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	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
2	Recommendations by the ClinGen Rett/Angelmanâ€like expert panel for geneâ€specific variant interpretation methods. Human Mutation, 2022, 43, 1097-1113.	2.5	8
3	Anthropometric Measures Correspond with Functional Motor Outcomes in Females with Rett Syndrome. Journal of Pediatrics, 2022, 244, 169-177.e3.	1.8	5
4	Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome. Contemporary Clinical Trials, 2022, 114, 106704.	1.8	30
5	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
6	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study. Journal of Neurodevelopmental Disorders, 2022, 14, 31.	3.1	15
7	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
8	Multisite Study of Evoked Potentials in Rett Syndrome. Annals of Neurology, 2021, 89, 790-802.	5.3	14
9	Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. Journal of Neurodevelopmental Disorders, 2021, 13, 40.	3.1	22
10	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
11	Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 3.	3.1	12
12	Multisystem comorbidities in classic Rett syndrome: a scoping review. BMJ Paediatrics Open, 2020, 4, e000731.	1.4	35
13	Detection of neurophysiological features in female R255X MeCP2 mutation mice. Neurobiology of Disease, 2020, 145, 105083.	4.4	11
14	Loss of MeCP2 Function Across Several Neuronal Populations Impairs Breathing Response to Acute Hypoxia. Frontiers in Neurology, 2020, 11, 593554.	2.4	13
15	Cortisol profiles and clinical severity in MECP2 duplication syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 19.	3.1	2
16	Consensus guidelines on managing Rett syndrome across the lifespan. BMJ Paediatrics Open, 2020, 4, e000717.	1.4	35
17	Pharmacological read-through of R294X Mecp2 in a novel mouse model of Rett syndrome. Human Molecular Genetics, 2020, 29, 2461-2470.	2.9	19
18	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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19	Rett syndrome and MECP2-related disorders. , 2020, , 269-284.		2
20	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. Frontiers in Integrative Neuroscience, 2020, 14, 7.	2.1	24
21	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
22	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
23	Towards a consensus on developmental regression. Neuroscience and Biobehavioral Reviews, 2019, 107, 3-5.	6.1	14
24	Can Rett syndrome be diagnosed before regression?. Neuroscience and Biobehavioral Reviews, 2019, 104, 158-159.	6.1	6
25	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. Annals of Neurology, 2019, 86, 332-343.	5.3	5
26	Hand stereotypies. Neurology, 2019, 92, e2594-e2603.	1.1	29
27	Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. Neurology, 2019, 92, e1912-e1925.	1.1	106
28	Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. Clinical Genetics, 2019, 95, 575-581.	2.0	37
29	Biliary Tract Disease in Girls and Young Women With Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, 799-805.	1.8	10
30	Behavioral profiles in Rett syndrome: Data from the natural history study. Brain and Development, 2019, 41, 123-134.	1.1	42
31	The course of awake breathing disturbances across the lifespan in Rett syndrome. Brain and Development, 2018, 40, 515-529.	1.1	60
32	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. Autism Research, 2018, 11, 212-222.	3.8	25
33	When Rett syndrome is due to genes other than MECP2. Translational Science of Rare Diseases, 2018, 3, 49-53.	1.5	19
34	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
35	Scoliosis in Rett Syndrome: Progression, Comorbidities, and Predictors. Pediatric Neurology, 2017, 70, 20-25.	2.1	46
36	Assessment of Caregiver Inventory for Rett Syndrome. Journal of Autism and Developmental Disorders, 2017, 47, 1102-1112.	2.7	13

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37	Developmental Encephalopathies. , 2017, , 242-248.		4
38	Longitudinal course of epilepsy in Rett syndrome and related disorders. Brain, 2017, 140, 306-318.	7.6	80
39	Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. Scientific Reports, 2017, 7, 9117.	3.3	9
40	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
41	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
42	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
43	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
44	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. Pediatric Neurology, 2016, 58, 67-74.	2.1	25
45	Progressive Changes in a Distributed Neural Circuit Underlie Breathing Abnormalities in Mice Lacking MeCP2. Journal of Neuroscience, 2016, 36, 5572-5586.	3.6	30
46	Methyl-CpG binding-protein 2 function in cholinergic neurons mediates cardiac arrhythmogenesis. Human Molecular Genetics, 2016, 25, ddw326.	2.9	15
47	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. ELife, 2016, 5, .	6.0	86
48	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
49	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
50	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. Nature, 2015, 521, E1-E4.	27.8	159
51	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
52	The Changing Face of Survival in Rett Syndrome andÂMECP2-Related Disorders. Pediatric Neurology, 2015, 53, 402-411.	2.1	79
53	Improving Treatment Trial Outcomes for Rett Syndrome. Journal of Child Neurology, 2015, 30, 1743-1748.	1.4	47
54	Therapeutic Advances in Autism and Other Neurodevelopmental Disorders. Neurotherapeutics, 2015, 12, 519-520.	4.4	6

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55	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
56	Pubertal Development in Rett Syndrome Deviates From Typical Females. Pediatric Neurology, 2014, 51, 769-775.	2.1	32
57	Brief Report: MECP2 Mutations in People Without Rett Syndrome. Journal of Autism and Developmental Disorders, 2014, 44, 703-711.	2.7	37
58	Mutual antagonism between Sox10 and NFIA regulates diversification of glial lineages and glioma subtypes. Nature Neuroscience, 2014, 17, 1322-1329.	14.8	124
59	Developmental delay in Rett syndrome: data from the natural history study. Journal of Neurodevelopmental Disorders, 2014, 6, 20.	3.1	118
60	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
61	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
62	Abstract 19072: Cardiac Autonomic Dysfunction in Mice Lacking Methyl CpG Binding Protein 2. Circulation, 2014, 130, .	1.6	0
63	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
64	Breathing challenges in Rett Syndrome: Lessons learned from humans and animal models. Respiratory Physiology and Neurobiology, 2013, 189, 280-287.	1.6	107
65	Insulinotropic treatments exacerbate metabolic syndrome in mice lacking MeCP2 function. Human Molecular Genetics, 2013, 22, 2626-2633.	2.9	55
66	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
67	Overexpression of Methyl-CpG Binding Protein 2 Impairs T _H 1 Responses. Science Translational Medicine, 2012, 4, 163ra158.	12.4	52
68	Preclinical research in Rett syndrome: setting the foundation for translational success. DMM Disease Models and Mechanisms, 2012, 5, 733-745.	2.4	183
69	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
70	The relationship of Rett syndrome and MECP2 disorders to autism. Dialogues in Clinical Neuroscience, 2012, 14, 253-262.	3.7	90
71	Complexities of Rett Syndrome and MeCP2: Figure 1.. Journal of Neuroscience, 2011, 31, 7951-7959.	3.6	101
72	MeCP2 Is Critical within HoxB1-Derived Tissues of Mice for Normal Lifespan. Journal of Neuroscience, 2011, 31, 10359-10370.	3.6	75

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73	Unfolding neurodevelopmental disorders: The mystery of developing connections. Nature Medicine, 2011, 17, 1353-1355.	30.7	11
74	Pathogenesis of Lethal Cardiac Arrhythmias in Mecp2 Mutant Mice: Implication for Therapy in Rett Syndrome. Science Translational Medicine, 2011, 3, 113ra125.	12.4	72
75	Rett Syndrome and MECP2-Related Disorders. , 2011, , 776-800.		18
76	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
77	Rett syndrome: Revised diagnostic criteria and nomenclature. Annals of Neurology, 2010, 68, 944-950.	5.3	1,045
78	Rett syndrome diagnostic criteria: Lessons from the Natural History Study. Annals of Neurology, 2010, 68, 951-955.	5.3	111
79	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269.	27.8	1,042
80	Interested in a career as a clinician-scientist?. DMM Disease Models and Mechanisms, 2010, 3, 125-130.	2.4	6
81	Profiling Scoliosis in Rett Syndrome. Pediatric Research, 2010, 67, 435-439.	2.3	54
82	A Study of the Treatment of Rett Syndrome With Folate and Betaine. Journal of Child Neurology, 2009, 24, 551-556.	1.4	48
83	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
84	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
85	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
86	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
87	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
88	Rett Syndrome: A Prototypical Neurodevelopmental Disorder. Neuroscientist, 2004, 10, 118-128.	3.5	135
89	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
90	The Xenopus Dorsalizing Factor noggin Ventralizes Drosophila Embryos by Preventing DPP from Activating Its Receptor. Cell, 1996, 86, 607-617.	28.9	236