

# Matthew W State

## List of Publications by Year in descending order

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Version: 2024-02-01

104  
papers

31,692  
citations

10979

71  
h-index

27389

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. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
2	The contribution of de novo coding mutations to autism spectrum disorder. <i>Nature</i> , 2014, 515, 216-221.	13.7	2,188
3	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. <i>Nature</i> , 2012, 485, 237-241.	13.7	1,863
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	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007, 39, 319-328.	9.4	1,272
6	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
7	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
8	Common genetic variants on 5p14.1 associate with autism spectrum disorders. <i>Nature</i> , 2009, 459, 528-533.	13.7	912
9	Sequence Variants in SLITRK1 Are Associated with Tourette's Syndrome. <i>Science</i> , 2005, 310, 317-320.	6.0	878
10	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. <i>Science</i> , 2018, 359, 693-697.	6.0	851
11	Coexpression Networks Implicate Human Midfetal Deep Cortical Projection Neurons in the Pathogenesis of Autism. <i>Cell</i> , 2013, 155, 997-1007.	13.5	825
12	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. <i>Science</i> , 2018, 362, .	6.0	805
13	Autism spectrum disorder. <i>Nature Reviews Disease Primers</i> , 2020, 6, 5.	18.1	746
14	Rare independent mutations in renal salt handling genes contribute to blood pressure variation. <i>Nature Genetics</i> , 2008, 40, 592-599.	9.4	728
15	Genomic Analysis of Non- <i>NF2</i> Meningiomas Reveals Mutations in <i>TRAF7</i> , <i>KLF4</i> , <i>AKT1</i> , and <i>SMO</i> . <i>Science</i> , 2013, 339, 1077-1080.	6.0	714
16	Comprehensive functional genomic resource and integrative model for the human brain. <i>Science</i> , 2018, 362, .	6.0	618
17	Practice Parameter for the Assessment and Treatment of Children and Adolescents With Autism Spectrum Disorder. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2014, 53, 237-257.	0.3	558
18	Functional and Evolutionary Insights into Human Brain Development through Global Transcriptome Analysis. <i>Neuron</i> , 2009, 62, 494-509.	3.8	555

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19	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. <i>Science</i> , 2018, 362, .	6.0	516
20	Molecular Cytogenetic Analysis and Resequencing of Contactin Associated Protein-Like 2 in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2008, 82, 165-173.	2.6	494
21	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. <i>Nature</i> , 2010, 467, 207-210.	13.7	457
22	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
23	Gene hunting in autism spectrum disorder: on the path to precision medicine. <i>Lancet Neurology</i> , The, 2015, 14, 1109-1120.	4.9	374
24	The PsychENCODE project. <i>Nature Neuroscience</i> , 2015, 18, 1707-1712.	7.1	371
25	Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9.	2.6	357
26	The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. <i>Nature Communications</i> , 2015, 6, 6404.	5.8	316
27	L-Histidine Decarboxylase and Tourette's Syndrome. <i>New England Journal of Medicine</i> , 2010, 362, 1901-1908.	13.9	304
28	The conundrums of understanding genetic risks for autism spectrum disorders. <i>Nature Neuroscience</i> , 2011, 14, 1499-1506.	7.1	287
29	Mutations in <i>BCKD-kinase</i> Lead to a Potentially Treatable Form of Autism with Epilepsy. <i>Science</i> , 2012, 338, 394-397.	6.0	272
30	Genome-wide association study of intracranial aneurysm identifies three new risk loci. <i>Nature Genetics</i> , 2010, 42, 420-425.	9.4	262
31	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
32	Susceptibility loci for intracranial aneurysm in European and Japanese populations. <i>Nature Genetics</i> , 2008, 40, 1472-1477.	9.4	247
33	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. <i>Neuron</i> , 2013, 77, 235-242.	3.8	242
34	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. <i>Nature Genetics</i> , 2018, 50, 727-736.	9.4	235
35	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. <i>Science</i> , 2018, 362, .	6.0	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. <i>Circulation Research</i> , 2014, 115, 884-896.	2.0	229

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37	Transcriptome and epigenome landscape of human cortical development modeled in organoids. <i>Science</i> , 2018, 362, .	6.0	220
38	Histidine Decarboxylase Deficiency Causes Tourette Syndrome: Parallel Findings in Humans and Mice. <i>Neuron</i> , 2014, 81, 77-90.	3.8	212
39	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. <i>American Journal of Human Genetics</i> , 2012, 91, 987-997.	2.6	201
40	Molecular and cellular reorganization of neural circuits in the human lineage. <i>Science</i> , 2017, 358, 1027-1032.	6.0	192
41	Recent Advances in the Genetics of Autism. <i>Biological Psychiatry</i> , 2007, 61, 429-437.	0.7	170
42	Estrogens Suppress a Behavioral Phenotype in Zebrafish Mutants of the Autism Risk Gene, CNTNAP2. <i>Neuron</i> , 2016, 89, 725-733.	3.8	170
43	Autism and autism spectrum disorders: diagnostic issues for the coming decade. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2009, 50, 108-115.	3.1	169
44	Rare Copy Number Variants in Tourette Syndrome Disrupt Genes in Histaminergic Pathways and Overlap with Autism. <i>Biological Psychiatry</i> , 2012, 71, 392-402.	0.7	167
45	Disruption of Contactin 4 (CNTN4) Results in Developmental Delay and Other Features of 3p Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2004, 74, 1286-1293.	2.6	162
46	Genome-wide Transcriptome Profiling Reveals the Functional Impact of Rare De Novo and Recurrent CNVs in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2012, 91, 38-55.	2.6	160
47	The Genetics of Child Psychiatric Disorders: Focus on Autism and Tourette Syndrome. <i>Neuron</i> , 2010, 68, 254-269.	3.8	156
48	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. <i>Neuron</i> , 2017, 94, 486-499.e9.	3.8	155
49	The Autism Sequencing Consortium: Large-Scale, High-Throughput Sequencing in Autism Spectrum Disorders. <i>Neuron</i> , 2012, 76, 1052-1056.	3.8	153
50	De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. <i>Cell Reports</i> , 2014, 9, 16-23.	2.9	151
51	The Emerging Biology of Autism Spectrum Disorders. <i>Science</i> , 2012, 337, 1301-1303.	6.0	147
52	Loss of $\beta$ -catenin function in severe autism. <i>Nature</i> , 2015, 520, 51-56.	13.7	145
53	Psychiatric Disorders: Diagnosis to Therapy. <i>Cell</i> , 2014, 157, 201-214.	13.5	140
54	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	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?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133
56	Autism genetics: strategies, challenges, and opportunities. <i>Autism Research</i> , 2008, 1, 4-17.	2.1	123
57	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. <i>Nature Neuroscience</i> , 2017, 20, 1661-1668.	7.1	122
58	Autism spectrum disorders: from genes to neurobiology. <i>Current Opinion in Neurobiology</i> , 2015, 30, 92-99.	2.0	121
59	Mutations in <i>DSTYK</i> and Dominant Urinary Tract Malformations. <i>New England Journal of Medicine</i> , 2013, 369, 621-629.	13.9	119
60	Recent developments in the genetics of autism spectrum disorders. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 310-315.	1.5	118
61	DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. <i>Molecular Autism</i> , 2014, 5, 22.	2.6	111
62	The Psychiatric Cell Map Initiative: A Convergent Systems Biological Approach to Illuminating Key Molecular Pathways in Neuropsychiatric Disorders. <i>Cell</i> , 2018, 174, 505-520.	13.5	108
63	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
64	The developmental transcriptome of the human brain. <i>Current Opinion in Neurology</i> , 2014, 27, 149-156.	1.8	100
65	Lost in Translation: Traversing the Complex Path from Genomics to Therapeutics in Autism Spectrum Disorder. <i>Neuron</i> , 2018, 100, 406-423.	3.8	98
66	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. <i>Cell Reports</i> , 2018, 24, 3441-3454.e12.	2.9	91
67	Whole-Genome and RNA Sequencing Reveal Variation and Transcriptomic Coordination in the Developing Human Prefrontal Cortex. <i>Cell Reports</i> , 2020, 31, 107489.	2.9	91
68	Syndromic patent ductus arteriosus: Evidence for haploinsufficient TFAP2B mutations and identification of a linked sleep disorder. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 2975-2979.	3.3	85
69	Cerebrospinal fluid levels of oxytocin in Prader-Willi syndrome: a preliminary report. <i>Biological Psychiatry</i> , 1998, 44, 1349-1352.	0.7	84
70	Developmentally regulated and evolutionarily conserved expression of SLITRK1 in brain circuits implicated in Tourette syndrome. <i>Journal of Comparative Neurology</i> , 2009, 513, 21-37.	0.9	84
71	Neonatal Tbr1 Dosage Controls Cortical Layer 6 Connectivity. <i>Neuron</i> , 2018, 100, 831-845.e7.	3.8	83
72	Genomics, convergent neuroscience and progress in understanding autism spectrum disorder. <i>Nature Reviews Neuroscience</i> , 2022, 23, 323-341.	4.9	81

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73	Mapping a Mendelian Form of Intracranial Aneurysm to 1p34.3-p36.13. <i>American Journal of Human Genetics</i> , 2005, 76, 172-179.	2.6	80
74	Recent advances in Tourette syndrome. <i>Current Opinion in Neurology</i> , 2011, 24, 119-125.	1.8	77
75	Epigenetic abnormalities associated with a chromosome 18(q21-q22) inversion and a Gilles de la Tourette syndrome phenotype. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 4684-4689.	3.3	73
76	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
77	The inheritance of Tourette Disorder: A review. <i>Journal of Obsessive-Compulsive and Related Disorders</i> , 2014, 3, 380-385.	0.7	70
78	Autism Spectrum Disorder Genetics and the Search for Pathological Mechanisms. <i>American Journal of Psychiatry</i> , 2021, 178, 30-38.	4.0	70
79	A Chromatin Accessibility Atlas of the Developing Human Telencephalon. <i>Cell</i> , 2020, 182, 754-769.e18.	13.5	69
80	Effects of Electroconvulsive Therapy in Adolescents with Severe Endogenous Depression Resistant to Pharmacotherapy. <i>Biological Psychiatry</i> , 1998, 43, 335-338.	0.7	66
81	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
82	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
83	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. <i>European Journal of Human Genetics</i> , 2015, 23, 165-172.	1.4	57
84	Parallel in vivo analysis of large-effect autism genes implicates cortical neurogenesis and estrogen in risk and resilience. <i>Neuron</i> , 2021, 109, 788-804.e8.	3.8	54
85	Prader-Willi Syndrome. <i>American Journal of Psychiatry</i> , 1998, 155, 1265-1273.	4.0	53
86	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. <i>Nature Communications</i> , 2021, 12, 3968.	5.8	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. <i>PLoS Genetics</i> , 2015, 11, e1004852.	1.5	47
88	The Genetic Etiology of Tourette Syndrome: Large-Scale Collaborative Efforts on the Precipice of Discovery. <i>Frontiers in Neuroscience</i> , 2016, 10, 351.	1.4	45
89	The genetics of Tourette disorder. <i>Current Opinion in Genetics and Development</i> , 2011, 21, 302-309.	1.5	41
90	The Tourette International Collaborative Genetics (TIC Genetics) study, finding the genes causing Tourette syndrome: objectives and methods. <i>European Child and Adolescent Psychiatry</i> , 2015, 24, 141-151.	2.8	41

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91	Recent challenges to the psychiatric diagnostic nosology: a focus on the genetics and genomics of neurodevelopmental disorders. <i>International Journal of Epidemiology</i> , 2014, 43, 465-475.	0.9	39
92	Autism risk gene POGZ promotes chromatin accessibility and expression of clustered synaptic genes. <i>Cell Reports</i> , 2021, 37, 110089.	2.9	38
93	Pre- and perinatal complications in relation to Tourette syndrome and co-occurring obsessive-compulsive disorder and attention-deficit/hyperactivity disorder. <i>Journal of Psychiatric Research</i> , 2016, 82, 126-135.	1.5	36
94	Enhancing WNT Signaling Restores Cortical Neuronal Spine Maturation and Synaptogenesis in Tbr1 Mutants. <i>Cell Reports</i> , 2020, 31, 107495.	2.9	32
95	Leveraging large genomic datasets to illuminate the pathobiology of autism spectrum disorders. <i>Neuropsychopharmacology</i> , 2021, 46, 55-69.	2.8	31
96	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. <i>Molecular Autism</i> , 2014, 5, 31.	2.6	27
97	Neurodevelopmental disorder risk gene <i>DYRK1A</i> is required for ciliogenesis and brain size in <i>Xenopus</i> embryos. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	27
98	Tourette disorder and other tic disorders. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 147, 343-354.	1.0	24
99	Elevated common variant genetic risk for tourette syndrome in a densely-affected pedigree. <i>Molecular Psychiatry</i> , 2021, 26, 7522-7529.	4.1	8
100	The Erosion of Phenotypic Specificity in Psychiatric Genetics: Emerging Lessons from CNTNAP2. <i>Biological Psychiatry</i> , 2011, 69, 816-817.	0.7	5
101	Leveraging Genetics and Genomics to Define the Causes of Mental Illness. <i>Biological Psychiatry</i> , 2015, 77, 3-5.	0.7	5
102	A surprising METamorphosis: Autism genetics finds a common functional variant. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 16621-16622.	3.3	3
103	Constance E. Lieber, Theodore R. Stanley, and the Enduring Impact of Philanthropy on Psychiatry Research. <i>Biological Psychiatry</i> , 2016, 80, 84-86.	0.7	2
104	Reply to 3p deletion and (skewed) literature review. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1060-1060.	0.7	0