



PERF[®]
Pediatric Epilepsy Research Foundation

POST AWARD REPORT



Investigators: Grace Gombolay, MD and Coral Stredny, MD **Grant Award Year:** 2022

Amount of PERF[®] Grant: \$300,000

☐ **Planning Grant**

Investigator Institution: Emory University (GG) / Boston Children's Hospital

Project Title: CONNECT: Registry/**CON**quering **Neu**roinflammation and **Epilepsies** Consor**Tium**

Project Description: This three-year registry grant is to continue establish a multi-center prospective patient registry including patients with rare neuroinflammatory and inflammatory epilepsy syndromes. These diseases include autoimmune encephalitis, Rasmussen encephalitis, and Landau-Kleffner syndrome. For this grant the goal was to include additional sites and enroll additional participants. The next steps are to begin magnetic resonance imaging analyses of MRIs from patients in this cohort and also cytokine analyses.

Project Goals/Objectives:

1. Expand CONNECT – The goal is to add 9 sites over the next 3 years. In year 1 we have added 3 additional sites to equal 6 sites, with 6 additional sites identified to be added in the next 2 years.
2. Characterize MRI and EEG features and their relationship to predict severity and outcomes. We first began with a retrospective multi-center study that demonstrated T2 frontal and occipital lesions associate with poor outcomes in children with NMDA receptor encephalitis, which was presented as an abstract and published as a manuscript. The next step will be to assess EEG features and their associations with outcomes.
3. Determine blood and CSF biomarkers and their associations with outcomes. We have identified potential biomarkers in blood and CSF that associate with one year outcomes in anti-NMDA receptor encephalitis in 6 participants. The next steps will be to

examine these cytokines in a larger cohort. We have collected samples to move towards accomplishing this aim.

Outcome of Research: We have met some of the aims in this project such as expanding CONNECT and now having a multicenter biorepository. We have done some MRI and EEG feature analyses but are working on a larger study in examining MRI volumetrics using NeuroQuant and performing cytokine analyses which we plan to do during our no cost extension period.

Subsequent Funding ☐ None to report.

Year: 2025

Funder: NIH U54 Rare Disease Clinical Research Network Grant

Amount: \$7,572,234

Project: ARISEN (Autoimmunity, Rasmussen's, Inflammation & Status Epilepticus Research Network)

Year: 2025

Funder: NIH K23

Amount: \$1,098,225

Project: The Role of T cells in children with anti-NMDA receptor encephalitis

Year: Click or tap here to enter text.

Funder: Click or tap here to enter text.

Amount: Click or tap here to enter text.

Project: Click or tap here to enter text.

Subsequent Publications ☐ None to report.

1. Gombolay G, Brenton JN, Yang JH, Stredny CM, Kammeyer R, Otten CE, Vu N, Santoro JD, Robles-Lopez K, Christiana A, Steriade C, Morris M, Gorman M, Moodley M, Hardy D, Kornbluh AB, Kahn I, Sepeta LN, Yeshokumar A; Conquering Neuroinflammation and Epilepsies Consortium (CONNECT). MRI Features and Their Association With Outcomes in Children With Anti-NMDA Receptor Encephalitis. *Neurol Neuroimmunol Neuroinflamm*. 2023 May 26;10(4):e200130. doi: 10.1212/NXI.0000000000200130. PMID: 37236807; PMCID: PMC10219134.

2. Gombolay G, Brenton JN, Yang JH, Stredny CM, Kammeyer R, Fisher KS, Sandweiss AJ, Erickson TA, Kannan V, Otten C, Steriade C, Vu N, Santoro JD, Robles-Lopez K, Goodrich R, Otallah S, Arellano J, Christiana A, Morris M, Gorman MP, Kornbluh AB, Kahn I, Sepeta L, Jiang Y, Muscal E, Murray KO, Moodley M, Hardy D. Isolated Psychiatric Symptoms in Children With Anti-N-Methyl-d Aspartate Receptor Encephalitis. *Pediatr Neurol*. 2024 Oct;159:12-15. doi:

10.1016/j.pediatrneurol.2024.07.009. Epub 2024 Jul 17. PMID: 39094249; PMCID: PMC11381152.

[Subsequent Presentations](#) ☐ None to report.

Abstracts:

1. Gombolay GY, Brenton JN, Yang JH, Stredny CM, Kammeyer R, Otten C, Vu N, Santoro JD, Robles-Lopze K, Christiana A, Steriade C, Morris M, Gorman MM, Moodley M, Hardy D, Kornbluh A, Kahn I, Sepeta L, Yeshokumar A. MRI features and their association with outcomes in children with anti-NMDA receptor encephalitis, American Academy of Neurology, Boston, MA, 2023 (Oral)
2. Gombolay GY, Brenton JN, Yang JH, Stredny CM, Kammeyer R, Otten C, Vu N, Santoro JD, Robles-Lopze K, Christiana A, Steriade C, Morris M, Gorman MM, Moodley M, Hardy D, Kornbluh A, Kahn I, Sepeta L. Initial MRI, EEG, and CSF White Cell Count Are Abnormal in Children with Anti-NMDA Receptor Encephalitis with Isolated Psychiatric Symptoms American Academy of Neurology, Denver, CO, 2024 (Poster).
3. Gombolay GY, Brenton JN, Yang JH, Stredny CM, Kammeyer R, Fisher KS, Sandweiss AJ, Erickson TA, Kannan V, Otten C, Vu N, Santoro JD, Robles-Lopez K, Goodrich R, Otallah S, Arellano J, Christiana A, Morris M, Gorman M, Kornbluh A, Kahn I, Sepeta L, Jiang Y, Muscal E, Murray KO, Moodley M, Hardy D, Steriade C. EEG Features That Associate with One-Year Outcomes in Pediatric Anti-NMDA Receptor Encephalitis, American Academy of Neurology, San Diego, CA, 2025 (Poster)

As of 10.21.2025