# Jeffrey L. Neul

## List of Publications by Citations



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|---|---|---|---|
| 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 | Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. *Nature*, **2010**, 468, 263-9 | 50.4 | 849 |
| 83 | Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, **2010**, 68, 944-50 | 9.4 | 804 |
| 82 | Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. *Genetics in Medicine*, **2006**, 8, 784-92 | 8.1 | 225 |
| 81 | The Xenopus dorsalizing factor noggin ventralizes Drosophila embryos by preventing DPP from activating its receptor. *Cell*, **1996**, 86, 607-17 | 56.2 | 215 |
| 80 | 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, 947-58 | 13.9 | 202 |
| 79 | 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, 21966-71 | 11.5 | 198 |
| 78 | Methyl-CpG-binding protein 2 (MECP2) mutation type is associated with disease severity in Rett syndrome. *Journal of Medical Genetics*, **2014**, 51, 152-8 | 5.8 | 181 |
| 77 | A partial loss of function allele of methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. *Human Molecular Genetics*, **2008**, 17, 1718-27 | 5.6 | 157 |
| 76 | Preclinical research in Rett syndrome: setting the foundation for translational success. *DMM Disease Models and Mechanisms*, **2012**, 5, 733-45 | 4.1 | 154 |
| 75 | Neurologic complications associated with influenza A in children during the 2003-2004 influenza season in Houston, Texas. *Pediatrics*, **2004**, 114, e626-33 | 7.4 | 134 |
| 74 | Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. *Nature*, **2015**, 521, E1-4 | 50.4 | 119 |
| 73 | Female Mecp2(+/-) mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. *Human Molecular Genetics*, **2013**, 22, 96-109 | 5.6 | 117 |
| 72 | Rett syndrome: a prototypical neurodevelopmental disorder. *Neuroscientist*, **2004**, 10, 118-28 | 7.6 | 114 |
| 71 | Gastrointestinal and nutritional problems occur frequently throughout life in girls and women with Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2012**, 55, 292-8 | 2.8 | 104 |
| 70 | Developmental delay in Rett syndrome: data from the natural history study. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 20 | 4.6 | 94 |
| 69 | Mutual antagonism between Sox10 and NFIA regulates diversification of glial lineages and glioma subtypes. *Nature Neuroscience*, **2014**, 17, 1322-9 | 25.5 | 93 |
| 68 | Spatially restricted activation of the SAX receptor by SCW modulates DPP/TKV signaling in Drosophila dorsal-ventral patterning. *Cell*, **1998**, 95, 483-94 | 56.2 | 90 |

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|---|---|---|---|
| 67 | Complexities of Rett syndrome and MeCP2. *Journal of Neuroscience*, **2011**, 31, 7951-9 | 6.6 | 89 |
| 66 | Breathing challenges in Rett syndrome: lessons learned from humans and animal models. *Respiratory Physiology and Neurobiology*, **2013**, 189, 280-7 | 2.8 | 87 |
| 65 | Rett syndrome diagnostic criteria: lessons from the Natural History Study. *Annals of Neurology*, **2010**, 68, 951-5 | 9.4 | 86 |
| 64 | MeCP2 is critical within HoxB1-derived tissues of mice for normal lifespan. *Journal of Neuroscience*, **2011**, 31, 10359-70 | 6.6 | 69 |
| 63 | The relationship of Rett syndrome and MECP2 disorders to autism. *Dialogues in Clinical Neuroscience*, **2012**, 14, 253-62 | 5.7 | 68 |
| 62 | Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. *ELife*, **2016**, 5, | 8.9 | 65 |
| 61 | The Changing Face of Survival in Rett Syndrome and MECP2-Related Disorders. *Pediatric Neurology*, **2015**, 53, 402-11 | 2.9 | 58 |
| 60 | Pathogenesis of lethal cardiac arrhythmias in Mecp2 mutant mice: implication for therapy in Rett syndrome. *Science Translational Medicine*, **2011**, 3, 113ra125 | 17.5 | 57 |
| 59 | Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. *Genetics in Medicine*, **2017**, 19, 13-19 | 8.1 | 52 |
| 58 | Longitudinal course of epilepsy in Rett syndrome and related disorders. *Brain*, **2017**, 140, 306-318 | 11.2 | 52 |
| 57 | Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. *Neurology*, **2019**, 92, e1912-e1925 | 6.5 | 46 |
| 56 | Profiling scoliosis in Rett syndrome. *Pediatric Research*, **2010**, 67, 435-9 | 3.2 | 46 |
| 55 | Rett syndrome like phenotypes in the R255X Mecp2 mutant mouse are rescued by MECP2 transgene. *Human Molecular Genetics*, **2015**, 24, 2662-72 | 5.6 | 39 |
| 54 | Overexpression of methyl-CpG binding protein 2 impairs T(H)1 responses. *Science Translational Medicine*, **2012**, 4, 163ra158 | 17.5 | 39 |
| 53 | Loss of MeCP2 in the rat models regression, impaired sociability and transcriptional deficits of Rett syndrome. *Human Molecular Genetics*, **2016**, 25, 3284-3302 | 5.6 | 38 |
| 52 | A Double-Blind, Randomized, Placebo-Controlled Clinical Study of Trofinetide in the Treatment of Rett Syndrome. *Pediatric Neurology*, **2017**, 76, 37-46 | 2.9 | 37 |
| 51 | Insulinotropic treatments exacerbate metabolic syndrome in mice lacking MeCP2 function. *Human Molecular Genetics*, **2013**, 22, 2626-33 | 5.6 | 37 |
| 50 | The course of awake breathing disturbances across the lifespan in Rett syndrome. *Brain and Development*, **2018**, 40, 515-529 | 2.2 | 33 |

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| 49 | A study of the treatment of Rett syndrome with folate and betaine. *Journal of Child Neurology*, **2009**, 24, 551-6 | 2.5 | 29 |
| 48 | Age of diagnosis in Rett syndrome: patterns of recognition among diagnosticians and risk factors for late diagnosis. *Pediatric Neurology*, **2015**, 52, 585-91.e2 | 2.9 | 28 |
| 47 | Vitamin D deficiency is prevalent in girls and women with Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2011**, 53, 569-74 | 2.8 | 28 |
| 46 | Improving Treatment Trial Outcomes for Rett Syndrome: The Development of Rett-specific Anchors for the Clinical Global Impression Scale. *Journal of Child Neurology*, **2015**, 30, 1743-8 | 2.5 | 26 |
| 45 | Scoliosis in Rett Syndrome: Progression, Comorbidities, and Predictors. *Pediatric Neurology*, **2017**, 70, 20-25 | 2.9 | 25 |
| 44 | Brief report: MECP2 mutations in people without Rett syndrome. *Journal of Autism and Developmental Disorders*, **2014**, 44, 703-11 | 4.6 | 23 |
| 43 | Origin of a Non-Clarke's Column Division of the Dorsal Spinocerebellar Tract and the Role of Caudal Proprioceptive Neurons in Motor Function. *Cell Reports*, **2015**, 13, 1258-1271 | 10.6 | 23 |
| 42 | Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. *Clinical Genetics*, **2019**, 95, 575-581 | 4 | 21 |
| 41 | Progressive Changes in a Distributed Neural Circuit Underlie Breathing Abnormalities in Mice Lacking MeCP2. *Journal of Neuroscience*, **2016**, 36, 5572-86 | 6.6 | 21 |
| 40 | Pubertal development in Rett syndrome deviates from typical females. *Pediatric Neurology*, **2014**, 51, 769-75 | 2.9 | 19 |
| 39 | Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. *Autism Research*, **2018**, 11, 212-222 | 5.1 | 18 |
| 38 | Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. *Pediatric Neurology*, **2016**, 58, 67-74 | 2.9 | 17 |
| 37 | When Rett syndrome is due to genes other than. *Translational Science of Rare Diseases*, **2018**, 3, 49-53 | 3.3 | 15 |
| 36 | Behavioral profiles in Rett syndrome: Data from the natural history study. *Brain and Development*, **2019**, 41, 123-134 | 2.2 | 14 |
| 35 | The array of clinical phenotypes of males with mutations in Methyl-CpG binding protein 2. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2019**, 180, 55-67 | 3.5 | 14 |
| 34 | Hand stereotypies: Lessons from the Rett Syndrome Natural History Study. *Neurology*, **2019**, 92, e2594-e2603 | 2.9 | 12 |
| 33 | Treatment of cardiac arrhythmias in a mouse model of Rett syndrome with Na+-channel-blocking antiepileptic drugs. *DMM Disease Models and Mechanisms*, **2015**, 8, 363-71 | 4.1 | 12 |
| 32 | Comparison of Core Features in Four Developmental Encephalopathies in the Rett Natural History Study. *Annals of Neurology*, **2020**, 88, 396-406 | 9.4 | 11 |

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| 31 | Methyl-CpG binding-protein 2 function in cholinergic neurons mediates cardiac arrhythmogenesis. *Human Molecular Genetics*, **2016**, 25, 4983-4995 | 5.6 | 11 |
| 30 | Loss of MeCP2 Causes Urological Dysfunction and Contributes to Death by Kidney Failure in Mouse Models of Rett Syndrome. *PLoS ONE*, **2016**, 11, e0165550 | 3.7 | 11 |
| 29 | 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, 386-92 | 2.8 | 10 |
| 28 | Unfolding neurodevelopmental disorders: the mystery of developing connections. *Nature Medicine*, **2011**, 17, 1353-5 | 50.5 | 10 |
| 27 | Assessment of Caregiver Inventory for Rett Syndrome. *Journal of Autism and Developmental Disorders*, **2017**, 47, 1102-1112 | 4.6 | 9 |
| 26 | Pharmacological read-through of R294X Mecp2 in a novel mouse model of Rett syndrome. *Human Molecular Genetics*, **2020**, 29, 2461-2470 | 5.6 | 8 |
| 25 | Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. *Frontiers in Integrative Neuroscience*, **2020**, 14, 7 | 3.2 | 8 |
| 24 | Multisystem comorbidities in classic Rett syndrome: a scoping review. *BMJ Paediatrics Open*, **2020**, 4, e000731 | 2.4 | 8 |
| 23 | Towards a consensus on developmental regression. *Neuroscience and Biobehavioral Reviews*, **2019**, 107, 3-5 | 9 | 7 |
| 22 | Biliary Tract Disease in Girls and Young Women With Rett Syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2019**, 68, 799-805 | 2.8 | 7 |
| 21 | Consensus guidelines on managing Rett syndrome across the lifespan. *BMJ Paediatrics Open*, **2020**, 4, e000717 | 2.4 | 6 |
| 20 | Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 3 | 4.6 | 6 |
| 19 | 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**, 161A, 1638-46 | 2.5 | 4 |
| 18 | Loss of MeCP2 Function Across Several Neuronal Populations Impairs Breathing Response to Acute Hypoxia. *Frontiers in Neurology*, **2020**, 11, 593554 | 4.1 | 4 |
| 17 | Safety and efficacy of genetic MECP2 supplementation in the R294X mouse model of Rett syndrome. *Genes, Brain and Behavior*, **2021**, e12739 | 3.6 | 4 |
| 16 | Embryonic hindbrain patterning genes delineate distinct cardio-respiratory and metabolic homeostatic populations in the adult. *Scientific Reports*, **2017**, 7, 9117 | 4.9 | 3 |
| 15 | Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome.. *Contemporary Clinical Trials*, **2022**, 106704 | 2.3 | 3 |
| 14 | 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 | 2.2 | 3 |

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| 13 | Detection of neurophysiological features in female R255X MeCP2 mutation mice. *Neurobiology of Disease*, **2020**, 145, 105083 | 7.5 | 3 |
| 12 | Phenotypic features in MECP2 duplication syndrome: Effects of age. *American Journal of Medical Genetics, Part A*, **2021**, 185, 362-369 | 2.5 | 3 |
| 11 | Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. *Pediatric Neurology*, **2021**, 123, 30-37 | 2.9 | 3 |
| 10 | Can Rett syndrome be diagnosed before regression?. *Neuroscience and Biobehavioral Reviews*, **2019**, 104, 158-159 | 9 | 2 |
| 9 | Rett Syndrome and MECP2-Related Disorders **2011**, 776-800 | | 2 |
| 8 | Cortisol profiles and clinical severity in MECP2 duplication syndrome. *Journal of Neurodevelopmental Disorders*, **2020**, 12, 19 | 4.6 | 2 |
| 7 | Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 40 | 4.6 | 2 |
| 6 | Developmental Encephalopathies **2017**, 242-248 | | 1 |
| 5 | Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. *Annals of Neurology*, **2019**, 86, 332-343 | 9.4 | 1 |
| 4 | Rett syndrome and MECP2-related disorders **2020**, 269-284 | | 1 |
| 3 | Multisite Study of Evoked Potentials in Rett Syndrome. *Annals of Neurology*, **2021**, 89, 790-802 | 9.4 | 1 |
| 2 | Analysis of X-inactivation status in a Rett syndrome natural history study cohort.. *Molecular Genetics & Genomic Medicine*, **2022**, e1917 | 2.3 | 0 |
| 1 | Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study.. *Journal of Neurodevelopmental Disorders*, **2022**, 14, 31 | 4.6 | 0 |