

# Jeffrey L. Neul

## List of Publications by Year in descending order

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Version: 2024-02-01

90  
papers

7,219  
citations

87723

38  
h-index

60497

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. <i>Annals of Neurology</i> , 2010, 68, 944-950.	2.8	1,045
2	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. <i>Nature</i> , 2010, 468, 263-269.	13.7	1,042
3	Methyl-CpG-binding protein 2 (MECP2) mutation type is associated with disease severity in Rett syndrome. <i>Journal of Medical Genetics</i> , 2014, 51, 152-158.	1.5	246
4	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. <i>Genetics in Medicine</i> , 2006, 8, 784-792.	1.1	245
5	Loss of MeCP2 in aminergic neurons causes cell-autonomous defects in neurotransmitter synthesis and specific behavioral abnormalities. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 21966-21971.	3.3	240
6	The Xenopus Dorsalizing Factor noggin Ventralizes Drosophila Embryos by Preventing DPP from Activating Its Receptor. <i>Cell</i> , 1996, 86, 607-617.	13.5	236
7	Deletion of Mecp2 in Sim1-Expressing Neurons Reveals a Critical Role for MeCP2 in Feeding Behavior, Aggression, and the Response to Stress. <i>Neuron</i> , 2008, 59, 947-958.	3.8	230
8	Preclinical research in Rett syndrome: setting the foundation for translational success. <i>DMM Disease Models and Mechanisms</i> , 2012, 5, 733-745.	1.2	183
9	A partial loss of function allele of Methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. <i>Human Molecular Genetics</i> , 2008, 17, 1718-1727.	1.4	173
10	Neurologic Complications Associated With Influenza A in Children During the 2003-2004 Influenza Season in Houston, Texas. <i>Pediatrics</i> , 2004, 114, e626-e633.	1.0	160
11	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. <i>Nature</i> , 2015, 521, E1-E4.	13.7	159
12	Female Mecp2 <sup>+/Δ</sup> mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. <i>Human Molecular Genetics</i> , 2013, 22, 96-109.	1.4	158
13	Gastrointestinal and Nutritional Problems Occur Frequently Throughout Life in Girls and Women With Rett Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2012, 55, 292-298.	0.9	144
14	Rett Syndrome: A Prototypical Neurodevelopmental Disorder. <i>Neuroscientist</i> , 2004, 10, 118-128.	2.6	135
15	Mutual antagonism between Sox10 and NFIA regulates diversification of glial lineages and glioma subtypes. <i>Nature Neuroscience</i> , 2014, 17, 1322-1329.	7.1	124
16	Developmental delay in Rett syndrome: data from the natural history study. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 20.	1.5	118
17	Rett syndrome diagnostic criteria: Lessons from the Natural History Study. <i>Annals of Neurology</i> , 2010, 68, 951-955.	2.8	111
18	Breathing challenges in Rett Syndrome: Lessons learned from humans and animal models. <i>Respiratory Physiology and Neurobiology</i> , 2013, 189, 280-287.	0.7	107

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19	Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. <i>Neurology</i> , 2019, 92, e1912-e1925.	1.5	106
20	Complexities of Rett Syndrome and MeCP2: Figure 1.. <i>Journal of Neuroscience</i> , 2011, 31, 7951-7959.	1.7	101
21	Spatially Restricted Activation of the SAX Receptor by SCW Modulates DPP/TKV Signaling in <i>Drosophila</i> Dorsal-Ventral Patterning. <i>Cell</i> , 1998, 95, 483-494.	13.5	95
22	The relationship of Rett syndrome and <i>MECP2</i> disorders to autism. <i>Dialogues in Clinical Neuroscience</i> , 2012, 14, 253-262.	1.8	90
23	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. <i>ELife</i> , 2016, 5, .	2.8	86
24	Longitudinal course of epilepsy in Rett syndrome and related disorders. <i>Brain</i> , 2017, 140, 306-318.	3.7	80
25	A Double-Blind, Randomized, Placebo-Controlled Clinical Study of Trofinetide in the Treatment of Rett Syndrome. <i>Pediatric Neurology</i> , 2017, 76, 37-46.	1.0	80
26	The Changing Face of Survival in Rett Syndrome and <i>MECP2</i> -Related Disorders. <i>Pediatric Neurology</i> , 2015, 53, 402-411.	1.0	79
27	<i>MECP2</i> Is Critical within <i>HoxB1</i> -Derived Tissues of Mice for Normal Lifespan. <i>Journal of Neuroscience</i> , 2011, 31, 10359-10370.	1.7	75
28	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in <i>MECP2</i> . <i>Genetics in Medicine</i> , 2017, 19, 13-19.	1.1	74
29	Pathogenesis of Lethal Cardiac Arrhythmias in <i>Mecp2</i> Mutant Mice: Implication for Therapy in Rett Syndrome. <i>Science Translational Medicine</i> , 2011, 3, 113ra125.	5.8	72
30	The course of awake breathing disturbances across the lifespan in Rett syndrome. <i>Brain and Development</i> , 2018, 40, 515-529.	0.6	60
31	Insulinotropic treatments exacerbate metabolic syndrome in mice lacking <i>MeCP2</i> function. <i>Human Molecular Genetics</i> , 2013, 22, 2626-2633.	1.4	55
32	Profiling Scoliosis in Rett Syndrome. <i>Pediatric Research</i> , 2010, 67, 435-439.	1.1	54
33	Rett syndrome like phenotypes in the R255X <i>Mecp2</i> mutant mouse are rescued by <i>MECP2</i> transgene. <i>Human Molecular Genetics</i> , 2015, 24, 2662-2672.	1.4	54
34	Overexpression of Methyl-CpG Binding Protein 2 Impairs T <sub>H</sub> 1 Responses. <i>Science Translational Medicine</i> , 2012, 4, 163ra158.	5.8	52
35	Loss of <i>MeCP2</i> in the rat models regression, impaired sociability and transcriptional deficits of Rett syndrome. <i>Human Molecular Genetics</i> , 2016, 25, 3284-3302.	1.4	52
36	A Study of the Treatment of Rett Syndrome With Folate and Betaine. <i>Journal of Child Neurology</i> , 2009, 24, 551-556.	0.7	48

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37	Improving Treatment Trial Outcomes for Rett Syndrome. <i>Journal of Child Neurology</i> , 2015, 30, 1743-1748.	0.7	47
38	The array of clinical phenotypes of males with mutations in <i>Methyl-CpG binding protein 2</i> . <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 55-67.	1.1	47
39	Scoliosis in Rett Syndrome: Progression, Comorbidities, and Predictors. <i>Pediatric Neurology</i> , 2017, 70, 20-25.	1.0	46
40	Age of Diagnosis in Rett Syndrome: Patterns of Recognition Among Diagnosticians and Risk Factors for Late Diagnosis. <i>Pediatric Neurology</i> , 2015, 52, 585-591.e2.	1.0	44
41	Behavioral profiles in Rett syndrome: Data from the natural history study. <i>Brain and Development</i> , 2019, 41, 123-134.	0.6	42
42	Origin of a Non-Clarke's Column Division of the Dorsal Spinocerebellar Tract and the Role of Caudal Proprioceptive Neurons in Motor Function. <i>Cell Reports</i> , 2015, 13, 1258-1271.	2.9	40
43	Vitamin D Deficiency Is Prevalent in Girls and Women With Rett Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2011, 53, 569-574.	0.9	40
44	Brief Report: MECP2 Mutations in People Without Rett Syndrome. <i>Journal of Autism and Developmental Disorders</i> , 2014, 44, 703-711.	1.7	37
45	Characterizing the phenotypic effect of Xq28 duplication size in <i>MECP2</i> duplication syndrome. <i>Clinical Genetics</i> , 2019, 95, 575-581.	1.0	37
46	Multisystem comorbidities in classic Rett syndrome: a scoping review. <i>BMJ Paediatrics Open</i> , 2020, 4, e000731.	0.6	35
47	Consensus guidelines on managing Rett syndrome across the lifespan. <i>BMJ Paediatrics Open</i> , 2020, 4, e000717.	0.6	35
48	Pubertal Development in Rett Syndrome Deviates From Typical Females. <i>Pediatric Neurology</i> , 2014, 51, 769-775.	1.0	32
49	Progressive Changes in a Distributed Neural Circuit Underlie Breathing Abnormalities in Mice Lacking MeCP2. <i>Journal of Neuroscience</i> , 2016, 36, 5572-5586.	1.7	30
50	Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome. <i>Contemporary Clinical Trials</i> , 2022, 114, 106704.	0.8	30
51	Hand stereotypies. <i>Neurology</i> , 2019, 92, e2594-e2603.	1.5	29
52	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. <i>Pediatric Neurology</i> , 2016, 58, 67-74.	1.0	25
53	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. <i>Autism Research</i> , 2018, 11, 212-222.	2.1	25
54	Comparison of Core Features in Four Developmental Encephalopathies in the Rett Natural History Study. <i>Annals of Neurology</i> , 2020, 88, 396-406.	2.8	25

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55	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. <i>Frontiers in Integrative Neuroscience</i> , 2020, 14, 7.	1.0	24
56	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2021, 13, 40.	1.5	22
57	A Psychometric Evaluation of the Motor-Behavioral Assessment Scale for Use as an Outcome Measure in Rett Syndrome Clinical Trials. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2020, 125, 493-509.	0.8	22
58	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. <i>Pediatric Neurology</i> , 2021, 123, 30-37.	1.0	21
59	When Rett syndrome is due to genes other than MECP2. <i>Translational Science of Rare Diseases</i> , 2018, 3, 49-53.	1.6	19
60	Pharmacological read-through of R294X MeCP2 in a novel mouse model of Rett syndrome. <i>Human Molecular Genetics</i> , 2020, 29, 2461-2470.	1.4	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. <i>DMM Disease Models and Mechanisms</i> , 2015, 8, 363-71.	1.2	15
63	Methyl-CpG binding-protein 2 function in cholinergic neurons mediates cardiac arrhythmogenesis. <i>Human Molecular Genetics</i> , 2016, 25, ddx326.	1.4	15
64	Safety and efficacy of genetic <sc><i>MECP2</i></sc> supplementation in the <sc>R294X</sc> mouse model of Rett syndrome. <i>Genes, Brain and Behavior</i> , 2022, 21, e12739.	1.1	15
65	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 31.	1.5	15
66	Towards a consensus on developmental regression. <i>Neuroscience and Biobehavioral Reviews</i> , 2019, 107, 3-5.	2.9	14
67	Multisite Study of Evoked Potentials in Rett Syndrome. <i>Annals of Neurology</i> , 2021, 89, 790-802.	2.8	14
68	Loss of MeCP2 Causes Urological Dysfunction and Contributes to Death by Kidney Failure in Mouse Models of Rett Syndrome. <i>PLoS ONE</i> , 2016, 11, e0165550.	1.1	13
69	Assessment of Caregiver Inventory for Rett Syndrome. <i>Journal of Autism and Developmental Disorders</i> , 2017, 47, 1102-1112.	1.7	13
70	Loss of MeCP2 Function Across Several Neuronal Populations Impairs Breathing Response to Acute Hypoxia. <i>Frontiers in Neurology</i> , 2020, 11, 593554.	1.1	13
71	Phenotypic features in <sc><i>MECP2</i></sc> duplication syndrome: Effects of age. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 362-369.	0.7	13
72	Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2021, 13, 3.	1.5	12

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73	Unfolding neurodevelopmental disorders: The mystery of developing connections. <i>Nature Medicine</i> , 2011, 17, 1353-1355.	15.2	11
74	Low Bone Mineral Mass Is Associated With Decreased Bone Formation and Diet in Girls With Rett Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2014, 59, 386-392.	0.9	11
75	Detection of neurophysiological features in female R255X MeCP2 mutation mice. <i>Neurobiology of Disease</i> , 2020, 145, 105083.	2.1	11
76	Biliary Tract Disease in Girls and Young Women With Rett Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2019, 68, 799-805.	0.9	10
77	Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. <i>Scientific Reports</i> , 2017, 7, 9117.	1.6	9
78	Recommendations by the ClinGen Rett/Angelman-like expert panel for gene-specific variant interpretation methods. <i>Human Mutation</i> , 2022, 43, 1097-1113.	1.1	8
79	Interested in a career as a clinician-scientist?. <i>DMM Disease Models and Mechanisms</i> , 2010, 3, 125-130.	1.2	6
80	Therapeutic Advances in Autism and Other Neurodevelopmental Disorders. <i>Neurotherapeutics</i> , 2015, 12, 519-520.	2.1	6
81	Can Rett syndrome be diagnosed before regression?. <i>Neuroscience and Biobehavioral Reviews</i> , 2019, 104, 158-159.	2.9	6
82	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. <i>Annals of Neurology</i> , 2019, 86, 332-343.	2.8	5
83	Anthropometric Measures Correspond with Functional Motor Outcomes in Females with Rett Syndrome. <i>Journal of Pediatrics</i> , 2022, 244, 169-177.e3.	0.9	5
84	Detection of rarely identified multiple mutations in <i>MECP2</i> gene do not contribute to enhanced severity in rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1638-1646.	0.7	4
85	Developmental Encephalopathies. , 2017, , 242-248.		4
86	Analysis of X-inactivation status in a Rett syndrome natural history study cohort. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1917.	0.6	4
87	Cortisol profiles and clinical severity in MECP2 duplication syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2020, 12, 19.	1.5	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. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2017, 56, S168-S169.	0.3	1
90	Abstract 19072: Cardiac Autonomic Dysfunction in Mice Lacking Methyl CpG Binding Protein 2. <i>Circulation</i> , 2014, 130, .	1.6	0