 | Enhanced activity of human serotonin transporter variants associated with autism. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2009, 364, 163-173. | 1.8 | 120 |
| 68 | Fine mapping and association studies in a candidate region for autism on chromosome 2q31-q32. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 535-544. | 1.1 | 12 |
| 69 | Modeling rare gene variation to gain insight into the oldest biomarker in autism: construction of the serotonin transporter Gly56Ala knock-in mouse. <i>Journal of Neurodevelopmental Disorders</i> , 2009, 1, 158-171. | 1.5 | 43 |
| 70 | Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. <i>Nature</i> , 2009, 459, 569-573. | 13.7 | 1,270 |
| 71 | Common genetic variants on 5p14.1 associate with autism spectrum disorders. <i>Nature</i> , 2009, 459, 528-533. | 13.7 | 912 |
| 72 | A genome-wide linkage and association scan reveals novel loci for autism. <i>Nature</i> , 2009, 461, 802-808. | 13.7 | 570 |

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|----|--|------|-----------|
| 73 | Microduplications of 16p11.2 are associated with schizophrenia. <i>Nature Genetics</i> , 2009, 41, 1223-1227. | 9.4 | 646 |
| 74 | Distinct Genetic Risk Based on Association of <i>MET</i> in Families With Co-occurring Autism and Gastrointestinal Conditions. <i>Pediatrics</i> , 2009, 123, 1018-1024. | 1.0 | 141 |
| 75 | Our vision for Autism Research. <i>Autism Research</i> , 2008, 1, 71-72. | 2.1 | 3 |
| 76 | Genetic evidence implicating multiple genes in the MET receptor tyrosine kinase pathway in autism spectrum disorder. <i>Autism Research</i> , 2008, 1, 159-168. | 2.1 | 143 |
| 77 | Heterogeneity and the design of genetic studies in autism. <i>Autism Research</i> , 2008, 1, 205-206. | 2.1 | 3 |
| 78 | Affiliative Behaviors and Beyond: It's the Phenotype, Stupid. <i>Biological Psychiatry</i> , 2008, 63, 909-910. | 0.7 | 2 |
| 79 | Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699. | 13.9 | 663 |
| 80 | Insights into the Pathogenesis of Autism. <i>Science</i> , 2008, 321, 208-209. | 6.0 | 53 |
| 81 | Molecular Genetics of the Platelet Serotonin System in First-Degree Relatives of Patients with Autism. <i>Neuropsychopharmacology</i> , 2008, 33, 353-360. | 2.8 | 57 |
| 82 | Contribution of SHANK3 Mutations to Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2007, 81, 1289-1297. | 2.6 | 604 |
| 83 | Strong Association of De Novo Copy Number Mutations with Autism. <i>Science</i> , 2007, 316, 445-449. | 6.0 | 2,497 |
| 84 | Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007, 39, 319-328. | 9.4 | 1,272 |
| 85 | 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. <i>Neuroscience Letters</i> , 2006, 394, 74-78. | 1.0 | 14 |
| 86 | Variation in ITGB3 is associated with whole-blood serotonin level and autism susceptibility. <i>European Journal of Human Genetics</i> , 2006, 14, 923-931. | 1.4 | 82 |
| 87 | Lack of Association Between Autism and <i>SLC25A12</i> . <i>American Journal of Psychiatry</i> , 2006, 163, 929-931. | 4.0 | 36 |
| 88 | A genetic variant that disrupts MET transcription is associated with autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 16834-16839. | 3.3 | 389 |
| 89 | Analysis of the RELN gene as a genetic risk factor for autism. <i>Molecular Psychiatry</i> , 2005, 10, 563-571. | 4.1 | 181 |
| 90 | Genome-wide and Ordered-Subset linkage analyses provide support for autism loci on 17q and 19p with evidence of phenotypic and interlocus genetic correlates. <i>BMC Medical Genetics</i> , 2005, 6, 1. | 2.1 | 130 |

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|-----|---|-----|-----------|
| 91 | 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 |
| 92 | 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 |
| 93 | 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 |
| 94 | Autosomal Dominant Lateral Temporal Epilepsy: Two Families with Novel Mutations in the LGI1 Gene. Epilepsia, 2004, 45, 218-222. | 2.6 | 39 |
| 95 | Examination of NRCAM, LRRN3, KIAA0716, and LAMB1 as autism candidate genes. BMC Medical Genetics, 2004, 5, 12. | 2.1 | 56 |
| 96 | 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 |
| 97 | A linkage disequilibrium map of the 1-Mb 15q12 GABA receptor subunit cluster and association to autism. American Journal of Medical Genetics Part A, 2004, 131B, 51-59. | 2.4 | 135 |
| 98 | Autism and 15q11-q13 disorders: Behavioral, genetic, and pathophysiological issues. Mental Retardation and Developmental Disabilities Research Reviews, 2004, 10, 284-291. | 3.5 | 112 |
| 99 | Analysis of the autism chromosome 2 linkage region: GAD1 and other candidate genes. Neuroscience Letters, 2004, 372, 209-214. | 1.0 | 32 |
| 100 | Partial duplication of the APBA2 gene in chromosome 15q13 corresponds to duplicon structures. BMC Genomics, 2003, 4, 15. | 1.2 | 20 |
| 101 | Defining the autism minimum candidate gene region on chromosome 7. American Journal of Medical Genetics Part A, 2003, 117B, 90-96. | 2.4 | 32 |
| 102 | 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. | 1.0 | 14 |
| 103 | Dense linkage disequilibrium mapping in the 15q11-q13 maternal expression domain yields evidence for association in autism. Molecular Psychiatry, 2003, 8, 624-634. | 4.1 | 60 |
| 104 | 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 |
| 105 | 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 |
| 106 | Linkage Disequilibrium at the Angelman Syndrome Gene UBE3A in Autism Families. Genomics, 2001, 77, 105-113. | 1.3 | 154 |
| 107 | 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 |
| 108 | Partial and generalized epilepsy with febrile seizures plus and a novel SCN1A mutation. Neurology, 2001, 57, 2265-2272. | 1.5 | 193 |

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|-----|--|-----|-----------|
| 109 | Appetitive behavior, compulsivity, and neurochemistry in Prader-Willi syndrome. <i>Mental Retardation and Developmental Disabilities Research Reviews</i> , 2000, 6, 125-130. | 3.5 | 75 |
| 110 | The spectrum of mutations in UBE3A causing Angelman syndrome. <i>Human Molecular Genetics</i> , 1999, 8, 129-135. | 1.4 | 150 |
| 111 | Integrated YAC Contig Map of the Prader-Willi/Angelman Region on Chromosome 15q11-q13 with Average STS Spacing of 35 kb. <i>Genome Research</i> , 1998, 8, 146-157. | 2.4 | 72 |
| 112 | Neuronally-expressed necdin gene: an imprinted candidate gene in Prader-Willi syndrome. <i>Lancet</i> , The, 1997, 350, 1520-1521. | 6.3 | 53 |
| 113 | Sequencing and Functional Analysis of the SNRPN Promoter: In Vitro Methylation Abolishes Promoter Activity. <i>Genome Research</i> , 1997, 7, 642-648. | 2.4 | 16 |
| 114 | The E6-AP Ubiquitin-Protein Ligase (<i>UBE3A</i>) Gene Is Localized within a Narrowed Angelman Syndrome Critical Region. <i>Genome Research</i> , 1997, 7, 368-377. | 2.4 | 106 |
| 115 | De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (<i>UBE3A</i>) in Angelman syndrome. <i>Nature Genetics</i> , 1997, 15, 74-77. | 9.4 | 801 |
| 116 | Imprinted expression of the murine Angelman syndrome gene, <i>Ube3a</i> , in hippocampal and Purkinje neurons. <i>Nature Genetics</i> , 1997, 17, 75-78. | 9.4 | 466 |
| 117 | Angelman syndrome in an inbred family. <i>Human Genetics</i> , 1996, 97, 294-298. | 1.8 | 5 |
| 118 | Detection of imprinting mutations in Angelman syndrome using a probe for exon 1 of SNRPN. <i>American Journal of Medical Genetics Part A</i> , 1996, 63, 414-415. | 2.4 | 13 |
| 119 | Validation studies of SNRPN methylation as a diagnostic test for Prader-Willi syndrome. , 1996, 66, 77-80. | | 87 |
| 120 | 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 |
| 121 | Identification of a novel paternally expressed transcript adjacent to snRPN in the Prader-Willi syndrome critical region.. <i>Genome Research</i> , 1996, 6, 742-746. | 2.4 | 23 |
| 122 | Mouse/human sequence divergence in a region with a paternal-specific methylation imprint at the human H19 locus. <i>Human Molecular Genetics</i> , 1996, 5, 1155-1161. | 1.4 | 85 |
| 123 | Tissue-specific and allele-specific replication timing control in the imprinted human Prader-Willi syndrome region.. <i>Genes and Development</i> , 1995, 9, 808-820. | 2.7 | 67 |
| 124 | Imprinting analysis of three genes in the Prader-Willi/Angelman region: SNRPN, E6-associated protein, and PAR-2 (D15S225E). <i>Human Molecular Genetics</i> , 1994, 3, 309-315. | 1.4 | 125 |
| 125 | Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. <i>Nature Genetics</i> , 1994, 8, 52-58. | 9.4 | 418 |
| 126 | Tissue specific expression of FMR1 provides evidence for a functional role in fragile X syndrome. <i>Nature Genetics</i> , 1993, 3, 36-43. | 9.4 | 358 |

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|-----|--|------|-----------|
| 127 | Human and murine FMR-1: alternative splicing and translational initiation downstream of the CCG repeat. <i>Nature Genetics</i> , 1993, 4, 244-251. | 9.4 | 247 |
| 128 | A complete YAC contig of the Prader-Willi/Angelman chromosome region (15q11-q13) and refined localization of the SNRPN gene. <i>Genomics</i> , 1993, 18, 546-552. | 1.3 | 100 |
| 129 | DNA methylation represses FMR-1 transcription in fragile X syndrome. <i>Human Molecular Genetics</i> , 1992, 1, 397-400. | 1.4 | 674 |
| 130 | PCR amplification and analysis of yeast artificial chromosomes. <i>Genomics</i> , 1992, 13, 1303-1306. | 1.3 | 11 |
| 131 | Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. <i>American Journal of Medical Genetics Part A</i> , 1992, 43, 237-243. | 2.4 | 82 |
| 132 | Variation of the CCG repeat at the fragile X site results in genetic instability: Resolution of the Sherman paradox. <i>Cell</i> , 1991, 67, 1047-1058. | 13.5 | 2,007 |
| 133 | Identification of a gene (FMR-1) containing a CCG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. <i>Cell</i> , 1991, 65, 905-914. | 13.5 | 3,285 |
| 134 | Strategy for molecular cloning of the fragile X site DNA. <i>American Journal of Medical Genetics Part A</i> , 1988, 30, 613-623. | 2.4 | 12 |