Jeffrey L. Neul

List of Publications by Year
in descending order

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90 papers	7,219 citations	87888 38 h-index	60623 81 g-index
95 all docs	95 docs citations	95 times ranked	6550 citing authors

#	Article	IF	Citations
1	Rett syndrome: Revised diagnostic criteria and nomenclature. Annals of Neurology, 2010, 68, 944-950.	5.3	1,045
2	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269.	27.8	1,042
3	Methyl-CpG-binding protein 2(MECP2) mutation type is associated with disease severity in Rett syndrome. Journal of Medical Genetics, 2014, 51, 152-158.	3.2	246
4	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. Genetics in Medicine, 2006, 8, 784-792.	2.4	245
5	Loss of MeCP2 in aminergic neurons causes cell-autonomous defects in neurotransmitter synthesis and specific behavioral abnormalities. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21966-21971.	7.1	240
6	The Xenopus Dorsalizing Factor noggin Ventralizes Drosophila Embryos by Preventing DPP from Activating Its Receptor. Cell, 1996, 86, 607-617.	28.9	236
7	Deletion of Mecp2 in Sim1-Expressing Neurons Reveals a Critical Role for MeCP2 in Feeding Behavior, Aggression, and the Response to Stress. Neuron, 2008, 59, 947-958.	8.1	230
8	Preclinical research in Rett syndrome: setting the foundation for translational success. DMM Disease Models and Mechanisms, 2012, 5, 733-745.	2.4	183
9	A partial loss of function allele of Methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. Human Molecular Genetics, 2008, 17, 1718-1727.	2.9	173
10	Neurologic Complications Associated With Influenza A in Children During the 2003-2004 Influenza Season in Houston, Texas. Pediatrics, 2004, 114, e626-e633.	2.1	160
11	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. Nature, 2015, 521, E1-E4.	27.8	159
12	Female Mecp2+/â^' mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. Human Molecular Genetics, 2013, 22, 96-109.	2.9	158
13	Gastrointestinal and Nutritional Problems Occur Frequently Throughout Life in Girls and Women With Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 292-298.	1.8	144
14	Rett Syndrome: A Prototypical Neurodevelopmental Disorder. Neuroscientist, 2004, 10, 118-128.	3.5	135
15	Mutual antagonism between Sox10 and NFIA regulates diversification of glial lineages and glioma subtypes. Nature Neuroscience, 2014, 17, 1322-1329.	14.8	124
16	Developmental delay in Rett syndrome: data from the natural history study. Journal of Neurodevelopmental Disorders, 2014, 6, 20.	3.1	118
17	Rett syndrome diagnostic criteria: Lessons from the Natural History Study. Annals of Neurology, 2010, 68, 951-955.	5.3	111
18	Breathing challenges in Rett Syndrome: Lessons learned from humans and animal models. Respiratory Physiology and Neurobiology, 2013, 189, 280-287.	1.6	107

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19	Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. Neurology, 2019, 92, e1912-e1925.	1.1	106
20	Complexities of Rett Syndrome and MeCP2: Figure 1.. Journal of Neuroscience, 2011, 31, 7951-7959.	3.6	101
21	Spatially Restricted Activation of the SAX Receptor by SCW Modulates DPP/TKV Signaling in Drosophila Dorsalâ€“Ventral Patterning. Cell, 1998, 95, 483-494.	28.9	95
22	The relationship of Rett syndrome and MECP2 disorders to autism. Dialogues in Clinical Neuroscience, 2012, 14, 253-262.	3.7	90
23	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. ELife, 2016, 5, .	6.0	86
24	Longitudinal course of epilepsy in Rett syndrome and related disorders. Brain, 2017, 140, 306-318.	7.6	80
25	A Double-Blind, Randomized, Placebo-Controlled Clinical Study of Trofinetide in the Treatment of Rett Syndrome. Pediatric Neurology, 2017, 76, 37-46.	2.1	80
26	The Changing Face of Survival in Rett Syndrome andÂMECP2-Related Disorders. Pediatric Neurology, 2015, 53, 402-411.	2.1	79
27	MeCP2 Is Critical within HoxB1-Derived Tissues of Mice for Normal Lifespan. Journal of Neuroscience, 2011, 31, 10359-10370.	3.6	75
28	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	2.4	74
29	Pathogenesis of Lethal Cardiac Arrhythmias in Mecp2 Mutant Mice: Implication for Therapy in Rett Syndrome. Science Translational Medicine, 2011, 3, 113ra125.	12.4	72
30	The course of awake breathing disturbances across the lifespan in Rett syndrome. Brain and Development, 2018, 40, 515-529.	1.1	60
31	Insulinotropic treatments exacerbate metabolic syndrome in mice lacking MeCP2 function. Human Molecular Genetics, 2013, 22, 2626-2633.	2.9	55
32	Profiling Scoliosis in Rett Syndrome. Pediatric Research, 2010, 67, 435-439.	2.3	54
33	Rett syndrome like phenotypes in the R255X Mecp2 mutant mouse are rescued by MECP2 transgene. Human Molecular Genetics, 2015, 24, 2662-2672.	2.9	54
34	Overexpression of Methyl-CpG Binding Protein 2 Impairs T _H 1 Responses. Science Translational Medicine, 2012, 4, 163ra158.	12.4	52
35	Loss of MeCP2 in the rat models regression, impaired sociability and transcriptional deficits of Rett syndrome. Human Molecular Genetics, 2016, 25, 3284-3302.	2.9	52
36	A Study of the Treatment of Rett Syndrome With Folate and Betaine. Journal of Child Neurology, 2009, 24, 551-556.	1.4	48

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37	Improving Treatment Trial Outcomes for Rett Syndrome. Journal of Child Neurology, 2015, 30, 1743-1748.	1.4	47
38	The array of clinical phenotypes of males with mutations in Methylâ€CpG binding protein 2. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 55-67.	1.7	47
39	Scoliosis in Rett Syndrome: Progression, Comorbidities, and Predictors. Pediatric Neurology, 2017, 70, 20-25.	2.1	46
40	Age of Diagnosis in Rett Syndrome: Patterns of Recognition Among Diagnosticians and Risk Factors for Late Diagnosis. Pediatric Neurology, 2015, 52, 585-591.e2.	2.1	44
41	Behavioral profiles in Rett syndrome: Data from the natural history study. Brain and Development, 2019, 41, 123-134.	1.1	42
42	Origin of a Non-Clarkeâ€™s Column Division of the Dorsal Spinocerebellar Tract and the Role of Caudal Proprioceptive Neurons in Motor Function. Cell Reports, 2015, 13, 1258-1271.	6.4	40
43	Vitamin D Deficiency Is Prevalent in Girls and Women With Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2011, 53, 569-574.	1.8	40
44	Brief Report: MECP2 Mutations in People Without Rett Syndrome. Journal of Autism and Developmental Disorders, 2014, 44, 703-711.	2.7	37
45	Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. Clinical Genetics, 2019, 95, 575-581.	2.0	37
46	Multisystem comorbidities in classic Rett syndrome: a scoping review. BMJ Paediatrics Open, 2020, 4, e000731.	1.4	35
47	Consensus guidelines on managing Rett syndrome across the lifespan. BMJ Paediatrics Open, 2020, 4, e000717.	1.4	35
48	Pubertal Development in Rett Syndrome Deviates From Typical Females. Pediatric Neurology, 2014, 51, 769-775.	2.1	32
49	Progressive Changes in a Distributed Neural Circuit Underlie Breathing Abnormalities in Mice Lacking MeCP2. Journal of Neuroscience, 2016, 36, 5572-5586.	3.6	30
50	Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome. Contemporary Clinical Trials, 2022, 114, 106704.	1.8	30
51	Hand stereotypies. Neurology, 2019, 92, e2594-e2603.	1.1	29
52	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. Pediatric Neurology, 2016, 58, 67-74.	2.1	25
53	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. Autism Research, 2018, 11, 212-222.	3.8	25
54	Comparison of Core Features in Four Developmental Encephalopathies in the Rett Natural History Study. Annals of Neurology, 2020, 88, 396-406.	5.3	25

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55	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. Frontiers in Integrative Neuroscience, 2020, 14, 7.	2.1	24
56	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. Journal of Neurodevelopmental Disorders, 2021, 13, 40.	3.1	22
57	A Psychometric Evaluation of the Motor-Behavioral Assessment Scale for Use as an Outcome Measure in Rett Syndrome Clinical Trials. American Journal on Intellectual and Developmental Disabilities, 2020, 125, 493-509.	1.6	22
58	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
59	When Rett syndrome is due to genes other than MECP2. Translational Science of Rare Diseases, 2018, 3, 49-53.	1.5	19
60	Pharmacological read-through of R294X Mecp2 in a novel mouse model of Rett syndrome. Human Molecular Genetics, 2020, 29, 2461-2470.	2.9	19
61	Rett Syndrome and MECP2-Related Disorders. , 2011, , 776-800.		18
62	Treatment of cardiac arrhythmias in Rett Syndrome with sodium channel blocking antiepileptic drugs. DMM Disease Models and Mechanisms, 2015, 8, 363-71.	2.4	15
63	Methyl-CpG binding-protein 2 function in cholinergic neurons mediates cardiac arrhythmogenesis. Human Molecular Genetics, 2016, 25, ddw326.	2.9	15
64	Safety and efficacy of genetic <scp>MECP2</scp> supplementation in the <scp>R294X</scp> mouse model of Rett syndrome. Genes, Brain and Behavior, 2022, 21, e12739.	2.2	15
65	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study. Journal of Neurodevelopmental Disorders, 2022, 14, 31.	3.1	15
66	Towards a consensus on developmental regression. Neuroscience and Biobehavioral Reviews, 2019, 107, 3-5.	6.1	14
67	Multisite Study of Evoked Potentials in Rett Syndrome. Annals of Neurology, 2021, 89, 790-802.	5.3	14
68	Loss of MeCP2 Causes Urological Dysfunction and Contributes to Death by Kidney Failure in Mouse Models of Rett Syndrome. PLoS ONE, 2016, 11, e0165550.	2.5	13
69	Assessment of Caregiver Inventory for Rett Syndrome. Journal of Autism and Developmental Disorders, 2017, 47, 1102-1112.	2.7	13
70	Loss of MeCP2 Function Across Several Neuronal Populations Impairs Breathing Response to Acute Hypoxia. Frontiers in Neurology, 2020, 11, 593554.	2.4	13
71	Phenotypic features in <scp>MECP2</scp> duplication syndrome: Effects of age. American Journal of Medical Genetics, Part A, 2021, 185, 362-369.	1.2	13
72	Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 3.	3.1	12

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73	Unfolding neurodevelopmental disorders: The mystery of developing connections. Nature Medicine, 2011, 17, 1353-1355.	30.7	11
74	Low Bone Mineral Mass Is Associated With Decreased Bone Formation and Diet in Girls With Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2014, 59, 386-392.	1.8	11
75	Detection of neurophysiological features in female R255X MeCP2 mutation mice. Neurobiology of Disease, 2020, 145, 105083.	4.4	11
76	Biliary Tract Disease in Girls and Young Women With Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, 799-805.	1.8	10
77	Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. Scientific Reports, 2017, 7, 9117.	3.3	9
78	Recommendations by the ClinGen Rett/Angelmanâ€like expert panel for geneâ€specific variant interpretation methods. Human Mutation, 2022, 43, 1097-1113.	2.5	8
79	Interested in a career as a clinician-scientist?. DMM Disease Models and Mechanisms, 2010, 3, 125-130.	2.4	6
80	Therapeutic Advances in Autism and Other Neurodevelopmental Disorders. Neurotherapeutics, 2015, 12, 519-520.	4.4	6
81	Can Rett syndrome be diagnosed before regression?. Neuroscience and Biobehavioral Reviews, 2019, 104, 158-159.	6.1	6
82	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. Annals of Neurology, 2019, 86, 332-343.	5.3	5
83	Anthropometric Measures Correspond with Functional Motor Outcomes in Females with Rett Syndrome. Journal of Pediatrics, 2022, 244, 169-177.e3.	1.8	5
84	Detection of rarely identified multiple mutations in MECP2 gene do not contribute to enhanced severity in rett syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1638-1646.	1.2	4
85	Developmental Encephalopathies. , 2017, , 242-248.		4
86	Analysis of <scp>Xâ€inactivation</scp> status in a Rett syndrome natural history study cohort. Molecular Genetics & Genomic Medicine, 2022, 10, e1917.	1.2	4
87	Cortisol profiles and clinical severity in MECP2 duplication syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 19.	3.1	2
88	Rett syndrome and MECP2-related disorders. , 2020, , 269-284.		2
89	1.49 Trofinetide, A Novel IGF-1 Related Treatment for Neurodevelopmental Disorders, Demonstrates Efficacy for Children and Adolescents With Rett Syndrome. Journal of the American Academy of Child and Adolescent Psychiatry, 2017, 56, S168-S169.	0.5	1
90	Abstract 19072: Cardiac Autonomic Dysfunction in Mice Lacking Methyl CpG Binding Protein 2. Circulation, 2014, 130, .	1.6	0