

# Jeffrey L. Neul

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

84  
papers

5,358  
citations

34  
h-index

73  
g-index

95  
ext. papers

6,324  
ext. citations

8.2  
avg, IF

5.23  
L-index

#	Paper	IF	Citations
84	Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome.. <i>Contemporary Clinical Trials</i> , <b>2022</b> , 106704	2.3	3
83	Analysis of X-inactivation status in a Rett syndrome natural history study cohort.. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2022</b> , e1917	2.3	0
82	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study.. <i>Journal of Neurodevelopmental Disorders</i> , <b>2022</b> , 14, 31	4.6	0
81	Safety and efficacy of genetic MECP2 supplementation in the R294X mouse model of Rett syndrome. <i>Genes, Brain and Behavior</i> , <b>2021</b> , e12739	3.6	4
80	Phenotypic features in MECP2 duplication syndrome: Effects of age. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 362-369	2.5	3
79	Multisite Study of Evoked Potentials in Rett Syndrome. <i>Annals of Neurology</i> , <b>2021</b> , 89, 790-802	9.4	1
78	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. <i>Journal of Neurodevelopmental Disorders</i> , <b>2021</b> , 13, 40	4.6	2
77	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. <i>Pediatric Neurology</i> , <b>2021</b> , 123, 30-37	2.9	3
76	Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. <i>Journal of Neurodevelopmental Disorders</i> , <b>2021</b> , 13, 3	4.6	6
75	Pharmacological read-through of R294X Mecp2 in a novel mouse model of Rett syndrome. <i>Human Molecular Genetics</i> , <b>2020</b> , 29, 2461-2470	5.6	8
74	Comparison of Core Features in Four Developmental Encephalopathies in the Rett Natural History Study. <i>Annals of Neurology</i> , <b>2020</b> , 88, 396-406	9.4	11
73	Rett syndrome and MECP2-related disorders <b>2020</b> , 269-284		1
72	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. <i>Frontiers in Integrative Neuroscience</i> , <b>2020</b> , 14, 7	3.2	8
71	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> , <b>2020</b> , 125, 493-509	2.2	3
70	Multisystem comorbidities in classic Rett syndrome: a scoping review. <i>BMJ Paediatrics Open</i> , <b>2020</b> , 4, e000731	2.4	8
69	Detection of neurophysiological features in female R255X MeCP2 mutation mice. <i>Neurobiology of Disease</i> , <b>2020</b> , 145, 105083	7.5	3
68	Loss of MeCP2 Function Across Several Neuronal Populations Impairs Breathing Response to Acute Hypoxia. <i>Frontiers in Neurology</i> , <b>2020</b> , 11, 593554	4.1	4

67	Cortisol profiles and clinical severity in MECP2 duplication syndrome. <i>Journal of Neurodevelopmental Disorders</i> , <b>2020</b> , 12, 19	4.6	2
66	Consensus guidelines on managing Rett syndrome across the lifespan. <i>BMJ Paediatrics Open</i> , <b>2020</b> , 4, e000717	2.4	6
65	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. <i>Annals of Neurology</i> , <b>2019</b> , 86, 332-343	9.4	1
64	Hand stereotypies: Lessons from the Rett Syndrome Natural History Study. <i>Neurology</i> , <b>2019</b> , 92, e2594-e2603	2.6	12
63	Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. <i>Neurology</i> , <b>2019</b> , 92, e1912-e1925	6.5	46
62	Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. <i>Clinical Genetics</i> , <b>2019</b> , 95, 575-581	4	21
61	Towards a consensus on developmental regression. <i>Neuroscience and Biobehavioral Reviews</i> , <b>2019</b> , 107, 3-5	9	7
60	Can Rett syndrome be diagnosed before regression?. <i>Neuroscience and Biobehavioral Reviews</i> , <b>2019</b> , 104, 158-159	9	2
59	Biliary Tract Disease in Girls and Young Women With Rett Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2019</b> , 68, 799-805	2.8	7
58	Behavioral profiles in Rett syndrome: Data from the natural history study. <i>Brain and Development</i> , <b>2019</b> , 41, 123-134	2.2	14
57	The array of clinical phenotypes of males with mutations in Methyl-CpG binding protein 2. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2019</b> , 180, 55-67	3.5	14
56	The course of awake breathing disturbances across the lifespan in Rett syndrome. <i>Brain and Development</i> , <b>2018</b> , 40, 515-529	2.2	33
55	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. <i>Autism Research</i> , <b>2018</b> , 11, 212-222	5.1	18
54	When Rett syndrome is due to genes other than. <i>Translational Science of Rare Diseases</i> , <b>2018</b> , 3, 49-53	3.3	15
53	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 13-19	8.1	52
52	Scoliosis in Rett Syndrome: Progression, Comorbidities, and Predictors. <i>Pediatric Neurology</i> , <b>2017</b> , 70, 20-25	2.9	25
51	Assessment of Caregiver Inventory for Rett Syndrome. <i>Journal of Autism and Developmental Disorders</i> , <b>2017</b> , 47, 1102-1112	4.6	9
50	Developmental Encephalopathies <b>2017</b> , 242-248		1

49	Longitudinal course of epilepsy in Rett syndrome and related disorders. <i>Brain</i> , <b>2017</b> , 140, 306-318	11.2	52
48	Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. <i>Scientific Reports</i> , <b>2017</b> , 7, 9117	4.9	3
47	A Double-Blind, Randomized, Placebo-Controlled Clinical Study of Trofinetide in the Treatment of Rett Syndrome. <i>Pediatric Neurology</i> , <b>2017</b> , 76, 37-46	2.9	37
46	Methyl-CpG binding-protein 2 function in cholinergic neurons mediates cardiac arrhythmogenesis. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4983-4995	5.6	11
45	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. <i>ELife</i> , <b>2016</b> , 5,	8.9	65
44	Loss of MeCP2 Causes Urological Dysfunction and Contributes to Death by Kidney Failure in Mouse Models of Rett Syndrome. <i>PLoS ONE</i> , <b>2016</b> , 11, e0165550	3.7	11
43	Loss of MeCP2 in the rat models regression, impaired sociability and transcriptional deficits of Rett syndrome. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 3284-3302	5.6	38
42	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. <i>Pediatric Neurology</i> , <b>2016</b> , 58, 67-74	2.9	17
41	Progressive Changes in a Distributed Neural Circuit Underlie Breathing Abnormalities in Mice Lacking MeCP2. <i>Journal of Neuroscience</i> , <b>2016</b> , 36, 5572-86	6.6	21
40	The Changing Face of Survival in Rett Syndrome and MECP2-Related Disorders. <i>Pediatric Neurology</i> , <b>2015</b> , 53, 402-11	2.9	58
39	Improving Treatment Trial Outcomes for Rett Syndrome: The Development of Rett-specific Anchors for the Clinical Global Impression Scale. <i>Journal of Child Neurology</i> , <b>2015</b> , 30, 1743-8	2.5	26
38	Treatment of cardiac arrhythmias in a mouse model of Rett syndrome with Na <sup>+</sup> -channel-blocking antiepileptic drugs. <i>DMM Disease Models and Mechanisms</i> , <b>2015</b> , 8, 363-71	4.1	12
37	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> , <b>2015</b> , 13, 1258-1271	10.6	23
36	Age of diagnosis in Rett syndrome: patterns of recognition among diagnosticians and risk factors for late diagnosis. <i>Pediatric Neurology</i> , <b>2015</b> , 52, 585-91.e2	2.9	28
35	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. <i>Nature</i> , <b>2015</b> , 521, E1-4	50.4	119
34	Rett syndrome like phenotypes in the R255X Mecp2 mutant mouse are rescued by MECP2 transgene. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2662-72	5.6	39
33	Brief report: MECP2 mutations in people without Rett syndrome. <i>Journal of Autism and Developmental Disorders</i> , <b>2014</b> , 44, 703-11	4.6	23
32	Mutual antagonism between Sox10 and NFIA regulates diversification of glial lineages and glioma subtypes. <i>Nature Neuroscience</i> , <b>2014</b> , 17, 1322-9	25.5	93

31	Developmental delay in Rett syndrome: data from the natural history study. <i>Journal of Neurodevelopmental Disorders</i> , <b>2014</b> , 6, 20	4.6	94
30	Methyl-CpG-binding protein 2 (MECP2) mutation type is associated with disease severity in Rett syndrome. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 152-8	5.8	181
29	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> , <b>2014</b> , 59, 386-92	2.8	10
28	Pubertal development in Rett syndrome deviates from typical females. <i>Pediatric Neurology</i> , <b>2014</b> , 51, 769-75	2.9	19
27	Detection of rarely identified multiple mutations in MECP2 gene do not contribute to enhanced severity in Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 1638-46	2.5	4
26	Breathing challenges in Rett syndrome: lessons learned from humans and animal models. <i>Respiratory Physiology and Neurobiology</i> , <b>2013</b> , 189, 280-7	2.8	87
25	Insulinotropic treatments exacerbate metabolic syndrome in mice lacking MeCP2 function. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 2626-33	5.6	37
24	Female <i>Mecp2</i> (+/-) mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 96-109	5.6	117
23	Overexpression of methyl-CpG binding protein 2 impairs T(H)1 responses. <i>Science Translational Medicine</i> , <b>2012</b> , 4, 163ra158	17.5	39
22	Preclinical research in Rett syndrome: setting the foundation for translational success. <i>DMM Disease Models and Mechanisms</i> , <b>2012</b> , 5, 733-45	4.1	154
21	Gastrointestinal and nutritional problems occur frequently throughout life in girls and women with Rett syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2012</b> , 55, 292-8	2.8	104
20	The relationship of Rett syndrome and MECP2 disorders to autism. <i>Dialogues in Clinical Neuroscience</i> , <b>2012</b> , 14, 253-62	5.7	68
19	Complexities of Rett syndrome and MeCP2. <i>Journal of Neuroscience</i> , <b>2011</b> , 31, 7951-9	6.6	89
18	MeCP2 is critical within HoxB1-derived tissues of mice for normal lifespan. <i>Journal of Neuroscience</i> , <b>2011</b> , 31, 10359-70	6.6	69
17	Unfolding neurodevelopmental disorders: the mystery of developing connections. <i>Nature Medicine</i> , <b>2011</b> , 17, 1353-5	50.5	10
16	Pathogenesis of lethal cardiac arrhythmias in <i>Mecp2</i> mutant mice: implication for therapy in Rett syndrome. <i>Science Translational Medicine</i> , <b>2011</b> , 3, 113ra125	17.5	57
15	Rett Syndrome and MECP2-Related Disorders <b>2011</b> , 776-800		2
14	Vitamin D deficiency is prevalent in girls and women with Rett syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2011</b> , 53, 569-74	2.8	28

13	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. <i>Nature</i> , <b>2010</b> , 468, 263-9	50.4	849
12	Profiling scoliosis in Rett syndrome. <i>Pediatric Research</i> , <b>2010</b> , 67, 435-9	3.2	46
11	Rett syndrome: revised diagnostic criteria and nomenclature. <i>Annals of Neurology</i> , <b>2010</b> , 68, 944-50	9.4	804
10	Rett syndrome diagnostic criteria: lessons from the Natural History Study. <i>Annals of Neurology</i> , <b>2010</b> , 68, 951-5	9.4	86
9	A study of the treatment of Rett syndrome with folate and betaine. <i>Journal of Child Neurology</i> , <b>2009</b> , 24, 551-6	2.5	29
8	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> , <b>2009</b> , 106, 21966-71	11.5	198
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> , <b>2008</b> , 59, 947-58	13.9	202
6	A partial loss of function allele of methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 1718-27	5.6	157
5	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. <i>Genetics in Medicine</i> , <b>2006</b> , 8, 784-92	8.1	225
4	Neurologic complications associated with influenza A in children during the 2003-2004 influenza season in Houston, Texas. <i>Pediatrics</i> , <b>2004</b> , 114, e626-33	7.4	134
3	Rett syndrome: a prototypical neurodevelopmental disorder. <i>Neuroscientist</i> , <b>2004</b> , 10, 118-28	7.6	114
2	Spatially restricted activation of the SAX receptor by SCW modulates DPP/TKV signaling in <i>Drosophila</i> dorsal-ventral patterning. <i>Cell</i> , <b>1998</b> , 95, 483-94	56.2	90
1	The <i>Xenopus</i> dorsalizing factor noggin ventralizes <i>Drosophila</i> embryos by preventing DPP from activating its receptor. <i>Cell</i> , <b>1996</b> , 86, 607-17	56.2	215