Matthew W State

List of Publications by Year
in descending order

104 papers	31,692 citations	10986 71 h-index	27406 106 g-index
120 all docs	120 docs citations	120 times ranked	31030 citing authors

#	Article	IF	Citations
1	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
2	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	27.8	2,188
3	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. Nature, 2012, 485, 237-241.	27.8	1,863
4	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
5	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
6	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	8.1	1,219
7	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	8.1	1,146
8	Common genetic variants on 5p14.1 associate with autism spectrum disorders. Nature, 2009, 459, 528-533.	27.8	912
9	Sequence Variants in SLITRK1 Are Associated with Tourette's Syndrome. Science, 2005, 310, 317-320.	12.6	878
10	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. Science, 2018, 359, 693-697.	12.6	851
11	Coexpression Networks Implicate Human Midfetal Deep Cortical Projection Neurons in the Pathogenesis of Autism. Cell, 2013, 155, 997-1007.	28.9	825
12	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. Science, 2018, 362, .	12.6	805
13	Autism spectrum disorder. Nature Reviews Disease Primers, 2020, 6, 5.	30.5	746
14	Rare independent mutations in renal salt handling genes contribute to blood pressure variation. Nature Genetics, 2008, 40, 592-599.	21.4	728
15	Genomic Analysis of Non- NF2 Meningiomas Reveals Mutations in TRAF7 , KLF4 , AKT1 , and SMO. Science, 2013, 339, 1077-1080.	12.6	714
16	Comprehensive functional genomic resource and integrative model for the human brain. Science, 2018, 362, .	12.6	618
17	Practice Parameter for the Assessment and Treatment of Children and Adolescents With Autism Spectrum Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 237-257.	0.5	558
18	Functional and Evolutionary Insights into Human Brain Development through Global Transcriptome Analysis. Neuron, 2009, 62, 494-509.	8.1	555

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19	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. Science, 2018, 362, .	12.6	516
20	Molecular Cytogenetic Analysis and Resequencing of Contactin Associated Protein-Like 2 in Autism Spectrum Disorders. American Journal of Human Genetics, 2008, 82, 165-173.	6.2	494
21	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	27.8	457
22	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
23	Gene hunting in autism spectrum disorder: on the path to precision medicine. Lancet Neurology, The, 2015, 14, 1109-1120.	10.2	374
24	The PsychENCODE project. Nature Neuroscience, 2015, 18, 1707-1712.	14.8	371
25	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	4.9	357
26	The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. Nature Communications, 2015, 6, 6404.	12.8	316
27	L-Histidine Decarboxylase and Tourette's Syndrome. New England Journal of Medicine, 2010, 362, 1901-1908.	27.0	304
28	The conundrums of understanding genetic risks for autism spectrum disorders. Nature Neuroscience, 2011, 14, 1499-1506.	14.8	287
29	Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy. Science, 2012, 338, 394-397.	12.6	272
30	Genome-wide association study of intracranial aneurysm identifies three new risk loci. Nature Genetics, 2010, 42, 420-425.	21.4	262
31	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	3.5	253
32	Susceptibility loci for intracranial aneurysm in European and Japanese populations. Nature Genetics, 2008, 40, 1472-1477.	21.4	247
33	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	8.1	242
34	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	21.4	235
35	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	12.6	234
36	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896.	4.5	229

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37	Transcriptome and epigenome landscape of human cortical development modeled in organoids. Science, 2018, 362, .	12.6	220
38	Histidine Decarboxylase Deficiency Causes Tourette Syndrome: Parallel Findings in Humans and Mice. Neuron, 2014, 81, 77-90.	8.1	212
39	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
40	Molecular and cellular reorganization of neural circuits in the human lineage. Science, 2017, 358, 1027-1032.	12.6	192
41	Recent Advances in the Genetics of Autism. Biological Psychiatry, 2007, 61, 429-437.	1.3	170
42	Estrogens Suppress a Behavioral Phenotype in Zebrafish Mutants of the Autism Risk Gene, CNTNAP2. Neuron, 2016, 89, 725-733.	8.1	170
43	Autism and autism spectrum disorders: diagnostic issues for the coming decade. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2009, 50, 108-115.	5.2	169
44	Rare Copy Number Variants in Tourette Syndrome Disrupt Genes in Histaminergic Pathways and Overlap with Autism. Biological Psychiatry, 2012, 71, 392-402.	1.3	167
45	Disruption of Contactin 4 (CNTN4) Results in Developmental Delay and Other Features of 3p Deletion Syndrome. American Journal of Human Genetics, 2004, 74, 1286-1293.	6.2	162
46	Genome-wide Transcriptome Profiling Reveals the Functional Impact of Rare De Novo and Recurrent CNVs in Autism Spectrum Disorders. American Journal of Human Genetics, 2012, 91, 38-55.	6.2	160
47	The Genetics of Child Psychiatric Disorders: Focus on Autism and Tourette Syndrome. Neuron, 2010, 68, 254-269.	8.1	156
48	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	8.1	155
49	The Autism Sequencing Consortium: Large-Scale, High-Throughput Sequencing in Autism Spectrum Disorders. Neuron, 2012, 76, 1052-1056.	8.1	153
50	De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. Cell Reports, 2014, 9, 16-23.	6.4	151
51	The Emerging Biology of Autism Spectrum Disorders. Science, 2012, 337, 1301-1303.	12.6	147
52	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	27.8	145
53	Psychiatric Disorders: Diagnosis to Therapy. Cell, 2014, 157, 201-214.	28.9	140
54	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136

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55	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.	1.3	133
56	Autism genetics: strategies, challenges, and opportunities. Autism Research, 2008, 1, 4-17.	3.8	123
57	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	14.8	122
58	Autism spectrum disorders: from genes to neurobiology. Current Opinion in Neurobiology, 2015, 30, 92-99.	4.2	121
59	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	27.0	119
60	Recent developments in the genetics of autism spectrum disorders. Current Opinion in Genetics and Development, 2013, 23, 310-315.	3.3	118
61	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. Molecular Autism, 2014, 5, 22.	4.9	111
62	The Psychiatric Cell Map Initiative: A Convergent Systems Biological Approach to Illuminating Key Molecular Pathways in Neuropsychiatric Disorders. Cell, 2018, 174, 505-520.	28.9	108
63	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.	2.9	101
64	The developmental transcriptome of the human brain. Current Opinion in Neurology, 2014, 27, 149-156.	3.6	100
65	Lost in Translation: Traversing the Complex Path from Genomics to Therapeutics in Autism Spectrum Disorder. Neuron, 2018, 100, 406-423.	8.1	98
66	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	6.4	91
67	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. Cell Reports, 2020, 31, 107489.	6.4	91
68	Syndromic patent ductus arteriosus: Evidence for haploinsufficient TFAP2B mutations and identification of a linked sleep disorder. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 2975-2979.	7.1	85
69	Cerebrospinal fluid levels of oxytocin in Praderâ€“Willi syndrome: a preliminary report. Biological Psychiatry, 1998, 44, 1349-1352.	1.3	84
70	Developmentally regulated and evolutionarily conserved expression of SLITRK1 in brain circuits implicated in Tourette syndrome. Journal of Comparative Neurology, 2009, 513, 21-37.	1.6	84
71	Neonatal Tbr1 Dosage Controls Cortical Layer 6 Connectivity. Neuron, 2018, 100, 831-845.e7.	8.1	83
72	Genomics, convergent neuroscience and progress in understanding autism spectrum disorder. Nature Reviews Neuroscience, 2022, 23, 323-341.	10.2	81

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73	Mapping a Mendelian Form of Intracranial Aneurysm to 1p34.3-p36.13. American Journal of Human Genetics, 2005, 76, 172-179.	6.2	80
74	Recent advances in Tourette syndrome. Current Opinion in Neurology, 2011, 24, 119-125.	3.6	77
75	Epigenetic abnormalities associated with a chromosome 18(q21-q22) inversion and a Gilles de la Tourette syndrome phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 4684-4689.	7.1	73
76	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	1.3	70
77	The inheritance of Tourette Disorder: A review. Journal of Obsessive-Compulsive and Related Disorders, 2014, 3, 380-385.	1.5	70
78	Autism Spectrum Disorder Genetics and the Search for Pathological Mechanisms. American Journal of Psychiatry, 2021, 178, 30-38.	7.2	70
79	A Chromatin Accessibility Atlas of the Developing Human Telencephalon. Cell, 2020, 182, 754-769.e18.	28.9	69
80	Effects of Electroconvulsive Therapy in Adolescents with Severe Endogenous Depression Resistant to Pharmacotherapy. Biological Psychiatry, 1998, 43, 335-338.	1.3	66
81	Intellectual Disability Is Associated with Increased Runs of Homozygosity in Simplex Autism. American Journal of Human Genetics, 2013, 93, 103-109.	6.2	63
82	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.	3.8	59
83	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. European Journal of Human Genetics, 2015, 23, 165-172.	2.8	57
84	Parallel inÂvivo analysis of large-effect autism genes implicates cortical neurogenesis and estrogen in risk and resilience. Neuron, 2021, 109, 788-804.e8.	8.1	54
85	Prader-Willi Syndrome. American Journal of Psychiatry, 1998, 155, 1265-1273.	7.2	53
86	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. Nature Communications, 2021, 12, 3968.	12.8	48
87	No Evidence for Association of Autism with Rare Heterozygous Point Mutations in Contactin-Associated Protein-Like 2 (CNTNAP2), or in Other Contactin-Associated Proteins or Contactins. PLoS Genetics, 2015, 11, e1004852.	3.5	47
88	The Genetic Etiology of Tourette Syndrome: Large-Scale Collaborative Efforts on the Precipice of Discovery. Frontiers in Neuroscience, 2016, 10, 351.	2.8	45
89	The genetics of Tourette disorder. Current Opinion in Genetics and Development, 2011, 21, 302-309.	3.3	41
90	The Tourette International Collaborative Genetics (TIC Genetics) study, finding the genes causing Tourette syndrome: objectives and methods. European Child and Adolescent Psychiatry, 2015, 24, 141-151.	4.7	41

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91	Recent challenges to the psychiatric diagnostic nosology: a focus on the genetics and genomics of neurodevelopmental disorders. International Journal of Epidemiology, 2014, 43, 465-475.	1.9	39
92	Autism risk gene POGZ promotes chromatin accessibility and expression of clustered synaptic genes. Cell Reports, 2021, 37, 110089.	6.4	38
93	Pre- and perinatal complications in relation to Tourette syndrome and co-occurring obsessive-compulsive disorder and attention-deficit/hyperactivity disorder. Journal of Psychiatric Research, 2016, 82, 126-135.	3.1	36
94	Enhancing WNT Signaling Restores Cortical Neuronal Spine Maturation and Synaptogenesis in Tbr1 Mutants. Cell Reports, 2020, 31, 107495.	6.4	32
95	Leveraging large genomic datasets to illuminate the pathobiology of autism spectrum disorders. Neuropsychopharmacology, 2021, 46, 55-69.	5.4	31
96	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. Molecular Autism, 2014, 5, 31.	4.9	27
97	Neurodevelopmental disorder risk gene DYRK1A is required for ciliogenesis and brain size in Xenopus embryos. Development (Cambridge), 2020, 147, .	2.5	27
98	Tourette disorder and other tic disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 343-354.	1.8	24
99	Elevated common variant genetic risk for tourette syndrome in a densely-affected pedigree. Molecular Psychiatry, 2021, 26, 7522-7529.	7.9	8
100	The Erosion of Phenotypic Specificity in Psychiatric Genetics: Emerging Lessons from CNTNAP2. Biological Psychiatry, 2011, 69, 816-817.	1.3	5
101	Leveraging Genetics and Genomics to Define the Causes of Mental Illness. Biological Psychiatry, 2015, 77, 3-5.	1.3	5
102	A surprising METamorphosis: Autism genetics finds a common functional variant. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 16621-16622.	7.1	3
103	Constance E. Lieber, Theodore R. Stanley, and the Enduring Impact of Philanthropy on Psychiatry Research. Biological Psychiatry, 2016, 80, 84-86.	1.3	2
104	Reply to 3p deletion and (skewed) literature review. American Journal of Medical Genetics, Part A, 2010, 152A, 1060-1060.	1.2	0