

2025

Schwarz AC, Breuner CC, **Blume HK**. Retrospective Study of the Efficacy of Biofeedback Therapy for Pediatric Persistent Posttraumatic Headache. *J Child Neurol*. 2025 May;40(5):357-365. doi: 10.1177/08830738241312942. Epub 2025 Jan 17. PMID: 39819114.

Patrick KE, Shields AN, Dustin HA, Patel AD, McNally K. Cognitive screening informs referrals for neuropsychological evaluation in children with epilepsy. *Epilepsia*. 2025 Apr 26. doi: 10.1111/epi.18421. Epub ahead of print. PMID: 40286280

Reecher HM, Park SE, Ailion A, Berl MM, Hennrick H, Gabriel M, Boyer K, Cooper C, Decrow AM, Duong P, Hodges E, Loblein H, Marshall D, McNally K, **Patrick KE**, Romain J, Sepeta L, Zaccariello M, Koop JI; Pediatric Epilepsy Research Consortium PERC Surgery Workgroup. Association of the cognitive lateralization rating Index with surgical variables of a national cohort of pediatric patients with epilepsy. *Epilepsy Behav*. 2025 Apr 15;168:110404. doi: 10.1016/j.yebeh.2025.110404. Epub ahead of print. PMID: 40239615.

Alves CAPF, Rossi-Espagnet MC, Perez F, Manteghinejad A, Peterson JT, Ganetzky R, Napolitano A, Grassi F, George-Sankoh I, Yildiz H, Muraresku C, Falk MJ, Martinelli D, Longo D, Vanderver A, Gandolfo C, **Saneto RP**, Goldstein A, Vossough A. Single Large-Scale Mitochondrial Deletion Syndromes: Neuroimaging Phenotypes and Longitudinal Progression in Pediatric Patients. *AJNR Am J Neuroradiol*. 2025 Apr 10. doi: 10.3174/ajnr.A8670. Epub ahead of print. PMID: 40210455

Ostendorf AP, Loddenkemper T, **Morgan LA**, Appavu B, Farias-Moeller R, Harrar D, Press C, Abend NS, Gaillard WD, Bai S, Eisner M, McHenry L, Kroshus E, Vannatta K, Goodkin HP. Treating seizures faster: The Quality Improvement in Time to Treat Status Epilepticus (QuITT-SE) multicenter randomized stepped wedge clinical trial protocol. *Contemp Clin Trials*. 2025 Apr;151:107831. doi: 10.1016/j.cct.2025.107831. Epub 2025 Feb 8. PMID: 39929261.

Vitanza NA, Ronsley R, Choe M, Seidel K, Huang W, Rawlings-Rhea SD, Beam M, Steinmetzer L, Wilson AL, Brown C, Beebe A, Lindgren C, Gustafson JA, Wein A, Holtzclaw S, Hoepfner C, Goldstein HE, Browd SR, Hauptman JS, Lee A, Ojemann JG, Crotty EE, Leary SES, Perez FA, Wright JN, Alonso MM, Dun MD, Foster JB, Hurst D, Kong A, Thomsen A, Orentas RJ, Albert CM, Pinto N, Annesley C, Gardner RA, Ho O, Pattabhi S, **Gust J**, Wendler JP, Park JR, Jensen MC. Intracerebroventricular B7-H3-targeting CAR T cells for diffuse intrinsic pontine glioma: a phase 1 trial. *Nat Med*. 2025 Mar;31(3):861-868. doi: 10.1038/s41591-024-03451-3. Epub 2025 Jan 7. PMID: 39775044; PMCID: PMC11922736.

Hauser Chatterjee J, Monrad PA, Goldstein HE, Shaw DW, Ojemann JG, **Randle SC**, Hauptman JS, **Blume HK**. Indomethacin-responsive refractory headache: Two case reports in children after hemispherectomy for Rasmussen's encephalitis. *Headache*. 2025 Mar 25. doi: 10.1111/head.14928. Epub ahead of print. PMID: 40129243.

Castillo-Pinto C, Yu P, **Wainwright MS**, Kirschen MP. Impaired Cerebral Autoregulation in Children. *Pediatr Neurol*. 2025 Mar 13;167:9-16. doi: 10.1016/j.pediatrneurol.2025.03.003. Epub ahead of print. PMID: 40184896.

Gust J, Cole BL, Ronsley R, Wilson AL, Seidel K, Wendler J, Pattabhi S, Brown C, Rawlings-Rhea SD, Shtanukhina N, Browd SR, Hauptman JS, Lee A, Ojemann JG, Crotty EE, Leary SES, Perez FA, Wright JN, Albert CM, Pinto N, Gardner RA, Vitanza NA, Jensen MC, Park JR. Locoregional Infusion of EGFR806-CAR T Cells for Recurrent or Refractory Pediatric CNS Tumors: Results of the Completed BrainChild02 Phase 1 Clinical Trial. *Neuro Oncol*. 2025 Mar 12;noaf064. doi: 10.1093/neuonc/noaf064. Epub ahead of print. PMID: 40070357.

Hauser Chatterjee J, Monrad PA, Goldstein HE, Shaw DW, Ojemann JG, **Randle SC**, Hauptman JS, **Blume HK**. Indomethacin-responsive refractory headache: Two case reports in children after hemispherectomy for Rasmussen's encephalitis. *Headache*. 2025 Mar 25. doi: 10.1111/head.14928. Epub ahead of print. PMID: 40129243.

Fullerton HJ, Hills NK, Wintermark M, Dlamini N, **Amalie-Lefond C**, Dowling MM, Jordan LC, Friedman NR, Elkind MSV, Grose C; VIPS II Investigators*. Evidence of Varicella Zoster Virus Reactivation in Children With Arterial Ischemic Stroke: Results of the VIPS II Study. *J Am Heart Assoc*. 2025 Mar 18;14(6):e039184. doi: 10.1161/JAHA.124.039184. Epub 2025 Mar 18. PMID: 40099546.

Park L, Tsai YT, Lim HK, Faulhaber LD, Burleigh K, Faulhaber EM, Bose M, Shih AY, Hirayama AY, Turtle CJ, Annesley CE, Gardner RA, Gustafson HH, **Gust J**. Cytokine-mediated increase in endothelial-leukocyte interaction mediates brain capillary plugging during CAR T cell neurotoxicity. *bioRxiv [Preprint]*. 2025 Feb 25:2025.02.19.638920. doi: 10.1101/2025.02.19.638920. PMID: 40060404; PMCID: PMC11888194

Pediatric Neurology Faculty Publications 2014-2025

Ivaniuk A, Anselm IA, Bowen A, Cohen BH, Eminoglu FT, Estrella J, Gallagher RC, Ganetzky RD, Gannon J, Gorman GS, Greene C, Gropman AL, Haas RH, Hirano M, Kapoor S, Karaa A, Koenig MK, Kornblum C, Kose E, Larson A, Lichter-Konecki U, Lopriore P, Mancuso M, McFarland R, Moe AM, Morava E, Ng YS, **Saneto RP**, Scaglia F, Sue CM, Tarnopolsky M, Walker MA, Parikh S; as the Hong Kong Mitochondrial Diseases Interest Group; Cheuk-Wing F, Wong TS, Belaramani K, Chan CK, Chan WK, Chan WL, Cheung HW, Cheung KY, Chang SK, Cheung SN, Cheung TF, Cheung YF, Chong SJ, Chow CJ, Chung HB, Fan SF, Fok WJ, Fong KW, Fung TS, Hui KF, Hui TH, Hui J, Ko CH, Kwan MC, Kwok MA, Kwok SJ, Lai MS, Lam YO, Lam CW, Lau MC, Law CE, Law HF, Lee WC, Hencher Lee HC, Leung KH, Leung KY, Li SH, Ling TJ, Liu KT, Lo FM, Lui C, Luk CO, Luk HM, Ma CK, Ma K, Ma KH, Mew YN, Mo A, Hg SF, Poon WG, Sheng B, Szeto CC, Tai SM, Tang JL, Tse CA, Tsung LL, Wong HJ, Wong WW, Wong KK, Wong SS, Wong CV, Wong WS, Wong CF, Wu SP, Wu HJ, Yau MM, Yau KE, Yeung WL, Yeung HJ, Yip KE, Wu HJ, Young PT, Yuan G, Yuen YL, Yuen CL. Characterization of Factors Associated With Death in Deceased Patients With Mitochondrial Disorders: A Multicenter Cross-Sectional Survey. *Neurology*. 2025 Feb 25;104(4):e209779. doi: 10.1212/WNL.0000000000209779. Epub 2025 Jan 30. PMID: 39883904; PMCID: PMC11781783.

Schierbaum L, Quiroz V, Yang K, Rong J, Battaglia N, Zubair U, Christie M, Davis M, Calame D, Danzi MC, Finkel RS, Burns J, Gilbert DL, **Mingbunjerdusuk D**, Pruitt G, Pruitt N, Cobb J, Sadjadi R, Cashman CR, Blackstone C, Fink JK, Shy ME, Zuchner S, Ebrahimi-Fakhari D. The Spastic Paraplegia-Centers of Excellence Research Network (SP-CERN): Clinical Trial Readiness for Hereditary Spastic Paraplegia. *Neurol Genet*. 2025 Feb 21;11(2):e200249. doi: 10.1212/NXG.0000000000200249. PMID: 39996129; PMCID: PMC11849523.

Crotty EE, **Sato AA**, Abdelbaki MS. Integrating MAPK pathway inhibition into standard-of-care therapy for pediatric low-grade glioma. *Front Oncol*. 2025 Feb 11;15:1520316. doi: 10.3389/fonc.2025.1520316. PMID: 40007996; PMCID: PMC11850343.

Ostendorf AP, Loddenkemper T, **Morgan LA**, Appavu B, Farias-Moeller R, Harrar D, Press C, Abend NS, Gaillard WD, Bai S, Eisner M, McHenry L, Kroshus E, Vannatta K, Goodkin HP. Treating seizures faster: The quality improvement in time to treat status epilepticus (QuITT-SE) multicenter randomized stepped wedge clinical trial protocol. *Contemp Clin Trials*. 2025 Feb 8;151:107831. doi: 10.1016/j.cct.2025.107831. Epub ahead of print. PMID: 39929261.

Horvat CM, Barda AJ, Perez Claudio E, Au AK, Bauman A, Li Q, Li R, Munjal N, **Wainwright MS**, Boonchalermvichien T, Hochheiser H, Clark RSB. Interoperable Models for Identifying Critically Ill Children at Risk of Neurologic Morbidity. *JAMA Netw Open*. 2025 Feb 3;8(2):e2457469. doi: 10.1001/jamanetworkopen.2024.57469. PMID: 39903466; PMCID: PMC11795326.

Beqiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal ES, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Correction: Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data. *Neurocrit Care*. 2025 Feb;42(1):319-320. doi: 10.1007/s12028-024-02097-w. Erratum for: *Neurocrit Care*. 2023 Dec;39(3):593-599. doi: 10.1007/s12028-023-01846-7. PMID: 39187690.

Bacci JL, Nwogu IB, Zarea S, Guignet M, White HS, Stergachis A, Ems D, **Novotny EJ**. A community pharmacist intervention for people living with epilepsy. *J Am Pharm Assoc* (2003). 2025 Jan-Feb;65(1):102275. doi: 10.1016/j.japh.2024.102275. Epub 2024 Oct 28. PMID: 39490530.

Sun LR, Lee S, **Lee-Eng J**, Barry M, Galardi MM, Harrar D, Hassanein SM, Rivkin MJ, Torres M, Wilson JL, **Amlie-Lefond C**, Williams K; as the International Pediatric Stroke Study and the Pediatric Neurocritical Care Research Group. Tenecteplase for the Treatment of Pediatric Arterial Ischemic Stroke: A Safety Surveillance Report. *Neurology*. 2025 Feb 11;104(3):e210310. doi: 10.1212/WNL.0000000000210310. Epub 2025 Jan 13. PMID: 39805054.

Schwarz AC, Breuner CC, **Blume HK**. Retrospective Study of the Efficacy of Biofeedback Therapy for Pediatric Persistent Posttraumatic Headache. *J Child Neurol*. 2025 Jan 17:8830738241312942. doi: 10.1177/08830738241312942. Epub ahead of print. PMID: 39819114.

Vitanza NA, Ronsley R, Choe M, Seidel K, Huang W, Rawlings-Rhea SD, Beam M, Steinmetzer L, Wilson AL, Brown C, Beebe A, Lindgren C, Gustafson JA, Wein A, Holtzclaw S, Hoepfner C, Goldstein HE, Browd SR, Hauptman JS, Lee A, Ojemann JG, Crotty EE, Leary SES, Perez FA, Wright JN, Alonso MM, Dun MD, Foster JB, Hurst D, Kong A, Thomsen A, Orentas RJ, Albert CM, Pinto N, Annesley C, Gardner RA, Ho O, Patabhi S, **Gust J**, Wendler JP, Park JR, Jensen MC. Intracerebroventricular B7-H3-targeting CAR T cells for diffuse intrinsic pontine glioma: a phase 1 trial. *Nat Med*. 2025 Jan 7. doi: 10.1038/s41591-024-03451-3. Epub ahead of print. PMID: 39775044.

2024

Campbell LM, Fennema-Notestine C, Sundermann EE, Barrett A, Bondi MW, Ellis RJ, Franklin D, Gelman B, Gilbert PE, Grant I, Heaton RK, Moore DJ, Morgello S, Letendre S, **Patel PB**, Roesch S, Moore RC. The prefrontal cortex, but not the medial temporal lobe, is associated with episodic memory in middle-aged persons with HIV. *J Int Neuropsychol Soc*. 2024 Dec;30(10):966-976. doi: 10.1017/S1355617724000596. Epub 2024 Nov 15. PMID: 39545285; PMCID: PMC11735296

Goldstein HE, Poliakov A, Barry D, Warner MH, Shaw DW, **Novotny EJ**, **Saneto RP**, **Patrick KE**, **Bowen KS**, Hauptman JS, Ojemann JG, Shurtleff HA. Functional MRI for bilingual epilepsy surgery patients: serving a diverse pediatric cohort. *J Neurosurg Pediatr*. 2024 Dec 27:1-10. doi: 10.3171/2024.9.PEDS2471. Epub ahead of print. PMID: 39729583.

Barcia Aguilar C, Amengual-Gual M, Brenton JN, Chapman KE, Clark J, Gaillard WD, Goldstein JL, Goodkin HP, Kahoud R, Lai YC, Mikati MA, **Morgan LA**, Payne ET, Press CA, Reece L, Sands TT, Sannagowdara K, Sheehan T, Shellhaas RA, Tasker RC, **Wainwright MS**, Zhang B, Loddenkemper T; pSERG. Lack of association of first and second-line medication dosing and progression to refractory status epilepticus in children. *Seizure*. 2024 Dec;123:133-141. doi: 10.1016/j.seizure.2024.10.017. Epub 2024 Oct 28. PMID: 39550933.

Dickman JA, Keene JC, **Natarajan N**, **Morgan LA**, Carrasco M. A Comparison of Treatment Practices for Newborn Seizure Management Across Level II and III Neonatal Intensive Care Units in the United States. *Pediatr Neurol*. 2024 Dec;161:108-112. doi: 10.1016/j.pediatrneurol.2024.09.006. Epub 2024 Sep 14. PMID: 39368245.

Edmonds B, Ngo JP, Groves A, Reyes B, Gott RA, Chia DJ, Mirbaha H, Magaki S, Khanlou N, Pineles SL, Salamon N, Thompson RM, Newman M, Rajaraman RR, Hussain SA, Fallah A, Russell B, Nariai H. Multi-disciplinary team approach for pediatric hemimegalencephaly: Insights from a single institutional case series. *Epilepsia Open*. 2024 Dec;9(6):2510-2517. doi: 10.1002/epi4.13079. Epub 2024 Oct 23. PMID: 39442533; PMCID: PMC11633684.

Hidalgo-Gutierrez A, Shintaku J, Ramon J, Barriocanal-Casado E, Pesini A, **Saneto RP**, Garrabou G, Milisenda JC, Matas-Garcia A, Gort L, Ugarteburu O, Gu Y, Koganti L, Wang T, Tadesse S, Meneri M, Sciacco M, Wang S, Tanji K, Horwitz MS, Dorschner MO, Mansukhani M, Comi GP, Ronchi D, Marti R, Ribes A, Tort F, Hirano M. Guanylate Kinase 1 Deficiency: A Novel and Potentially Treatable Mitochondrial DNA Depletion/Deletions Disease. *Ann Neurol*. 2024 Dec;96(6):1209-1224. doi: 10.1002/ana.27071. Epub 2024 Sep 4. PMID: 39230499; PMCID: PMC11563867.

Patel PB, Prince DK, Bolenzius J, Ch'en P, Chiarella J, Kolind S, Vavasour I, Pedersen T, Levendovszky SR, Spudich S, Marra C, Paul R. Medical comorbidities and lower myelin content are associated with poor cognition in young adults with perinatally acquired HIV. *AIDS*. 2024 Nov 15;38(14):1932-1939. doi: 10.1097/QAD.0000000000003989. Epub 2024 Aug 7. PMID: 39110577; PMCID: PMC11524773.

Seerha H, Maher K, Barry D, **Amalie-Lefond C**. Anticoagulation for Septic Cerebral Venous Thrombosis in Childhood. *J Pediatr Hematol Oncol*. 2024 Nov 1;46(8):404-408. doi: 10.1097/MPH.0000000000002949. Epub 2024 Sep 9. PMID: 39262384.

Sepulveda CJ, Walsh E, Carlin K, **Saneto RP**. Using a Previsit Questionnaire for Initial Visits in a Pediatric Mitochondrial Clinic: Perspectives of Parents, a Specialty Physician, and a Clinical Coordinator. *J Child Neurol*. 2024 Nov;39(13-14):453-460. doi: 10.1177/08830738241278388. Epub 2024 Sep 13. PMID: 39268553.

Bacci JL, Nwogu IB, Zarea S, Guignet M, White HS, Stergachis A, Ems D, **Novotny EJ**. A community pharmacist intervention for people living with epilepsy. *J Am Pharm Assoc (2003)*. 2024 Oct 25:102275. doi: 10.1016/j.japh.2024.102275. Epub ahead of print. PMID: 39490530.

Barcia Aguilar C, Amengual-Gual M, Brenton JN, Chapman KE, Clark J, Gaillard WD, Goldstein JL, Goodkin HP, Kahoud R, Lai YC, Mikati MA, **Morgan LA**, Payne ET, Press CA, Reece L, Sands TT, Sannagowdara K, Sheehan T, Shellhaas RA, Tasker RC, **Wainwright MS**, Zhang B, Loddenkemper T; pSERG. Lack of association of first and second-line medication dosing and progression to refractory status epilepticus in children. *Seizure*. 2024 Oct 28;123:133-141. doi: 10.1016/j.seizure.2024.10.017. Epub ahead of print. PMID: 39550933

Nakai R, Varnum S, Field RL, Shi H, Giwa R, Jia W, Krysa SJ, Cohen EF, Borcharding N, **Saneto RP**, Tsai RC, Suganuma M, Ohta H, Yokota T, Brestoff JR. Mitochondria transfer-based therapies reduce the morbidity and mortality of Leigh syndrome. *Nat Metab*. 2024 Oct;6(10):1886-1896. doi: 10.1038/s42255-024-01125-5. Epub 2024 Sep 2. PMID: 39223312.

Pediatric Neurology Faculty Publications 2014-2025

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

Patterson Gentile C, Rosenthal S, **Blume H**, Rastogi RG, McVige J, Bicknese A, Ladak A, Zaveri H, Greene K, Barlow K. American Headache Society white paper on treatment of post-traumatic headache from concussion in youth. *Headache*. 2024 Oct;64(9):1148-1162. doi: 10.1111/head.14795. Epub 2024 Jul 29. PMID: 39073141.

Edmonds B, Ngo JP, Groves A, Reyes B, Gott RA, Chia DJ, Mirbaha H, Magaki S, Khanlou N, Pineles SL, Salamon N, Thompson RM, Newman M, Rajaraman RR, Hussain SA, Fallah A, Russell B, Nariai H. Multi-disciplinary team approach for pediatric hemimegalencephaly: Insights from a single institutional case series. *Epilepsia Open*. 2024 Oct 23. doi: 10.1002/epi4.13079. Epub ahead of print. PMID: 39442533.

Farach LS, Richard MA, Wulsin AC, Bebin EM, Krueger DA, Sahin M, Porter BE, McPherson TO, Peters JM, O'Kelley S, Taub KS, Rajaraman R, **Randle SC**, McClintock WM, Koenig MK, Frost MD, Werner K, Nolan DA, Wong M, Cutter G, Northrup H, Au KS; PREVeNT Study Group. Drug-Resistant Epilepsy in Tuberous Sclerosis Complex Is Associated With TSC2 Genotype: More Findings From the Preventing Epilepsy Using Vigatrin (PREVeNT) Trial. *Pediatr Neurol*. 2024 Oct;159:62-71. doi: 10.1016/j.pediatrneurol.2024.06.012. Epub 2024 Jul 4. PMID: 39142021.

Dickman JA, Keene JC, **Natarajan N**, **Morgan LA**, Carrasco M. A Comparison of Treatment Practices for Newborn Seizure Management Across Level II and III Neonatal Intensive Care Units in the United States. *Pediatr Neurol*. 2024 Sep 14;161:108-112. doi: 10.1016/j.pediatrneurol.2024.09.006. Epub ahead of print. PMID: 39368245.

Horvat CM, Barda AJ, Claudio EP, Au AK, Bauman A, Li Q, Li R, Munjal N, **Wainwright M**, Boonchalermvichien T, Hochheiser H, Clark RSB. Development, External Validation, and Biomolecular Corroboration of Interoperable Models for Identifying Critically Ill Children at Risk of Neurologic Morbidity. *medRxiv [Preprint]*. 2024 Sep 18:2024.09.17.24313649. doi: 10.1101/2024.09.17.24313649. PMID: 39371157; PMCID: PMC11451808.

Gust J, Shah NN. CARDinal signs of variant ICANS. *Blood*. 2024 Sep 26;144(13):1352-1354. doi: 10.1182/blood.2024025664. PMID: 39325485.

Patel ND, **Wainwright MS**, Moore A, Suz P, Muangman S, Vavilala MS. Establishing normal Lindegaard Ratio in healthy children 10-16 years of age. *Childs Nerv Syst*. 2024 Sep;40(9):2829-2833. doi: 10.1007/s00381-024-06467-9. Epub 2024 Jun 21. PMID: 38907117.

Sepulveda CJ, Walsh E, Carlin K, **Saneto RP**. Using a Previsit Questionnaire for Initial Visits in a Pediatric Mitochondrial Clinic: Perspectives of Parents, a Specialty Physician, and a Clinical Coordinator. *J Child Neurol*. 2024 Sep 13:8830738241278388. doi: 10.1177/08830738241278388. Epub ahead of print. PMID: 39268553.

Guignet M, Steve White H, **Novotny EJ**, Benedict Nwogu I, Zarea S, Stergachis A, Ems D, Bacci JL. Community Pharmacist-Centered training program improves confidence in delivering epilepsy care. *Epilepsy Behav*. 2024 Sep;158:109933. doi: 10.1016/j.yebeh.2024.109933. Epub 2024 Jul 5. PMID: 38970894.

Seerha H, Maher K, Barry D, **Amlie-Lefond C**. Anticoagulation for Septic Cerebral Venous Thrombosis in Childhood. *J Pediatr Hematol Oncol*. 2024 Sep 9. doi: 10.1097/MPH.0000000000002949. Epub ahead of print. PMID: 39262384.

Hidalgo-Gutierrez A, Shintaku J, Ramon J, Barriocanal-Casado E, Pesini A, **Saneto RP**, Garrabou G, Milisenda JC, Matas-Garcia A, Gort L, Ugarteburu O, Gu Y, Koganti L, Wang T, Tadesse S, Meneri M, Sciacco M, Wang S, Tanji K, Horwitz MS, Dorschner MO, Mansukhani M, Comi GP, Ronchi D, Marti R, Ribes A, Tort F, Hirano M. Guanylate Kinase 1 Deficiency: A Novel and Potentially Treatable Mitochondrial DNA Depletion/Deletions Disease. *Ann Neurol*. 2024 Sep 4. doi: 10.1002/ana.27071. Epub ahead of print. PMID: 39230499.

Nakai R, Varnum S, Field RL, Shi H, Giwa R, Jia W, Krysa SJ, Cohen EF, Borcharding N, **Saneto RP**, Tsai RC, Suganuma M, Ohta H, Yokota T, Brestoff JR. Mitochondria transfer-based therapies reduce the morbidity and mortality of Leigh syndrome. *Nat Metab*. 2024 Sep 2. doi: 10.1038/s42255-024-01125-5. Epub ahead of print. PMID: 39223312.

Beqiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal ES, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Correction: Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data.

Pediatric Neurology Faculty Publications 2014-2025

- Neurocrit Care. 2024 Aug 26. doi: 10.1007/s12028-024-02097-w. Epub ahead of print. Erratum for: Neurocrit Care. 2023 Dec;39(3):593-599. doi: 10.1007/s12028-023-01846-7. PMID: 39187690.
- Vitanza NA, Choe M, Brown C, Beebe A, Kong A, Rogers L, Jacob S, Mano E, Abuan K, Mgebroff S, Lindgren C, Gustafson JA, Wilson AL, Noll A, Ronsley R, Crotty EE, Leary SES, Foster JB, Pinto N, **Gust J**, Gardner RA, Park JR, Jensen MC. Locoregional CAR T Cells for the Treatment of CNS Tumors in Children: Investigational Drug Service Pharmacy Activities. J Hematol Oncol Pharm. 2024 Aug;14(4):148-154. PMID: 39238483; PMCID: PMC11375759.
- Patel PB**, Prince DK, Bolenzius J, Chen P, Chiarella J, Kolind S, Vavasour I, Pedersen T, Levendovszky SR, Spudich S, Marra C, Paul R. Medical comorbidities and lower myelin content are associated with poor cognition in young adults with perinatally acquired HIV. AIDS. 2024 Aug 7. doi: 10.1097/QAD.0000000000003989. Epub ahead of print. PMID: 39110577.
- Bose-Brill S, Hirabayashi K, Schwimmer E, Pajor NM, Rao S, Mejias A, Jhaveri R, Forrest CB, Bailey LC, Christakis DA, Thacker D, Hanley PC, **Patel PB**, Cogen JD, Block JP, Prahalad P, Lorman V, Lee GM; Researching COVID to Enhance Recovery consortium. Pediatric Nirmatrelvir/Ritonavir Prescribing Patterns During the COVID-19 Pandemic. Hosp Pediatr. 2024 Aug 1;14(8):e341-e348. doi: 10.1542/hpeds.2023-007132. PMID: 39076115; PMCID: PMC11287060.
- Hanin A, Jimenez AD, Gopaul M, Asbell H, Aydemir S, Basha MM, Batra A, Damien C, Day GS, Eka O, Eschbach K, Fatima S, Fields MC, Foreman B, Gerard EE, Gofton TE, Haider HA, Hantus ST, Hocker S, Jongeling A, Kalkach Aparicio M, Kandula P, Kang P, Kazazian K, Kellogg MA, Kim M, Lee JW, Marcuse LV, McGraw CM, Mohamed W, Orozco J, Pimentel CM, Punia V, Ramirez AM, Steriade C, Struck AF, Taraschenko O, Treister AK, **Wainwright MS**, Yoo JY, Zafar S, Zhou DJ, Zutshi D, Gaspard N, Hirsch LJ. Trends in management of patients with new-onset refractory status epilepticus (NORSE) from 2016 to 2023: An interim analysis. Epilepsia. 2024 Aug;65(8):e148-e155. doi: 10.1111/epi.18014. Epub 2024 Jun 5. PMID: 38837761.
- Ahn S, **Edmonds B**, Rajaraman RR, Rao LM, Hussain SA, Matsumoto JH, Sankar R, Salamon N, Fallah A, Nariai H. Bilateral centromedian nucleus of thalamus responsive neurostimulation for pediatric-onset drug-resistant epilepsy. Epilepsia. 2024 Aug;65(8):e131-e140. doi: 10.1111/epi.18031. Epub 2024 Jun 7. PMID: 38845459; PMCID: PMC11315622.
- Rael VE, Yano JA, Huizar JP, Slayden LC, Weiss MA, Turcotte EA, Terry JM, Zuo W, Thiffault I, Pastinen T, Farrow EG, Jenkins JL, Becker ML, Wong SC, Stevens AM, **Otten C**, Allenspach EJ, Bonner DE, Bernstein JA, Wheeler MT, Saxton RA; Undiagnosed Diseases Network; Liu B, Majer O, Barton GM. Large-scale mutational analysis identifies UNC93B1 variants that drive TLR-mediated autoimmunity in mice and humans. J Exp Med. 2024 Aug 5;221(8):e20232005. doi: 10.1084/jem.20232005. Epub 2024 May 23. PMID: 38780621; PMCID: PMC11116816.
- Harel T, Spicher C, Scheer E, Buchan JG, Cech J, Folland C, Frey T, Holtz AM, Innes AM, Keren B, Macken WL, Marcellis C, **Otten CE**, Paolucci SA, Petit F, Pfundt R, Pitceathly RDS, Rauch A, Ravenscroft G, Sanchez R, Steindl K, Tammer F, Tyndall A, Devys D, Vincent SD, Elpeleg O, Tora L. De novo variants in ATXN7L3 lead to developmental delay, hypotonia and distinctive facial features. Brain. 2024 Aug 1;147(8):2732-2744. doi: 10.1093/brain/awae160. PMID: 38753057.
- Patterson Gentile C, Rosenthal S, **Blume H**, Rastogi RG, McVige J, Bicknese A, Ladak A, Zaveri H, Greene K, Barlow K. American Headache Society white paper on treatment of post-traumatic headache from concussion in youth. Headache. 2024 Jul 29. doi: 10.1111/head.14795. Epub ahead of print. PMID: 39073141.
- 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 Jul 17;159:12-15. doi: 10.1016/j.pediatrneurol.2024.07.009. Epub ahead of print. PMID: 39094249.
- Guignet M, Steve White H, **Novotny EJ**, Benedict Nwogu I, Zarea S, Stergachis A, Ems D, Bacci JL. Community Pharmacist-Centered training program improves confidence in delivering epilepsy care. Epilepsy Behav. 2024 Jul 5;158:109933. doi: 10.1016/j.yebeh.2024.109933. Epub ahead of print. PMID: 38970894.
- Farach LS MD, Richard MA PhD, Wulsin AC MD, PhD, Bebin EM MD, Krueger DA MD, PhD, Sahin M MD, PhD, Porter BE MD, PhD, McPherson TO PhD, Peters JM MD, PhD, O'Kelley S PhD, Taub KS MD, Rajaraman R MD, **Randle SC** MD, McClintock WM MD, Koenig MK MD, Frost MD MD, Werner K MD, PhD, Nolan DA MD, Wong M MD, PhD, Cutter G PhD, Northrup H MD, Au KS PhD; PREVeNT Study Group. Drug-Resistant Epilepsy in Tuberous Sclerosis Complex Is

Pediatric Neurology Faculty Publications 2014-2025

Associated With TSC2 Genotype: More Findings From the Preventing Epilepsy Using Vigatin (PREVeNT) Trial. *Pediatr Neurol.* 2024 Jul 4;159:62-71. doi: 10.1016/j.pediatrneurol.2024.06.012. Epub ahead of print. PMID: 39142021.

Cuccurullo C, Cerulli Irelli E, Ugga L, Riva A, D'Amico A, Cabet S, Lesca G, Bilo L, Zara F, Iliescu C, Barca D, Fung F, Helbig K, Ortiz-Gonzalez X, Schelhaas HJ, Willemsen MH, van der Linden I, Canafoglia L, Courage C, Gommarschi S, Gonzalez-Alegre P, Bardakjian T, Syrbe S, Schuler E, Lemke JR, Vari S, Roende G, Bak M, Huq M, Powis Z, Johannesen KM, Hammer TB, Møller RS, Rabin R, Pappas J, Zupanc ML, Zadeh N, Cohen J, Naidu S, Krey I, **Saneto R**, Thies J, Licchetta L, Tinuper P, Bisulli F, Minardi R, Bayat A, Villeneuve N, Molinari F, Salimi Dafsari H, Moller B, Le Roux M, Houdayer C, Vecchi M, Mammi I, Fiorini E, Proietti J, Ferri S, Cantalupo G, Battaglia DI, Gambardella ML, Contaldo I, Brogna C, Trivisano M, De Dominicis A, Bova SM, Gardella E, Striano P, Coppola A. Clinical features and genotype-phenotype correlations in epilepsy patients with de novo DYNC1H1 variants. *Epilepsia.* 2024 Jul 2. doi: 10.1111/epi.18054. Epub ahead of print. PMID: 38953796.

Fink EL, Alcamo AM, Lovett M, Hartman M, Williams C, Garcia A, Rasmussen L, Pal R, Drury K, MackDiaz E, Ferrazzano PA, Dervan L, Appavu B, Snooks K, Stulce C, Rubin P, Pate B, Toney N, Robertson CL, **Wainwright MS**, Roa JD, Schober ME, Slomine BS. Corrigendum: Post-discharge outcomes of hospitalized children diagnosed with acute SARS-CoV-2 or MIS-C. *Front Pediatr.* 2024 Jul 1;12:1440609. doi: 10.3389/fped.2024.1440609. Erratum for: *Front Pediatr.* 2024 Feb 12;12:1340385. doi: 10.3389/fped.2024.1340385. PMID: 39011061; PMCID: PMC11247001.

Reecher HM, Bearden DJ, Koop JI, Berl MM, **Patrick KE**, Ailion AS. The changing landscape of electrical stimulation language mapping with subdural electrodes and stereoelectroencephalography for pediatric epilepsy: A literature review and commentary. *Epilepsia.* 2024 Jul;65(7):1879-1898. doi: 10.1111/epi.18009. Epub 2024 May 24. PMID: 38787551.

Siegel BI, **Gust J**. How Cancer Harms the Developing Brain: Long-Term Outcomes in Pediatric Cancer Survivors. *Pediatr Neurol.* 2024 Jul;156:91-98. doi: 10.1016/j.pediatrneurol.2024.03.031. Epub 2024 Apr 8. PMID: 38735088.

Patel ND, **Wainwright MS**, Moore A, Suz P, Muangman S, Vavilala MS. Establishing normal Lindegaard Ratio in healthy children 10-16 years of age. *Childs Nerv Syst.* 2024 Jun 21. doi: 10.1007/s00381-024-06467-9. Epub ahead of print. PMID: 38907117.

Ahn S, **Edmonds B**, Rajaraman RR, Rao LM, Hussain SA, Matsumoto JH, Sankar R, Salamon N, Fallah A, Nariai H. Bilateral centromedian nucleus of thalamus responsive neurostimulation for pediatric-onset drug-resistant epilepsy. *Epilepsia.* 2024 Jun 7. doi: 10.1111/epi.18031. Epub ahead of print. PMID: 38845459.

Hanin A, Jimenez AD, Gopaul M, Asbell H, Aydemir S, Basha MM, Batra A, Damien C, Day GS, Eka O, Eschbach K, Fatima S, Fields MC, Foreman B, Gerard EE, Gofton TE, Haider HA, Hantus ST, Hocker S, Jongeling A, Kalkach Aparicio M, Kandula P, Kang P, Kazazian K, Kellogg MA, Kim M, Lee JW, Marcuse LV, McGraw CM, Mohamed W, Orozco J, Pimentel CM, Punia V, Ramirez AM, Steriade C, Struck AF, Taraschenko O, Treister AK, **Wainwright MS**, Yoo JY, Zafar S, Zhou DJ, Zutshi D, Gaspard N, Hirsch LJ. Trends in management of patients with new-onset refractory status epilepticus (NORSE) from 2016 to 2023: An interim analysis. *Epilepsia.* 2024 Jun 5. doi: 10.1111/epi.18014. Epub ahead of print. PMID: 38837761.

Lin V, Tian C, Wahlster S, **Castillo-Pinto C**, Mainali S, Johnson NJ. Temperature Control in Acute Brain Injury: An Update. *Semin Neurol.* 2024 Jun;44(3):308-323. doi: 10.1055/s-0044-1785647. Epub 2024 Apr 9. PMID: 38593854.

Francoeur C, Alcamo AM, Robertson CL, **Wainwright MS**, Roa JD, Lovett ME, Stulce C, Yacoub M, Potera RM, Zivick E, Holloway A, Nagpal A, Wellnitz K, Even KM, Brunow de Carvalho W, Rodriguez IS, Schwartz SP, Walker TC, Campos-Miño S, Dervan LA, Geneslaw AS, Sewell TB, Pryce P, Silver WG, Lin JE, Vargas WS, Topjian A, McGuire JL, Domínguez Rojas JA, Tasayco-Muñoz J, Hong SJ, Muller WJ, Doerfler M, Williams CN, Drury K, Bhagat D, Nelson A, Price D, Dapul H, Santos L, Kahoud R, Appavu B, Guilliams KP, Agner SC, Walson KH, Rasmussen L, Pal R, Janas A, Ferrazzano P, Farias-Moeller R, Snooks KC, Chang CH, Iolster T, Erklauer JC, Jorro Baron F, Wassmer E, Yoong M, Jardine M, Mohammad Z, Deep A, Kendirli T, Lidsky K, Dallefeld S, Flockton H, Agrawal S, Siruguppa KS, Waak M, Gutiérrez-Mata A, Butt W, Bogantes-Ledezma S, Sevilla-Acosta F, Umaña-Calderón A, Ulate-Campos A, Yock-Corrales A, Talisa VB, Kanthimathinathan HK, Schober ME, Fink EL; Global Consortium Study of Neurologic Dysfunction in COVID-19 (GCS-NeuroCOVID) Investigators. Severe Pediatric Neurological Manifestations With SARS-CoV-2 or MIS-C Hospitalization and New Morbidity. *JAMA Netw Open.* 2024 Jun 3;7(6):e2414122. doi: 10.1001/jamanetworkopen.2024.14122. PMID: 38857050; PMCID: PMC11165382.

Fullerton HJ, Hills NK, Wintermark M, Dlamini N, **Amlie-Lefond C**, Dowling MM, Jordan LC, Bernard TJ, Friedman NR, Elkind MSV, Grose C; VIPS II Investigators. Evidence of varicella zoster virus (VZV) reactivation in children with arterial

Pediatric Neurology Faculty Publications 2014-2025

ischemic stroke: Results of the VIPS II Study. medRxiv [Preprint]. 2024 May 28;2024.05.26.24307958. doi: 10.1101/2024.05.26.24307958. PMID: 38853955; PMCID: PMC11160817.

Reecher HM, Bearden DJ, Koop JI, Berl MM, **Patrick KE**, Ailion AS. The changing landscape of electrical stimulation language mapping with subdural electrodes and stereoelectroencephalography for pediatric epilepsy: A literature review and commentary. *Epilepsia*. 2024 May 24. doi: 10.1111/epi.18009. Epub ahead of print. PMID: 38787551.

Ch'en P, **Patel PB**, Ramirez M. Caregivers' and Health Care Providers' Cultural Perceptions of and Experiences With Latino Patients With Dementia. *Neurol Clin Pract*. 2024 Aug;14(4):e200307. doi: 10.1212/CPJ.0000000000200307. Epub 2024 May 23. PMID: 38855714; PMCID: PMC11157425.

Harel T, Spicher C, Scheer E, Buchan JG, Cech J, Folland C, Frey T, Holtz AM, Innes AM, Keren B, Macken WL, Marcellis C, **Otten CE**, Paolucci SA, Petit F, Pfundt R, Pitceathly RDS, Rauch A, Ravenscroft G, Sanchez R, Steindl K, Tammer F, Tyndall A, Devys D, Vincent SD, Elpeleg O, Tora L. De novo variants in ATXN7L3 lead to developmental delay, hypotonia and distinctive facial features. *Brain*. 2024 May 16;awae160. doi: 10.1093/brain/awae160. Epub ahead of print. PMID: 38753057.

Numis AL, Glass HC, Comstock BA, Gonzalez F, Maitre NL, Massey SL, Mayock DE, Mietzsch U, **Natarajan N**, Sokol GM, Bonifacio S, Van Meurs K, Thomas C, Ahmad K, Heagerty P, Juul SE, Wu YW, Wusthoff CJ. Relationship of Neonatal Seizure Burden Before Treatment and Response to Initial Antiseizure Medication. *J Pediatr*. 2024 May;268:113957. doi: 10.1016/j.jpeds.2024.113957. Epub 2024 Feb 13. PMID: 38360261.

Bonda D, Kelly KA, Boop S, Feroze AH, **Randle SC**, Bindschadler M, Marashly A, Owens J, **Lockrow J**, Bozarth X, **Novotny E**, Friedman S, Goldstein HE, Grannan BL, Durfy S, Ojemann JG, Ko AL, Hauptman JS. Deep Brain Stimulation of Bilateral Centromedian Thalamic Nuclei in Pediatric Patients with Lennox-Gastaut Syndrome: An Institutional Experience. *World Neurosurg*. 2024 May;185:e631-e639. doi: 10.1016/j.wneu.2024.02.099. Epub 2024 Feb 24. PMID: 38403017.

Panchavati S, Daida A, **Edmonds B**, Miyakoshi M, Oana S, Ahn SS, Arnold C, Salamon N, Sankar R, Fallah A, Speier W, Nariai H. Uncovering spatiotemporal dynamics of the corticothalamic network at ictal onset. *Epilepsia*. 2024 Apr 25. doi: 10.1111/epi.17990. Epub ahead of print. PMID: 38662128.

Mietzsch U, Kolnik SE, Wood TR, **Natarajan N**, Gonzalez FF, Glass H, Mayock DE, Bonifacio SL, Van Meurs K, Comstock BA, Heagerty PJ, Wu TW, Wu YW, Juul SE; HEAL Trial Study Group. Evolution of the Sarnat exam and association with 2-year outcomes in infants with moderate or severe hypoxic-ischaemic encephalopathy: a secondary analysis of the HEAL Trial. *Arch Dis Child Fetal Neonatal Ed*. 2024 Apr 18;109(3):308-316. doi: 10.1136/archdischild-2023-326102. PMID: 38071538; PMCID: PMC11031347.

Lin V, Tian C, Wahlster S, **Castillo-Pinto C**, Mainali S, Johnson NJ. Temperature Control in Acute Brain Injury: An Update. *Semin Neurol*. 2024 Apr 9. doi: 10.1055/s-0044-1785647. Epub ahead of print. PMID: 38593854.

Siegel BI, **Gust J**. How Cancer Harms the Developing Brain: Long-Term Outcomes in Pediatric Cancer Survivors. *Pediatr Neurol*. 2024 Apr 8;156:91-98. doi: 10.1016/j.pediatrneurol.2024.03.031. Epub ahead of print. PMID: 38735088.

Chatterjee JH, **Blume HK**. Triptans in the Acute Migraine Management of Children and Adolescents: An Update. *Curr Pain Headache Rep*. 2024 Apr 6. doi: 10.1007/s11916-024-01213-x. Epub ahead of print. PMID: 38581536.

Terman SW, Kirkpatrick L, Kerr WT, **Akiyama LF**, Baajour W, Atilgan D, Dorotan MKC, Choi HW, French JA. Challenges and directions in epilepsy diagnostics and therapeutics: Proceedings of the 17th Epilepsy Therapies and Diagnostics Development conference. *Epilepsia*. 2024 Apr;65(4):846-860. doi: 10.1111/epi.17875. Epub 2024 Jan 5. PMID: 38135921; PMCID: PMC11018495.

Terman SW, Kirkpatrick L, **Akiyama LF**, Baajour W, Atilgan D, Dorotan MKC, Choi HW, French JA. Current state of the epilepsy drug and device pipeline. *Epilepsia*. 2024 Apr;65(4):833-845. doi: 10.1111/epi.17884. Epub 2024 Feb 12. PMID: 38345387; PMCID: PMC11018510.

Williams NC, **Morgan LA**, Friedman J, Siegler J. Ketamine Efficacy for Management of Status Epilepticus: Considerations for Prehospital Clinicians. *Air Med J*. 2024 Mar-Apr;43(2):84-89. doi: 10.1016/j.amj.2023.09.011. Epub 2023 Oct 11. PMID: 38490790.

Pediatric Neurology Faculty Publications 2014-2025

Trunfio M, Tang B, Okwuegbuna O, Iudicello JE, Bharti A, Moore DJ, Gelman BB, Morgello S, **Patel PB**, Rubin LH, Ances BM, Gianella S, Heaton RK, Ellis RJ, Letendre SL. Longitudinal analysis of CSF HIV RNA in untreated people with HIV: Identification of CSF controllers. *J Med Virol*. 2024 Mar;96(3):e29550. doi: 10.1002/jmv.29550. PMID: 38511593.

Dang UJ, Damsker JM, Guglieri M, Clemens PR, **Perlman SJ**, Smith EC, Horrocks I, Finkel RS, Mah JK, Deconinck N, Goemans NM, Haberlová J, Straub V, Mengle-Gaw L, Schwartz BD, Harper A, Shieh PB, De Waele L, Castro D, Yang ML, Ryan MM, McDonald CM, Tulinius M, Webster RI, Mcmillan HJ, Kuntz N, Rao VK, Baranello G, Spinty S, Childs AM, Sbrocchi AM, Selby KA, Monduy M, Nevo Y, Vilchez JJ, Nascimento-Osorio A, Niks EH, De Groot IJM, Katsaloulis M, Van Den Anker JN, Ward LM, Leinonen M, D'Alessandro AL, Hoffman EP. Efficacy and Safety of Vamorolone Over 48 Weeks in Boys With Duchenne Muscular Dystrophy: A Randomized Controlled Trial. *Neurology*. 2024 Mar 12;102(5):e208112. doi: 10.1212/WNL.0000000000208112. Epub 2024 Feb 9. PMID: 38335499.

Morgan LA, Sprigg BN, Barry D, Hrachovec JB, **Novotny EJ**, **Akiyama LF**, Allar N, Matlock JK, Dervan LA. Reducing Time to Electroencephalography in Pediatric Convulsive Status Epilepticus: A Quality Improvement Initiative. *Pediatr Neurol*. 2024 Mar;152:169-176. doi: 10.1016/j.pediatrneurol.2024.01.006. Epub 2024 Jan 8. PMID: 38295718.

Joshi CN, Karakas C, Eschbach K, Samanta D, Auguste K, Desai V, Singh R, McGoldrick P, Wolf S, Abel TJ, **Novotny E**, Oluigbo C, Reddy SB, Alexander A, Price A, Reeders P, Mcnamara N, Romanowski EF, Mutchnick I, Ostendorf AP, Shaikhouni A, Knox A, Aungaroon G, Olaya J, Muh CR. Pediatric neuromodulation for drug-resistant epilepsy: Survey of current practices, techniques, and outcomes across US epilepsy centers. *Epilepsia Open*. 2024 Feb 29. doi: 10.1002/epi4.12902. Epub ahead of print. PMID: 38421143

Bonda D, Kelly KA, Boop S, Feroze AH, **Randle SC**, Bindschadler M, Marashly A, Owens J, **Lockrow J**, Bozarth X, **Novotny E**, Friedman S, Goldstein HE, Grannan BL, Durfy S, Ojemann JG, Ko AL, Hauptman JS. Deep Brain Stimulation of Bilateral Centromedian Thalamic Nuclei in Pediatric Patients with Lennox-Gastaut Syndrome: An Institutional Experience. *World Neurosurg*. 2024 Feb 23:S1878-8750(24)00312-7. doi: 10.1016/j.wneu.2024.02.099. Epub ahead of print. PMID: 38403017.

Beqiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal ES, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Correction: Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data. *Neurocrit Care*. 2024 Feb;40(1):382-383. doi: 10.1007/s12028-023-01894-z. Erratum for: *Neurocrit Care*. 2023 Dec;39(3):593-599. PMID: 38057543.

Guerrero-Gonzalez JM, Kirk GR, Birn R, Bigler ED, **Bowen K**, Broman AT, Rosario BL, Butt W, Beers SR, Bell MJ, Alexander AL, Ferrazzano PA; ADAPT MRI Biomarkers Investigators. Multi-modal MRI of hippocampal morphometry and connectivity after pediatric severe TBI. *Brain Imaging Behav*. 2024 Feb;18(1):159-170. doi: 10.1007/s11682-023-00818-x. Epub 2023 Nov 13. PMID: 37955810; PMCID: PMC10844146.

Numis AL, Glass HC, Comstock BA, Gonzalez F, Maitre NL, Massey SL, Mayock DE, Mietzsch U, **Natarajan N**, Sokol GM, Bonifacio S, Van Meurs K, Thomas C, Ahmad K, Heagerty P, Juul SE, Wu YW, Wusthoff CJ. Relationship of Neonatal Seizure Burden Before Treatment and Response to Initial Antiseizure Medication. *J Pediatr*. 2024 Feb 13;268:113957. doi: 10.1016/j.jpeds.2024.113957. Epub ahead of print. PMID: 38360261.

Fink EL, Alcamo AM, Lovett M, Hartman M, Williams C, Garcia A, Rasmussen L, Pal R, Drury K, MackDiaz E, Ferrazzano PA, Dervan L, Appavu B, Snooks K, Stulce C, Rubin P, Pate B, Toney N, Robertson CL, **Wainwright MS**, Roa JD, Schober ME, Slomine BS. Post-discharge outcomes of hospitalized children diagnosed with acute SARS-CoV-2 or MIS-C. *Front Pediatr*. 2024 Feb 12;12:1340385. doi: 10.3389/fped.2024.1340385. PMID: 38410766; PMCID: PMC10895015.

Terman SW, Kirkpatrick L, **Akiyama LF**, Baajour W, Atilgan D, Dorotan MKC, Choi HW, French JA. Current state of the epilepsy drug and device pipeline. *Epilepsia*. 2024 Feb 12. doi: 10.1111/epi.17884. Epub ahead of print. PMID: 38345387.

Liu Y, Massenburg BB, **Otten CE**, Osorio MB, Lewis SP, Hottovy J, Tse RW. Selective nerve transfers to restore shoulder abduction and flexion in acute flaccid myelitis: A case report. *Microsurgery*. 2024 Jan;44(1):e31104. doi: 10.1002/micr.31104. Epub 2023 Aug 30. PMID: 37646277.

Goel K, Phillips HW, Chen JS, Ngo J, **Edmonds B**, Ha PX, Wang A, Weil A, Russell BE, Salamon N, Nariai H, Fallah A. Hemispheric epilepsy surgery for hemimegalencephaly: The UCLA experience. *Epilepsia*. 2024 Jan;65(1):57-72. doi: 10.1111/epi.17807. Epub 2023 Nov 11. PMID: 37873610

Pediatric Neurology Faculty Publications 2014-2025

Patrick KE, Kroshus E, Boyle LN, Wang J, Binjolkar M, Ebel BE, Rivara FP. Driving characteristics of young adults prior to and following concussion. *Traffic Inj Prev.* 2024;25(1):14-19. doi: 10.1080/15389588.2023.2250493. Epub 2023 Nov 30. PMID: 37728546; PMCID: PMC10872858.

Zaraa S, Steve White H, Stergachis A, **Novotny EJ Jr**, Protos C, Simic G, Bacci JL. Using design thinking to strengthen the community pharmacist's role in epilepsy care. *Epilepsy Behav.* 2024 Jan;150:109542. doi: 10.1016/j.yebeh.2023.109542. Epub 2023 Nov 29. PMID: 38035539.

Turner AD, Streb MM, Ouyang A, **Leonard SS**, Hall TA, Bosworth CC, Williams CN, Guerriero RM, Hartman ME, Said AS, Guilliama KP. Long-term Neurobehavioral and Functional Outcomes of Pediatric Extracorporeal Membrane Oxygenation Survivors. *ASAIO J.* 2024 Jan 12. doi: 10.1097/MAT.0000000000002135. Epub ahead of print. PMID: 38207105.

Maheer K, **Amlie-Lefond C**. Cow's Milk Anemia in Childhood Manifesting as Severe Cerebral Venous Sinus Thrombosis. *J Child Neurol.* 2024 Jan 23;8830738241227064. doi: 10.1177/08830738241227064. Epub ahead of print. PMID: 38263607

2023

Wang S, Wang B, Drury V, Drake S, Sun N, Alkhairi H, Arbelaes J, Duhn C; Tourette International Collaborative Genetics (TIC Genetics)(**includes Dararat Mingbunjerdasuk**); Bal VH, Langley K, Martin J, Hoekstra PJ, Dietrich A, Xing J, Heiman GA, Tischfield JA, Fernandez TV, Owen MJ, O'Donovan MC, Thapar A, State MW, Willsey AJ. Rare X-linked variants carry predominantly male risk in autism, Tourette syndrome, and ADHD. *Nat Commun.* 2023 Dec 6;14(1):8077. doi: 10.1038/s41467-023-43776-0. PMID: 38057346; PMCID: PMC10700338.

Beqiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal ES, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data. *Neurocrit Care.* 2023 Dec;39(3):593-599. doi: 10.1007/s12028-023-01846-7. Epub 2023 Sep 13. Erratum in: *Neurocrit Care.* 2023 Dec 6; PMID: 37704934.

Beqiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal ES, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Correction: Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data. *Neurocrit Care.* 2023 Dec 6. doi: 10.1007/s12028-023-01894-z. Epub ahead of print. Erratum for: *Neurocrit Care.* 2023 Dec;39(3):593-599. PMID: 38057543.

Patrick KE, Kroshus E, Boyle LN, Wang J, Binjolkar M, Ebel BE, Rivara FP. Driving characteristics of young adults prior to and following concussion. *Traffic Inj Prev.* 2024;25(1):14-19. doi: 10.1080/15389588.2023.2250493. Epub 2023 Nov 30. PMID: 37728546.

Terman SW, Kirkpatrick L, Kerr WT, **Akiyama LF**, Baajour W, Atilgan D, Dorotan MKC, Choi HW, French JA. Challenges and directions in epilepsy diagnostics and therapeutics: proceedings of the 17th Epilepsy Therapies and Diagnostics Development conference. *Epilepsia.* 2023 Dec 22. doi: 10.1111/epi.17875. Epub ahead of print. PMID: 38135921.

Bogetz JF, **Natarajan N**, Hauer J, Ramirez JM. Appreciating the Abilities of Children With Severe Neurologic Impairment. *Hosp Pediatr.* 2023 Dec 1;13(12):e392-e394. doi: 10.1542/hpeds.2023-007463. PMID: 37946661; PMCID: PMC10656431.

Foreman B, Kapinos G, **Wainwright MS**, Ngwenya LB, O'Phelan KH, LaRovere KL, Kirschen MP, Appavu B, Lazaridis C, Alkhachroum A, Maciel CB, Amorim E, Chang JJ, Gilmore EJ, Rosenthal ES, Park S. Practice Standards for the Use of Multimodality Neuromonitoring: A Delphi Consensus Process. *Crit Care Med.* 2023 Dec 1;51(12):1740-1753. doi: 10.1097/CCM.0000000000006016. Epub 2023 Nov 16. PMID: 37607072.

Zaraa S, Steve White H, Stergachis A, **Novotny EJ Jr**, Protos C, Simic G, Bacci JL. Using design thinking to strengthen the community pharmacist's role in epilepsy care. *Epilepsy Behav.* 2023 Nov 28;150:109542. doi: 10.1016/j.yebeh.2023.109542. Epub ahead of print. PMID: 38035539

Pediatric Neurology Faculty Publications 2014-2025

Glass HC, Numis AL, Comstock BA, Gonzalez FF, Mietzsch U, Bonifacio SL, Massey S, Thomas C, **Natarajan N**, Mayock DE, Sokol GM, Van Meurs KP, Ahmad KA, Maitre N, Heagerty PJ, Juul SE, Wu YW, Wusthoff CJ. Association of EEG Background and Neurodevelopmental Outcome in Neonates With Hypoxic-Ischemic Encephalopathy Receiving Hypothermia. *Neurology*. 2023 Nov 27;101(22):e2223-e2233. doi: 10.1212/WNL.0000000000207744. PMID: 37816642; PMCID: PMC10727206.

Mietzsch U, Kolnik SE, Wood TR, **Natarajan N**, Gonzalez FF, Glass H, Mayock DE, Bonifacio SL, Van Meurs K, Comstock BA, Heagerty PJ, Wu TW, Wu YW, Juul SE; HEAL Trial Study Group. Evolution of the Sarnat exam and association with 2-year outcomes in infants with moderate or severe hypoxic-ischaemic encephalopathy: a secondary analysis of the HEAL Trial. *Arch Dis Child Fetal Neonatal Ed*. 2023 Nov 27:fetalneonatal-2023-326102. doi: 10.1136/archdischild-2023-326102. Epub ahead of print. PMID: 38071538.

Uppal P, Smith J, **Castillo-Pinto C**, Wells EM, Jayakar A, Harrar D. Glial Fibrillary Acidic Protein (GFAP) Astrocytopathy: An Emerging Cause of Meningoencephalomyelitis in Children and Adolescents. *J Child Neurol*. 2023 Nov 20:8830738231214301. doi: 10.1177/08830738231214301. Epub ahead of print. PMID: 37981797.

Guerrero-Gonzalez JM, Kirk GR, Birn R, Bigler ED, **Bowen K**, Broman AT, Rosario BL, Butt W, Beers SR, Bell MJ, Alexander AL, Ferrazzano PA; ADAPT MRI Biomarkers Investigators. Multi-modal MRI of hippocampal morphometry and connectivity after pediatric severe TBI. *Brain Imaging Behav*. 2023 Nov 13. doi: 10.1007/s11682-023-00818-x. Epub ahead of print. PMID: 37955810.

Bogetz JF, **Natarajan N**, Hauer J, Ramirez JM. Appreciating the Abilities of Children With Severe Neurologic Impairment. *Hosp Pediatr*. 2023 Nov 10;13(12):e392-4. doi: 10.1542/hpeds.2023-007463. Epub ahead of print. PMID: 37946661; PMCID: PMC10656431.

Benedetti GM, **Wainwright MS**. Commentary: Characteristics and Outcomes of Children with Cerebral Sinus Venous Thrombosis. *Neurocrit Care*. 2023 Oct;39(2):306-308. doi: 10.1007/s12028-023-01787-1. Epub 2023 Jul 12. PMID: 37438551.

Goel K, Phillips HW, Chen JS, Ngo J, **Edmonds B**, Ha PX, Wang A, Weil A, Russell BE, Salamon N, Nariai H, Fallah A. Hemispheric Epilepsy Surgery for Hemimegalencephaly: The UCLA Experience. *Epilepsia*. 2023 Oct 24. doi: 10.1111/epi.17807. Epub ahead of print. PMID: 37873610.

Omura JC, Freeman CL, **Perlman SJ**, Fuentes MM. Inpatient rehabilitation admission for a patient with spinal muscular atrophy status post gene therapy. *PM R*. 2023 Oct;15(10):1361-1362. doi: 10.1002/pmrj.12948. Epub 2023 Feb 24. PMID: 36655390.

Glass HC, Numis AL, Comstock BA, Gonzalez FF, Mietzsch U, Bonifacio SL, Massey S, Thomas C, **Natarajan N**, Mayock DE, Sokol GM, Van Meurs KP, Ahmad KA, Maitre N, Heagerty PJ, Juul SE, Wu YW, Wusthoff CJ. Association of EEG Background and Neurodevelopmental Outcome in Neonates With Hypoxic-Ischemic Encephalopathy Receiving Hypothermia. *Neurology*. 2023 Oct 10;101(22):e2223-e2233. doi: 10.1212/WNL.0000000000207744. doi: 10.1212/WNL.0000000000207744. Epub ahead of print. PMID: 37816642.

Amengual-Gual M, Sánchez Fernández I, Vasquez A, Abend NS, Anderson A, Arya R, Barcia Aguilar C, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Lai YC, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Reece L, Riviello JJ, Sannagowdara K, Sheehan T, Tasker RC, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group (pSERG). Pediatric status epilepticus management by Emergency Medical Services (the pSERG cohort). *Seizure*. 2023 Oct;111:51-55. doi: 10.1016/j.seizure.2023.07.010. Epub 2023 Jul 16. PMID: 37523933.

Remore LG, Rifi Z, Nariai H, Eliashiv DS, Fallah A, **Edmonds BD**, Matsumoto JH, Salamon N, Tolossa M, Wei W, Locatelli M, Tsolaki EC, Bari AA. Structural connections of the centromedian nucleus of thalamus and their relevance for neuromodulation in generalized drug-resistant epilepsy: insight from a tractography study. *Ther Adv Neurol Disord*. 2023 Oct 10;16:17562864231202064. doi: 10.1177/17562864231202064. PMID: 37822361; PMCID: PMC10563482.

Gust J. BCMA-CAR T-cell treatment-associated parkinsonism. *Blood*. 2023 Oct 5;142(14):1181-1183. doi: 10.1182/blood.2023021860. PMID: 37796518.

Pediatric Neurology Faculty Publications 2014-2025

Kolnik SE, Marquard R, Brandon O, Puia-Dumitrescu M, Valentine G, Law JB, **Natarajan N**, Dighe M, Mourad PD, Wood TR, Mietzsch U. Preterm infants variability in cerebral near-infrared spectroscopy measurements in the first 72-h after birth. *Pediatr Res*. 2023 Oct;94(4):1408-1415. doi: 10.1038/s41390-023-02618-x. Epub 2023 May 3. PMID: 37138026.

Akiyama LF, **Saneto RP**. Early-Life Epilepsies. *Pediatr Ann*. 2023 Oct;52(10):e381-e387. doi: 10.3928/19382359-20230829-01. Epub 2023 Oct 1. PMID: 37820708.

Mietzsch U, Wood TR, Wu TW, **Natarajan N**, Glass HC, Gonzalez FF, Mayock DE, Comstock BA, Heagerty PJ, Juul SE, Wu YW; HEAL Study Group. Early Glycemic State and Outcomes of Neonates With Hypoxic-Ischemic Encephalopathy. *Pediatrics*. 2023 Oct 1;152(4):e2022060965. doi: 10.1542/peds.2022-060965. PMID: 37655394; PMCID: PMC10522925.

Edmonds B, Miyakoshi M, Gianmaria Remore L, Ahn S, Westley Phillips H, Daida A, Salamon N, Bari A, Sankar R, Matsumoto JH, Fallah A, Nariai H. Characteristics of ictal thalamic EEG in pediatric-onset neocortical focal epilepsy. *Clin Neurophysiol*. 2023 Oct;154:116-125. doi: 10.1016/j.clinph.2023.07.007. Epub 2023 Aug 6. PMID: 37595481.

Saneto RP, Karaa A. Optimized Nutrition in Mitochondrial Diseases Correlates with Improved Muscle Fatigue, Strength, and Quality of Life: You Are What You Eat, or Are You? *Neurotherapeutics*. 2023 Sep 28. doi: 10.1007/s13311-023-01445-6. Epub ahead of print. PMID: 37770714.

Lenahan A, Mietzsch U, Wood TR, Callahan KP, Weiss EM, Miller DE, German K, **Natarajan N**, Puia-Dumitrescu M, Esposito V, Kolnik S, Law JB. Characteristics, Genetic Testing, and Diagnoses of Infants with Neonatal Encephalopathy Not Due to Hypoxic Ischemic Encephalopathy: A Cohort Study. *J Pediatr*. 2023 Sep;260:113533. doi: 10.1016/j.jpeds.2023.113533. Epub 2023 Jun 1. PMID: 37269901.

Begiri E, Badjatia N, Ercole A, Foreman B, Hu P, Hu X, LaRovere K, Meyfroidt G, Moberg D, Robba C, Rosenthal E, Smielewski P, **Wainwright MS**, Park S; Curing Coma Campaign and its Contributing Members. Common Data Elements for Disorders of Consciousness: Recommendations from the Working Group on Physiology and Big Data. *Neurocrit Care*. 2023 Sep 13. doi: 10.1007/s12028-023-01846-7. Epub ahead of print. PMID: 37704934.

Fong SL, Utidjian L, Kaur M, Abend NS, **Wainwright MS**, Grande KM, Foskett N, Roebing R, Guerriero RM, Jain B, Rao S, Stoltzenberg M, Williams P, Yuen N, Dickinson K, McDonald J, Maltenfort M, Forrest CB. Safety of intravenous lacosamide in hospitalized children and neonates. *Epilepsia*. 2023 Sep;64(9):2297-2309. doi: 10.1111/epi.17676. Epub 2023 Jun 19. PMID: 37287398.

Patrick KE, Kroshus E, Boyle LN, Wang J, Binjolkar M, Ebel BE, Rivara FP. Driving characteristics of young adults prior to and following concussion. *Traffic Inj Prev*. 2023 Sep 20:1-6. doi: 10.1080/15389588.2023.2250493. Epub ahead of print. PMID: 37728546.

Lenahan A, Mietzsch U, Wood TR, Callahan KP, Weiss EM, Miller DE, German K, **Natarajan N**, Puia-Dumitrescu M, Esposito V, Kolnik S, Law JB. Characteristics, Genetic Testing, and Diagnoses of Infants with Neonatal Encephalopathy Not Due to Hypoxic Ischemic Encephalopathy: A Cohort Study. *J Pediatr*. 2023 Sep;260:113533. doi: 10.1016/j.jpeds.2023.113533. Epub 2023 Jun 1. PMID: 37269901.

Mietzsch U, Wood TR, Wu TW, **Natarajan N**, Glass HC, Gonzalez FF, Mayock DE, Comstock BA, Heagerty PJ, Juul SE, Wu YW; HEAL Study Group. Early Glycemic State and Outcomes of Neonates With Hypoxic-Ischemic Encephalopathy. *Pediatrics*. 2023 Sep 1:e2022060965. doi: 10.1542/peds.2022-060965. Epub ahead of print. PMID: 37655394.

Hong SJ, **Wainwright MS**, Abend NS, Appavu BL, Francoeur C, Erklauer JC, Guerguerian AM, Guerriero RM, Williams KP, Lee-Eng J, Loeb J, Lovett ME, Murphy SA, Pardo AC, Pineda JA, Ruzas CM, Topjian AA. A Survey of Pediatric Neurocritical Care Fellowship Training in North America. *Pediatr Neurol*. 2023 Sep;146:1-7. doi: 10.1016/j.pediatrneurol.2023.05.015. Epub 2023 May 30. PMID: 37356227.

Akiyama LF, Richards TR, Imada T, Dager SR, Wroblewski L, Kuhl PK. Age-specific average head template for typically developing 6-month-old infants. *PLoS One*. 2013 Sep 12;8(9):e73821. doi: 10.1371/journal.pone.0073821. PMID: 24069234; PMCID: PMC3772014.

Liu Y, Massenburg BB, **Otten CE**, Osorio MB, Lewis SP, Hottovy J, Tse RW. Selective nerve transfers to restore shoulder abduction and flexion in acute flaccid myelitis: A case report. *Microsurgery*. 2023 Aug 30. doi: 10.1002/micr.31104. Epub ahead of print. PMID: 37646277.

Pediatric Neurology Faculty Publications 2014-2025

Bebin EM, Peters JM, Porter BE, McPherson TO, O'Kelley S, Sahin M, Taub KS, Rajaraman R, **Randle SC**, McClintock WM, Koenig MK, Frost MD, Northrup HA, Werner K, Nolan DA, Wong M, Krefting JL, Biasini F, Peri K, Cutter G, Krueger DA; PREVeNT Study Group. Early Treatment with Vigabatrin Does Not Decrease Focal Seizures or Improve Cognition in Tuberous Sclerosis Complex: The PREVeNT Trial. *Ann Neurol*. 2023 Aug 28. doi: 10.1002/ana.26778. Epub ahead of print. PMID: 37638552

Crotty EE, Wilson AL, Davidson T, Tahiri S, **Gust J**, Griesinger AM, Venkataraman S, Park JR, Mueller S, Rood BR, Hwang EI, Wang LD, Vitanza NA. Cellular Therapy for Children with Central Nervous System Tumors: Mining and Mapping the Correlative Data. *Curr Oncol Rep*. 2023 Aug;25(8):847-855. doi: 10.1007/s11912-023-01423-3. Epub 2023 May 9. PMID: 37160547; PMCID: PMC10326126.

Panchavati S, Daida A, **Edmonds B**, Miyakoshi M, Oana S, Ahn SS, Arnold C, Salamon N, Sankar R, Fallah A, Speier W, Nariai H. Uncovering Spatiotemporal Dynamics of the Corticothalamic Network during Seizures. *medRxiv [Preprint]*. 2023 Aug 22:2023.08.21.23294382. doi: 10.1101/2023.08.21.23294382. PMID: 37662245; PMCID: PMC10473800.

Foreman B, Kapinos G, **Wainwright MS**, Ngwenya LB, O'Phelan KH, LaRovere KL, Kirschen MP, Appavu B, Lazaridis C, Alkhachroum A, Maciel CB, Amorim E, Chang JJ, Gilmore EJ, Rosenthal ES, Park S. Practice Standards for the Use of Multimodality Neuromonitoring: A Delphi Consensus Process. *Crit Care Med*. 2023 Aug 18. doi: 10.1097/CCM.0000000000006016. Epub ahead of print. PMID: 37607072.

Edmonds B, Miyakoshi M, Gianmaria Remore L, Ahn S, Westley Phillips H, Daida A, Salamon N, Bari A, Sankar R, Matsumoto JH, Fallah A, Nariai H. Characteristics of ictal thalamic EEG in pediatric-onset neocortical focal epilepsy. *Clin Neurophysiol*. 2023 Aug 6;154:116-125. doi: 10.1016/j.clinph.2023.07.007. Epub ahead of print. PMID: 37595481.

Rosenthal S, Mazzio E, Wellman Gilbert H, Guistolisi S, Marks J, Jewell J, Straley M, Silveira L, Messer R, **Martin J**, Mistry R, Yonker ME, Press CA. Efficacy of IV Valproic Acid and Oral Valproic Acid Tapers for the Treatment of Pediatric Headaches in the Emergency Department. *Neurol Clin Pract*. 2023 Aug;13(4):e200170. doi: 10.1212/CPJ.000000000000200170. Epub 2023 Jun 6. PMID: 37292258; PMCID: PMC10245683.

Edmonds B, Miyakoshi M, Gianmaria Remore L, Ahn S, Westley Phillips H, Daida A, Salamon N, Bari A, Sankar R, Matsumoto JH, Fallah A, Nariai H. Characteristics of ictal thalamic EEG in pediatric-onset neocortical focal epilepsy. *Clin Neurophysiol*. 2023 Aug 6;154:116-125. doi: 10.1016/j.clinph.2023.07.007. Epub ahead of print. PMID: 37595481.

Peariso K, Arya R, Glauser T, Abend NS, Barcia Aguilar C, Amengual-Gual M, Anderson A, Appavu BL, Brenton JN, Carpenter J, Chapman KE, Clark J, Gaillard WD, Gainza-Lein M, Goldstein J, Goodkin H, Grinspan Z, Guerriero RM, Horn PS, Huh L, Kahoud R, Kelley SA, Kossoff EH, Kapur K, Lai YC, Marquis BO, McDonough T, Mikati MA, **Morgan L**, **Novotny E**, Ostendorf AP, Payne ET, Piantino J, Riviello J, Sands T, Stafstrom CE, Tasker RC, Tchapyjnikov D, Vasquez A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; for Pediatric Status Epilepticus Research Group (pSERG). Early Clinical Variables Associated With Refractory Convulsive Status Epilepticus in Children. *Neurology*. 2023 Aug 1;101(5):e546-e557. doi: 10.1212/WNL.000000000000207472. Epub 2023 Jun 9. PMID: 37295955; PMCID: PMC10401687.

Maher K, Persa L, Barry D, **Lee-Eng J**, Dichek H, Joshi S, **Amlie-Lefond C**. Thrombophilia screening in the routine clinical care of children with arterial ischemic stroke. *Pediatr Blood Cancer*. 2023 Aug;70(8):e30381. doi: 10.1002/pbc.30381. Epub 2023 Apr 28. PMID: 37114761.

Hauser Chatterjee J, Boos MD, Shaw DW, Ellenbogen RG, Duffy EK. Resolution of a Focal Pruritic Rash Following Neurosurgical Decompression in an Adolescent With Chiari I and Syringomyelia. *Pediatr Neurol*. 2023 Aug;145:115-118. doi: 10.1016/j.pediatrneurol.2023.05.009. Epub 2023 May 19. PMID: 37329859.

Xu AL, Suresh KV, Gomez JA, Emans JB, Larson AN, Cahill PJ, Andras LM, White KK, Miller DJ, Murphy JS, Groves ML, Belzberg AJ, Hwang SW, Rosser TL, Staedtke V, Ullrich NJ, **Sato AA**, Blakeley JO, Schorry EK, Gross AM, Redding GJ, Sponseller PD; Pediatric Spine Study Group. Consensus-Based Best Practice Guidelines for the Management of Spinal Deformity and Associated Tumors in Pediatric Neurofibromatosis Type 1: Screening and Surveillance, Surgical Intervention, and Medical Therapy. *J Pediatr Orthop*. 2023 Aug 1;43(7):e531-e537. doi: 10.1097/BPO.0000000000002431. Epub 2023 May 29. PMID: 37253707

Morgan LA, Hrachovec JB, Goodkin HP. Pediatric Status Epilepticus: Treat Early and Avoid Delays. *Paediatr Drugs*. 2023 Jul;25(4):411-424. doi: 10.1007/s40272-023-00570-1. Epub 2023 May 13. PMID: 37178271.

Pediatric Neurology Faculty Publications 2014-2025

Karaa A, Bertini E, Carelli V, Cohen BH, Enns GM, Falk MJ, Goldstein A, Gorman GS, Haas R, Hirano M, Klopstock T, Koenig MK, Kornblum C, Lamperti C, Lehman A, Longo N, Molnar MJ, Parikh S, Phan H, Pitceathly RDS, **Saneto R**, Scaglia F, Servidei S, Tarnopolsky M, Toscano A, Van Hove JLK, Vissing J, Vockley J, Finman JS, Brown DA, Shiffer JA, Mancuso M; MMPOWER-3 Trial Investigators. Efficacy and Safety of Elamipretide in Individuals With Primary Mitochondrial Myopathy: The MMPOWER-3 Randomized Clinical Trial. *Neurology*. 2023 Jul 18;101(3):e238-e252. doi: 10.1212/WNL.0000000000207402. Epub 2023 Jun 2. PMID: 37268435; PMCID: PMC10382259.

Amengual-Gual M, Sánchez Fernández I, Vasquez A, Abend NS, Anderson A, Arya R, Barcia Aguilar C, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Lai YC, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Reece L, Riviello JJ, Sannagowdara K, Sheehan T, Tasker RC, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group (pSERG). Pediatric status epilepticus management by Emergency Medical Services (the pSERG cohort). *Seizure*. 2023 Jul 16;111:51-55. doi: 10.1016/j.seizure.2023.07.010. Epub ahead of print. PMID: 37523933.

Benedetti GM, **Wainwright MS**. Commentary: Characteristics and Outcomes of Children with Cerebral Sinus Venous Thrombosis. *Neurocrit Care*. 2023 Jul 12. doi: 10.1007/s12028-023-01787-1. Epub ahead of print. PMID: 37438551.

Ch'en PY, Gold LS, Lu Q, Ye T, Andrews JS, **Patel P**. Exploring risk factors for persistent neurocognitive sequelae after hospitalization for COVID-19. *Ann Clin Transl Neurol*. 2023 Jul;10(7):1200-1208. doi: 10.1002/acn3.51801. Epub 2023 Jun 2. PMID: 37265172; PMCID: PMC10351655.

Glass HC, Wusthoff CJ, Comstock BA, Numis AL, Gonzalez FF, Maitre N, Massey SL, Mayock DE, Mietzsch U, **Natarajan N**, Sokol GM, Bonifacio SL, Van Meurs KP, Thomas C, Ahmad KA, Heagerty PJ, Juul SE, Wu YW. Risk of seizures in neonates with hypoxic-ischemic encephalopathy receiving hypothermia plus erythropoietin or placebo. *Pediatr Res*. 2023 Jul;94(1):252-259. doi: 10.1038/s41390-022-02398-w. Epub 2022 Dec 5. PMID: 36470964; PMCID: PMC10239788.

Peariso K, Arya R, Glauser T, Abend NS, Aguilar CB, Amengual-Gual M, Anderson A, Appavu BL, Brenton JN, Carpenter J, Chapman KE, Clark J, Gaillard WD, Gaínza-Lein M, Goldstein J, Goodkin H, Grinspan Z, Guerriero RM, Horn PS, Huh L, Kahoud R, Kelley SA, Kossoff EH, Kapur K, Lai YC, Marquis BO, McDonough T, Mikati MA, **Morgan L**, **Novotny E**, Ostendorf AP, Payne ET, Piantino J, Riviello J, Sands T, Stafstrom CE, Tasker RC, Tchapyjnikov D, Vasquez-Avila A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; for Pediatric Status Epilepticus Research Group (pSERG). Early Clinical Variables Associated With Refractory Convulsive Status Epilepticus in Children. *Neurology*. 2023 Jun 9;101(11):e207472. doi: 10.1212/WNL.0000000000207472. Epub ahead of print. PMID: 37295955

Mazzio E, Mistry RD, Rosenthal S, Ser E, Jewell J, **Martin J**, Messer R, Straley M, Leonard J, Press CA. Neuroimaging for Pediatric Non-First-Time Seizures in the Emergency Department. *Neurol Clin Pract*. 2022 Jun;12(3):e7-e13. doi: 10.1212/CPJ.0000000000001165. PMID: 35747538; PMCID: PMC9208414.

Gray JM, Kramer ME, Suskauer SJ, Slomine BS. Functional Recovery During Inpatient Rehabilitation in Children With Anoxic or Hypoxic Brain Injury. *Arch Phys Med Rehabil*. 2023 Jun;104(6):918-924. doi: 10.1016/j.apmr.2023.01.018. Epub 2023 Feb 8. PMID: 36758714.

Fong SL, Utidjian L, Kaur M, Abend NS, **Wainwright MS**, Grande KM, Foscett N, Roebling R, Guerriero RM, Jain B, Rao S, Stoltenberg M, Williams P, Yuen N, Dickinson K, McDonald J, Maltenfort M, Forrest CB. Safety of intravenous lacosamide in hospitalized children and neonates. *Epilepsia*. 2023 Jun 7. doi: 10.1111/epi.17676. Epub ahead of print. PMID: 37287398.

Berl MM, Koop JI, Ailion A, Bearden DJ, Boyer K, Cooper CM, Decrow AM, Duong PH, Espe-Pfeifer P, Gabriel M, Hodges E, Marshall DF, McNally KA, Molnar AE, Olsen EK, Ono KE, **Patrick KE**, Paul BM, Romain J, Sepeta LN, Stilp RLH, Wilkening GN, Zaccariello M, Zelko F, Perry MS; PERC Surgery Workgroup (includes Edward Novotny). Leveraging expertise and optimizing clinical research: Initial success of a pediatric epilepsy surgery collaborative. *Epilepsia*. 2023 Jun;64(6):1554-1567. doi: 10.1111/epi.17579. Epub 2023 Mar 24. PMID: 36897767.

Akiyama LF, Roberts EA, Shurtleff HA, Barry D, **Saneto RP**, **Novotny EJ**, Young CC, Warner MH, Hauptman JS, Ojemann JG, Marshally A. Clinical outcomes of pediatric hemispherectomy following unsuccessful subhemispheric resection for refractory epilepsy: a case review study. *J Neurosurg Pediatr*. 2023 Jun 2:1-8. doi: 10.3171/2023.4.PEDS2328. Epub ahead of print. PMID: 37310054.

Pediatric Neurology Faculty Publications 2014-2025

- Karaa A, Bertini E, Carelli V, Cohen BH, Enns GM, Falk MJ, Goldstein A, Gorman GS, Haas R, Hirano M, Klopstock T, Koenig MK, Kornblum C, Lamperti C, Lehman A, Longo N, Molnar MJ, Parikh S, Phan H, Pitceathly RDS, **Saneto R**, Scaglia F, Servidei S, Tarnopolsky M, Toscano A, Van Hove JLK, Vissing J, Vockley J, Finman JS, Brown DA, Shiffer JA, Mancuso M; MMPOWER-3 Trial Investigators. Efficacy and Safety of Elamipretide in Individuals With Primary Mitochondrial Myopathy: The MMPOWER-3 Randomized Clinical Trial. *Neurology*. 2023 Jun 2;101(12):e207402. doi: 10.1212/WNL.000000000000207402. Epub ahead of print. PMID: 37268435.
- Lenahan A, Mietzsch U, Wood TR, Callahan KP, Weiss EM, Miller DE, German K, **Natarajan N**, Puia-Dumitrescu M, Esposito V, Kolnik S, Law JB. Characteristics, Genetic Testing, and Diagnoses of Infants with Neonatal Encephalopathy Not Due to Hypoxic Ischemic Encephalopathy: A Cohort Study. *J Pediatr*. 2023 Jun 1;260:113533. doi: 10.1016/j.jpeds.2023.113533. Epub ahead of print. PMID: 37269901.
- Rivara FP, Ebel BE, Binjolkar M, Wang J, Hanron A, Kroshus E, Boyle LN, **Patrick KE**. Cognitive Impairment and Driving Skills in Youth After Concussion. *J Neurotrauma*. 2023 Jun;40(11-12):1187-1196. doi: 10.1089/neu.2022.0308. Epub 2022 Dec 26. PMID: 36416238; PMCID: PMC10259608.
- Berl MM, Koop JI, Ailion A, Bearden DJ, Boyer K, Cooper CM, Decrow AM, Duong PH, Espe-Pfeifer P, Gabriel M, Hodges E, Marshall DF, McNally KA, Molnar AE, Olsen EK, Ono KE, **Patrick KE**, Paul BM, Romain J, Sepeta LN, Stilp RLH, Wilkening GN, Zaccariello M, Zelko F, Perry MS; PERC Surgery Workgroup. Leveraging expertise and optimizing clinical research: Initial success of a pediatric epilepsy surgery collaborative. *Epilepsia*. 2023 Jun;64(6):1554-1567. doi: 10.1111/epi.17579. Epub 2023 Mar 24. PMID: 36897767
- McCormick EM, Keller K, Taylor JP, Coffey AJ, Shen L, Krotoski D, Harding B; NICHD ClinGen U24 Mitochondrial Disease Gene Curation Expert Panel; Alves CAPF, Ardisson A, Bai R, de Barcelos IP, Bertini E, Bluske K, Christodoulou J, Clause AR, Copeland WC, Diaz GA, Diodato D, Dulik MC, Enns G, Feigenbaum A, Fratter C, Ghezzi D, Goldstein A, Gropman A, Haas R, Karaa A, Koenig MK, Monteleone B, Parikh S, Duenas BP, Rajkumar R, Saada A, **Saneto RP**, Sergeant K, Shoffner J, Smith C, Stanley C, Thiffault I, Thorburn D, Walker M, Wallace D, Wong LJ, Gai X, Falk MJ, Zolkipli-Cunningham Z, Rahman S. Expert panel curation of 113 primary mitochondrial disease genes for the Leigh syndrome spectrum. *Ann Neurol*. 2023 May 31. doi: 10.1002/ana.26716. Epub ahead of print. PMID: 37255483.
- Hong SJ, **Wainwright MS**, Abend NS, Appavu BL, Francoeur C, Erklauer JC, Guerguerian AM, Guerriero RM, Williams KP, **Lee-Eng J**, Loeb J, Lovett ME, Murphy SA, Pardo AC, Pineda JA, Ruzas CM, Topjian AA. A Survey of Pediatric Neurocritical Care Fellowship Training in North America. *Pediatr Neurol*. 2023 May 30;146:1-7. doi: 10.1016/j.pediatrneurol.2023.05.015. Epub ahead of print. PMID: 37356227.
- 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.000000000000200130. PMID: 37236807; PMCID: PMC10219134
- Boop S, Bonda D, **Randle S**, Leary S, Vitanza N, Crotty E, **Novotny E**, Friedman S, Ellenbogen RG, Durfy S, Goldstein H, Ojemann JG, Hauptman JS. A comparison of clinical outcomes for subependymal giant cell astrocytomas treated with laser interstitial thermal therapy, open surgical resection, and mTOR inhibitors. *Pediatr Neurosurg*. 2023 May 24. doi: 10.1159/000531210. Epub ahead of print. PMID: 37232001.
- Santomasso BD, **Gust J**, Perna F. How I treat unique and difficult-to-manage cases of CAR T-cell therapy-associated neurotoxicity. *Blood*. 2023 May 18;141(20):2443-2451. doi: 10.1182/blood.2022017604. PMID: 36877916; PMCID: PMC10329188.
- Kolnik SE, Marquard R, Brandon O, Puia-Dumitrescu M, Valentine G, Law JB, **Natarajan N**, Dighe M, Mourad PD, Wood TR, Mietzsch U. Preterm infants variability in cerebral near-infrared spectroscopy measurements in the first 72-h after birth. *Pediatr Res*. 2023 May 3. doi: 10.1038/s41390-023-02618-x. Epub ahead of print. PMID: 37138026.
- Hauser Chatterjee J**, Hartford EA, Law E, Barry D, **Blume H**. Sumatriptan as a First-Line Treatment for Headache in the Pediatric Emergency Department. *Pediatr Neurol*. 2023 May;142:68-75. doi: 10.1016/j.pediatrneurol.2023.01.016. Epub 2023 Feb 4. PMID: 36958085.
- Martin JA**, Sampey F, Feldman A, Silveira L, Press CA, Messer R, Barry M, Kaul P. Development and Evaluation of a Child Neurology Resident Curriculum for Communication Around Serious Illness. *J Child Neurol*. 2023 Apr;38(5):307-314. doi: 10.1177/08830738231172539. Epub 2023 May 8. PMID: 37157829

Pediatric Neurology Faculty Publications 2014-2025

Benedetti GM, **Morgan LA**, Sansevere AJ, Harrar DB, Guerriero RM, **Wainwright MS**, LaRovere KL, Kielian A, Ganesan SL, Press CA; Pediatric Quantitative EEG Strategic Taskforce (PedQuEST). The Spectrum of Quantitative EEG Utilization Across North America: A Cross-Sectional Survey. *Pediatr Neurol.* 2023 Apr;141:1-8. doi: 10.1016/j.pediatrneurol.2022.12.016. Epub 2023 Jan 6. PMID: 36731228.

Mahdi J, Dietrich J, Straathof K, Roddie C, Scott BJ, Davidson TB, Prolo LM, Batchelor TT, Campen CJ, Davis KL, **Gust J**, Lim M, Majzner RG, Park JR, Partap S, Ramakrishna S, Richards R, Schultz L, Vitanza NA, Wang LD, Mackall CL, Monje M. Tumor inflammation-associated neurotoxicity. *Nat Med.* 2023 Apr;29(4):803-810. doi: 10.1038/s41591-023-02276-w. Epub 2023 Apr 6. PMID: 37024595; PMCID: PMC10166099.

Bozarth XL, Lopez J, Fang H, **Lee-Eng J**, Duan Z, Deng X. Phenotypes and Genotypes in Patients with *SMC1A*-Related Developmental and Epileptic Encephalopathy. *Genes (Basel).* 2023 Mar 31;14(4):852. doi: 10.3390/genes14040852. PMID: 37107610; PMCID: PMC10138066.

Shelkowitz E, **Saneto RP**, Al-Hertani W, Lubout CMA, Stence NV, Brown MS, Long P, Walleigh D, Nelson JA, Perez FE, Shaw DWW, Michl EJ, Van Hove JLK. Correction: Ketogenic diet as a glycine lowering therapy in nonketotic hyperglycinemia and impact on brain glycine levels. *Orphanet J Rare Dis.* 2023 Mar 13;18(1):54. doi: 10.1186/s13023-023-02646-0. Erratum for: *Orphanet J Rare Dis.* 2022 Dec 5;17(1):423. PMID: 36915141; PMCID: PMC10012511.

Patel PB, **Patrick KE**, Benedetti GM, **Morgan LA**, **Bowen KS**, Wright JN, **Wainwright MS**. A Multidisciplinary Pediatric Neurology Clinic for Systematic Follow-up of Children with Neurologic Sequelae of COVID-19. *J Child Neurol.* 2023 Mar;38(3-4):121-129. doi: 10.1177/08830738231156950. Epub 2023 Mar 29. PMID: 36991568; PMCID: PMC10064197.

Qin ES, Gold LS, Singh N, Wysham KD, Hough CL, **Patel PB**, Bunnell AE, Andrews JS. Physical function and fatigue recovery at 6 months after hospitalization for COVID-19. *PM R.* 2023 Mar;15(3):314-324. doi: 10.1002/pmjr.12866. Epub 2022 Aug 6. PMID: 35726518; PMCID: PMC9353339.

Chang V, **Mingbunjerdsuk D**. Dystonia Responding to Levodopa in A Child with Myoclonus-Dystonia. *Mov Disord Clin Pract.* 2023 Mar 7;10(4):687-690. doi: 10.1002/mdc3.13708. PMID: 37070063; PMCID: PMC10105106.

Paul MS, Duncan AR, Genetti CA, Pan H, Jackson A, Grant PE, Shi J, Pinelli M, Brunetti-Pierri N, Garza-Flores A, Shahani D, **Saneto RP**, Zampino G, Leoni C, Agolini E, Novelli A, Blümlein Tobias B Haack U, Heinritz W, Matzker E, Alhaddad B, Jamra RA, Bartolomaeus T, AlHamdan S, Carapito R, Isidor B, Bahram S, Ritter A, Izumi K, Shakked BP, Barel O, Ben Zeev B, Begtrup A, Carere DA, Mullegama SV, Palculict TB, Calame DG, Schwan K, Aycinena ARP, Traberg R; Genomics England Research Consortium; Douzgou S, Pirt H, Ismayilova N, Banka S, Chao HT, Agrawal PB. Rare EIF4A2 variants are associated with a neurodevelopmental disorder characterized by intellectual disability, hypotonia, and epilepsy. *Am J Hum Genet.* 2023 Mar 2;110(3):548. doi: 10.1016/j.ajhg.2023.02.010. Erratum for: *Am J Hum Genet.* 2023 Jan 5;110(1):120-145. PMID: 36868207; PMCID: PMC10027494.

Wade SL, Walsh K, Slomine BS, Davis KC, Heard C, Maggard B, Sutcliffe M, Van Tubbergen M, McNally K, Deidrick K, Kirkwood MW, Lantagne A, **Ashman S**, Scratch S, Chesley G, Johnson-Kerner B, Johnson A, Cirincione L, Austin C. Widespread clinical implementation of the teen online problem-solving program: Progress, barriers, and lessons learned. *Front Rehabil Sci.* 2023 Feb 7;3:1089079. doi: 10.3389/fresc.2022.1089079. PMID: 36824180; PMCID: PMC9942775.

Ostendorf AP, Axteen ET, Eschbach K, Fedak Romanowski E, **Morgan LA**, Gross P, Narayanan UG, Glader L, Noritz G; Cerebral Palsy Research Network. Epilepsy and proxy-reported health-related quality of life in children and young people with non-ambulatory cerebral palsy. *Dev Med Child Neurol.* 2023 Feb;65(2):200-206. doi: 10.1111/dmcn.15336. Epub 2022 Jul 12. PMID: 35820144; PMCID: PMC10084160.

Castro D, **Henriquez A**. Atypical presentation of hypokalemic periodic paralysis: A case report. *Muscle Nerve.* 2023 Apr;67(4):E8-E9. doi: 10.1002/mus.27771. Epub 2023 Jan 27. PMID: 36507920.

Amlie-Lefond C. Pediatric Stroke-Are We Asking the Right Questions? The 2022 Sidney Carter Award Lecture. *Neurology.* 2023 Jan 24;100(4):192-198. doi: 10.1212/WNL.0000000000201487. Epub 2022 Nov 8. PMID: 36347625.

Omura JC, Freeman CL, **Perlman SJ**, Fuentes MM. Inpatient rehabilitation admission for a patient with spinal muscular atrophy status post gene therapy. *PM R.* 2023 Jan 19. doi: 10.1002/pmjr.12948. Epub ahead of print. PMID: 36655390.

Pediatric Neurology Faculty Publications 2014-2025

Vitanza NA, Wilson AL, Huang W, Seidel K, Brown C, Gustafson JA, Yokoyama JK, Johnson AJ, Baxter BA, Koning RW, Reid AN, Meechan M, Biery MC, Myers C, Rawlings-Rhea SD, Albert CM, Browd SR, Hauptman JS, Lee A, Ojemann JG, Berens ME, Dun MD, Foster JB, Crotty EE, Leary SES, Cole BL, Perez FA, Wright JN, Orentas RJ, Chour T, Newell EW, Whiteaker JR, Zhao L, Paulovich AG, Pinto N, **Gust J**, Gardner RA, Jensen MC, Park JR. Intraventricular B7-H3 CAR T Cells for Diffuse Intrinsic Pontine Glioma: Preliminary First-in-Human Bioactivity and Safety. *Cancer Discov.* 2023 Jan 9;13(1):114-131. doi: 10.1158/2159-8290.CD-22-0750. PMID: 36259971; PMCID: PMC9827115.

Paul MS, Duncan AR, Genetti CA, Pan H, Jackson A, Grant PE, Shi J, Pinelli M, Brunetti-Pierri N, Garza-Flores A, Shahani D, **Saneto RP**, Zampino G, Leoni C, Agolini E, Novelli A, Blümlein U, Haack TB, Heinritz W, Matzker E, Alhaddad B, Abou Jamra R, Bartolomaeus T, AlHamdan S, Carapito R, Isidor B, Bahram S, Ritter A, Izumi K, Shakked BP, Barel O, Ben Zeev B, Begtrup A, Carere DA, Mullegama SV, Palculict TB, Calame DG, Schwan K, Aycinena ARP, Traberg R; Genomics England Research Consortium; Douzgou S, Pirt H, Ismayilova N, Banka S, Chao HT, Agrawal PB. Rare EIF4A2 variants are associated with a neurodevelopmental disorder characterized by intellectual disability, hypotonia, and epilepsy. *Am J Hum Genet.* 2023 Jan 5;110(1):120-145. doi: 10.1016/j.ajhg.2022.11.011. Epub 2022 Dec 16. Erratum in: *Am J Hum Genet.* 2023 Mar 2;110(3):548. PMID: 36528028; PMCID: PMC9892767.

2022

Rivara FP, Ebel BE, Binjolkar M, Wang J, Hanron A, Kroshus E, Boyle LN, **Patrick KE**. Cognitive Impairment and Driving Skills in Youth after Concussion. *J Neurotrauma.* 2022 Dec 26. doi: 10.1089/neu.2022.0308. Epub ahead of print. PMID: 36416238

Shelkowitz E, **Saneto RP**, Al-Hertani W, Lubout CMA, Stence NV, Brown MS, Long P, Walleigh D, Nelson JA, Perez FE, Shaw DWW, Michl EJ, Van Hove JLK. Ketogenic diet as a glycine lowering therapy in nonketotic hyperglycinemia and impact on brain glycine levels. *Orphanet J Rare Dis.* 2022 Dec 5;17(1):423. doi: 10.1186/s13023-022-02581-6. PMID: 36471344; PMCID: PMC9720968.

Glass HC, Wusthoff CJ, Comstock BA, Numis AL, Gonzalez FF, Maitre N, Massey SL, Mayock DE, Mietzsch U, **Natarajan N**, Sokol GM, Bonifacio SL, Van Meurs KP, Thomas C, Ahmad KA, Heagerty PJ, Juul SE, Wu YW. Risk of seizures in neonates with hypoxic-ischemic encephalopathy receiving hypothermia plus erythropoietin or placebo. *Pediatr Res.* 2022 Dec 5. doi: 10.1038/s41390-022-02398-w. Epub ahead of print. PMID: 36470964.

Serrallach BL, Schafer ES, Kralik SK, Tran BH, Huisman TAGM, Wright JN, **Morgan LA**, Desai NK. Imaging Findings in Children Presenting with CNS Nelarabine Toxicity. *AJNR Am J Neuroradiol.* 2022 Dec;43(12):1802-1809. doi: 10.3174/ajnr.A7692. Epub 2022 Nov 3. PMID: 36328408.

Castro D, **Henriquez A**. Atypical presentation of hypokalemic periodic paralysis: A case report. *Muscle Nerve.* 2022 Dec 12. doi: 10.1002/mus.27771. Epub ahead of print. PMID: 36507920.

Chatterjee JH, Blume HK. Migraine and Mental Health in Pediatrics. *JAMA Pediatr.* 2022 Dec 1;176(12):1165-1166. doi: 10.1001/jamapediatrics.2022.3948. PMID: 36315116.

Amlie-Lefond C. Pediatric Stroke-Are We Asking the Right Questions? The 2022 Sidney Carter Award Lecture. *Neurology.* 2022 Nov 8;10.1212/WNL.0000000000201487. doi: 10.1212/WNL.0000000000201487. Epub ahead of print. PMID: 36347625.

Benedetti GM, **Amlie-Lefond C**. Caring for Our Sickest Patients: The Expanding Role of the Pediatric Neurohospitalist. *Neurology.* 2022 Nov 1;99(18):781-782. doi: 10.1212/WNL.0000000000201267. PMID: 36316123.

Chatterjee JH, Blume HK. Migraine and Mental Health in Pediatrics. *JAMA Pediatr.* 2022 Oct 31. doi: 10.1001/jamapediatrics.2022.3948. Epub ahead of print. PMID: 36315116.

Vitanza NA, Wilson AL, Huang W, Seidel K, Brown C, Gustafson JA, Yokoyama JK, Johnson AJ, Baxter BA, Koning RW, Reid AN, Meechan M, Biery MC, Myers C, Rawlings-Rhea SD, Albert CM, Browd SR, Hauptman JS, Lee A, Ojemann JG, Berens ME, Dun MD, Foster JB, Crotty EE, Leary SES, Cole BL, Perez FA, Wright JN, Orentas RJ, Chour T, Newell EW, Whiteaker JR, Zhao L, Paulovich AG, Pinto N, **Gust J**, Gardner RA, Jensen MC, Park JR. Intraventricular B7-H3 CAR T cells for diffuse intrinsic pontine glioma: preliminary first-in-human bioactivity and safety. *Cancer Discov.* 2022 Oct 19;CD-22-0750. doi: 10.1158/2159-8290.CD-22-0750. Epub ahead of print. PMID: 36259971.

Pediatric Neurology Faculty Publications 2014-2025

Guglieri M, Clemens PR, **Perlman SJ**, Smith EC, Horrocks I, Finkel RS, Mah JK, Deconinck N, Goemans N, Haberlova J, Straub V, Mengle-Gaw LJ, Schwartz BD, Harper AD, Shieh PB, De Waele L, Castro D, Yang ML, Ryan MM, McDonald CM, Tulinius M, Webster R, McMillan HJ, Kuntz NL, Rao VK, Baranello G, Spinty S, Childs AM, Sbrocchi AM, Selby KA, Monduy M, Nevo Y, Vilchez-Padilla JJ, Nascimento-Osorio A, Niks EH, de Groot IJM, Katsalouli M, James MK, van den Anker J, Damsker JM, Ahmet A, Ward LM, Jaros M, Shale P, Dang UJ, Hoffman EP. Efficacy and Safety of Vamorolone vs Placebo and Prednisone Among Boys With Duchenne Muscular Dystrophy: A Randomized Clinical Trial. *JAMA Neurol*. 2022 Oct 1;79(10):1005-1014. doi: 10.1001/jamaneurol.2022.2480. PMID: 36036925; PMCID: PMC9425287

Mayne EW, Mailo JA, Pabst L, Pulcine E, Harrar DB, Waak M, Rafay MF, Hassanein SM, **Amlie-Lefond C**, Jordan LC. Pediatric Stroke and Cardiac Disease: Challenges in Recognition and Management. *Semin Pediatr Neurol*. 2022 Oct;43:100992. doi: 10.1016/j.spen.2022.100992. Epub 2022 Aug 31. PMID: 36344023.

Surtees TL, Pearson R, Harrar DB, Lee S, **Amlie-Lefond CM**, Guillems KP. Acute Hospital Management of Pediatric Stroke. *Semin Pediatr Neurol*. 2022 Oct;43:100990. doi: 10.1016/j.spen.2022.100990. Epub 2022 Aug 19. PMID: 36344020.

Patel ND, Batra M, Udomphorn Y, **Wainwright M**, Vavilala MS. Cerebral Autoregulation in Healthy Term Newborns: Brief Report. *Pediatr Neurol*. 2022 Oct;135:4-5. doi: 10.1016/j.pediatrneurol.2022.07.001. Epub 2022 Jul

Persa L, Shaw DW, **Amlie-Lefond C**. Why Would a Child Have a Stroke? *J Child Neurol*. 2022 Oct 9:8830738221129916. doi: 10.1177/08830738221129916. Epub ahead of print. PMID: 36214173.

Hartford EA, **Blume H**, Barry D, **Hauser Chatterjee J**, Law E. Disparities in the emergency department management of pediatric migraine by race, ethnicity, and language preference. *Acad Emerg Med*. 2022 Sep;29(9):1057-1066. doi: 10.1111/acem.14550. Epub 2022 Jul 7. PMID: 35726699.

Olson V, Chang IJ, Merritt Nd JL, **Mingbunjerdasuk D**. Refractory Myoclonus as a Presentation of Metabolic Stroke in A Child With Cobalamin B Methylmalonic Acidemia After Liver and Kidney Transplant. *J Mov Disord*. 2022 Sep;15(3):281-283. doi: 10.14802/jmd.21196. Epub 2022 May 26. PMID: 35614015; PMCID: PMC9536905.

Natarajan N, Benedetti G, Perez FA, Wood TR, German KR, **Lockrow JP**, Puia-Dumitrescu M, Myers E, Mietzsch U. Association Between Early EEG Background and Outcomes in Infants With Mild HIE Undergoing Therapeutic Hypothermia. *Pediatr Neurol*. 2022 Sep;134:52-58. doi: 10.1016/j.pediatrneurol.2022.06.006. Epub 2022 Jun 19. PMID: 35835026.

Hanaford A, **Johnson SC**. The immune system as a driver of mitochondrial disease pathogenesis: a review of evidence. *Orphanet J Rare Dis*. 2022 Sep 2;17(1):335. doi: 10.1186/s13023-022-02495-3. PMID: 36056365; PMCID: PMC9438277.

Gust J, Rawlings-Rhea SD, Wilson AL, Tulberg NM, Sherman AL, Seidel KD, Wu QV, Park JR, Gardner RA, Annesley CE. GFAP and NFL increase during neurotoxicity from high baseline levels in pediatric CD19-CAR T-cell patients. *Blood Adv*. 2022 Aug 25:bloodadvances.2022008119. doi: 10.1182/bloodadvances.2022008119. Epub ahead of print. PMID: 36006611.

Moufawad El Achkar C, Rosen A, Kessler SK, **Steinman KJ**, Spence SJ, Ramocki M, Marco EJ, Green Snyder L, Spiro JE, Chung WK, Annapurna P, Sherr EH. Clinical Characteristics of Seizures and Epilepsy in Individuals With Recurrent Deletions and Duplications in the 16p11.2 Region. *Neurol Genet*. 2022 Aug 5;8(5):e200018. doi: 10.1212/NXG.000000000200018. PMID: 36531974; PMCID: PMC9756306.

Tran DK, Poliakov AV, Friedman SD, Goldstein HE, Shurtleff HA, **Bowen K**, **Patrick KE**, Warner M, **Novotny EJ**, Ojemann JG, Hauptman JS. Concordance of functional MRI memory task and resting-state functional MRI connectivity used in surgical planning for pediatric temporal lobe epilepsy. *J Neurosurg Pediatr*. 2022 Jul 29:1-6. doi: 10.3171/2022.6.PEDS221. Epub ahead of print. PMID: 35907201.

Erklauer JC, Thomas AX, Hong SJ, Appavu BL, Carpenter JL, Chiriboga-Salazar NR, Ferrazzano PA, Goldstein Z, Griffith JL, Guillems KP, Kirschen MP, Lidsky K, Lovett ME, McLaughlin B, Munoz Pareja JC, Murphy S, O'Donnell W, Riviello JJ, Schober ME, Topjian AA, **Wainwright MS**, Simon DW, Pediatric Neurocritical Care Research Group. A Virtual Community of Practice: An International Educational Series in Pediatric Neurocritical Care. *Children (Basel)*. 2022 Jul 20;9(7):1086. doi: 10.3390/children9071086. PMID: 35884070; PMCID: PMC9316633.

Pediatric Neurology Faculty Publications 2014-2025

- Tarazi RA, **Patrick KE**, Iampietro M, Apollonsky N. Hydroxyurea Use Associated with Nonverbal and Executive Skills in Sick Cell Anemia. *J Pediatr Psychol*. 2021 Jul 20;46(6):710-718. doi: 10.1093/jpepsy/jsab015. PMID: 33706380.
- Ostendorf AP, Axeen ET, Eschbach K, Fedak Romanowski E, **Morgan LA**, Gross P, Narayanan UG, Glader L, Noritz G; CEREBRAL PALSY RESEARCH NETWORK. Epilepsy and proxy-reported health-related quality of life in children and young people with non-ambulatory cerebral palsy. *Dev Med Child Neurol*. 2022 Jul 12. doi: 10.1111/dmcn.15336. Epub ahead of print. PMID: 35820144.
- Faulhaber LD, D'Costa O, Shih AY, **Gust J**. Antibody-based *in vivo* leukocyte label for two-photon brain imaging in mice. *Neurophotonics*. 2022 Jul;9(3):031917. doi: 10.1117/1.NPh.9.3.031917. Epub 2022 May 24. PMID: 35637871; PMCID: PMC9128835.
- Davies OMT, Garzon MC, Frieden IJ, Cottrell CE, Gripp KW, **Saneto RP**, Shwayder T, Mirzaa GM, Drolet BA. Cutaneous vascular anomalies associated with a mosaic variant of AKT3: Genetic analysis continues to refine the diagnosis, nomenclature, and classification of vascular anomalies. *J Am Acad Dermatol*. 2022 Jul;87(1):162-164. doi: 10.1016/j.jaad.2021.06.877. Epub 2021 Jul 6. PMID: 34237354; PMCID: PMC8733055.
- Goldstein HE, Poliakov A, Shaw DW, Barry D, Tran K, **Novotny EJ**, **Saneto RP**, Marashly A, Warner MH, Wright JN, Hauptman JS, Ojemann JG, Shurtleff HA. Precision medicine in pediatric temporal epilepsy surgery: optimization of outcomes through functional MRI memory tasks and tailored surgeries. *J Neurosurg Pediatr*. 2022 Jul 1:1-12. doi: 10.3171/2022.5.PEDS22148. Epub ahead of print. PMID: 35901731.
- Striano P, Auvin S, Collins A, Horvath R, Scheffer IE, Tzadok M, Miller I, Kay Koenig M, Lacy A, Davis R, Garcia-Cazorla A, **Saneto RP**, Brandabur M, Blair S, Koutsoukos T, De Vivo D. A randomized, double-blind trial of triheptanoin for drug-resistant epilepsy in glucose transporter 1 deficiency syndrome. *Epilepsia*. 2022 Jul;63(7):1748-1760. doi: 10.1111/epi.17263. Epub 2022 May 21. PMID: 35441706.
- Hartford EA, **H**, Barry D, **Hauser Chatterjee J**, Law E. Disparities in the emergency department management of pediatric migraine by race, ethnicity, and language preference. *Acad Emerg Med*. 2022 Jun 21. doi: 10.1111/acem.14550. Epub ahead of print. PMID: 35726699.
- Qin ES, Gold LS, Singh N, Wysham KD, Hough CL, **Patel PB**, Bunnell AE, Andrews JS. Physical function and fatigue recovery at 6 months after hospitalization for COVID-19. *PM R*. 2022 Jun 21;10.1002/pmrj.12866. doi: 10.1002/pmrj.12866. Epub ahead of print. PMID: 35726518; PMCID: PMC9353339.
- Natarajan N**, **Benedetti G**, Perez FA, Wood TR, German KR, **Lockrow JP**, Puia-Dumitrescu M, Myers E, Mietzsch U. Association Between Early EEG Background and Outcomes in Infants With Mild HIE Undergoing Therapeutic Hypothermia. *Pediatr Neurol*. 2022 Jun 19;134:52-58. doi: 10.1016/j.pediatrneurol.2022.06.006. Epub ahead of print. PMID: 35835026
- Saneto RP**, Perez FA. Mitochondria-Associated Membrane Scaffolding with Endoplasmic Reticulum: A Dynamic Pathway of Developmental Disease. *Front Mol Biosci*. 2022 Jun 14;9:908721. doi: 10.3389/fmolb.2022.908721. PMID: 35775081; PMCID: PMC9237565.
- Emmanuele V, Ganesh J, Vladutiu G, Haas R, Kerr D, **Saneto RP**, Cohen BH, Van Hove JLK, Scaglia F, Hoppel C, Rosales XQ, Barca E, Buchsbaum R, Thompson JL, DiMauro S, Hirano M; North American Mitochondrial Disease Consortium (NAMDC). Time to harmonize mitochondrial syndrome nomenclature and classification: A consensus from the North American Mitochondrial Disease Consortium (NAMDC). *Mol Genet Metab*. 2022 Jun;136(2):125-131. doi: 10.1016/j.ymgme.2022.05.001. Epub 2022 May 13. PMID: 35606253; PMCID: PMC9341219.
- Castillo-Pinto C**, Carpenter JL, Donofrio MT, Zhang A, Wernovsky G, Sinha P, Harrar D. Incidence and predictors of epilepsy in children with congenital heart disease. *Cardiol Young*. 2022 Jun;32(6):918-924. doi: 10.1017/S1047951121003279. Epub 2021 Aug 9. PMID: 34365987.
- Crotty E, **Gust J**. Pediatric ependymoma: New perspectives on older trials. *Neuro Oncol*. 2022 Jun 1;24(6):949-950. doi: 10.1093/neuonc/noac060. PMID: 35325202; PMCID: PMC9159413.
- Knight EMP, Amin S, Bahi-Buisson N, Benke TA, Cross JH, Demarest ST, Olson HE, Specchio N, Fleming TR, Aimetti AA, Gasior M, Devinsky O; Marigold Trial Group (**Saneto, RP**). Safety and efficacy of ganaxolone in patients with CDKL5 deficiency disorder: results from the double-blind phase of a randomised, placebo-controlled, phase 3 trial. *Lancet Neurol*.

Pediatric Neurology Faculty Publications 2014-2025

2022 May;21(5):417-427. doi: 10.1016/S1474-4422(22)00077-1. Erratum in: Lancet Neurol. 2022 Jul;21(7):e7. PMID: 35429480.

Olson V, Chang IJ, Merritt Nd JL, **Mingbunjerdasuk D**. Refractory Myoclonus as a Presentation of Metabolic Stroke in A Child With Cobalamin B Methylmalonic Acidemia After Liver and Kidney Transplant. J Mov Disord. 2022 May 26. doi: 10.14802/jmd.21196. Epub ahead of print. PMID: 35614015.

Chen WL, Pao E, **Owens J**, Glass I, Pritchard C, Shirts BH, Lockwood C, Mirzaa GM. The utility of cerebrospinal fluid-derived cell-free DNA in molecular diagnostics for the *PIK3CA*-related megalencephaly-capillary malformation (MCAP) syndrome: a case report. Cold Spring Harb Mol Case Stud. 2022 Apr 28;8(3):a006188. doi: 10.1101/mcs.a006188. PMID: 35483878; PMCID: PMC9059787.

Pirozzi F, Berkseth M, Shear R, Gonzalez L, Timms AE, Sulc J, Pao E, Oyama N, Forzano F, Conti V, Guerrini R, Doherty ES, Saitta SC, Lockwood CM, Pritchard CC, Dobyns WB, **Novotny E**, Wright JNN, **Saneto RP**, Friedman S, Hauptman J, Ojemann J, Kapur RP, Mirzaa GM. Profiling PI3K-AKT-MTOR variants in focal brain malformations reveals new insights for diagnostic care. Brain. 2022 Apr 29;145(3):925-938. doi: 10.1093/brain/awab376. Erratum in: Brain. 2022 Oct 19;: PMID: 35355055; PMCID: PMC9630661.

Maio N, **Saneto RP**, Steet R, Sotero de Menezes MA, Skinner C, Rouault TA. Disruption of cellular iron homeostasis by *IREB2* missense variants causes severe neurodevelopmental delay, dystonia and seizures. Brain Commun. 2022 Apr 19;4(3):fcac102. doi: 10.1093/braincomms/fcac102. PMID: 35602653; PMCID: PMC9118103.

Prasad N, **Castillo-Pinto C**, Safadi AL, Osborne B. Neurological Considerations for the Care of Patients With Severe Obesity. Neurohospitalist. 2022 Apr;12(2):264-267. doi: 10.1177/19418744211040697. Epub 2021 Aug 25. PMID: 35419155; PMCID: PMC8995583.

Williams CN, Hall TA, Francoeur C, Kurz J, Rasmussen L, Hartman ME, O'meara AI, Ferguson NM, Fink EL, Walker T, Drury K, Carpenter JL, Erklauer J, Press C, **Wainwright MS**, Lovett M, Dapul H, Murphy S, Risen S, Guerriero RM, Woodruff A, Guiliams KP; PEDIATRIC NEUROCITICAL CARE RESEARCH GROUP (PNCRG).. Continuing Care For Critically Ill Children Beyond Hospital Discharge: Current State of Follow-up. Hosp Pediatr. 2022 Apr 1;12(4):359-393. doi: 10.1542/hpeds.2021-006464. PMID: 35314865; PMCID: PMC9182716.

Vawter-Lee M, **Natarajan N**, Rang K, Horn PS, Pardo AC, Thomas CW. Topiramate Is Safe for Refractory Neonatal Seizures: A Multicenter Retrospective Cohort Study of Necrotizing Enterocolitis Risk. Pediatr Neurol. 2022 Apr;129:7-13. doi: 10.1016/j.pediatrneurol.2021.12.003. Epub 2021 Dec 30. PMID: 35131568.

Keene JC, **Morgan LA**, Abend NS, Bates SV, Bauer Huang SL, Chang T, Chu CJ, Glass HC, Massey SL, Ostrander B, Pardo AC, Press CA, Soul JS, Shellhaas RA, Thomas C, **Natarajan N**. Treatment of Neonatal Seizures: Comparison of Treatment Pathways From 11 Neonatal Intensive Care Units. Pediatr Neurol. 2022 Mar;128:67-74. doi: 10.1016/j.pediatrneurol.2021.10.004. Epub 2021 Oct 11. PMID: 34750046.

Keene JC, **Wainwright M**, **Morgan LA**, Mietzsch U, Musa N, **Bozarth XL**, **Natarajan N**. Retrospective Evaluation of First-line Levetiracetam use for Neonatal Seizures after Congenital Heart Defect repair with or without Extracorporeal Membrane Oxygenation. J Pediatr Pharmacol Ther. 2022;27(3):254-262. doi: 10.5863/1551-6776-27.3.254. Epub 2022 Mar 21. PMID: 35350164; PMCID: PMC8939278.

Fink EL, Robertson CL, **Wainwright MS**, Roa JD, Lovett ME, Stulce C, Yacoub M, Potera RM, Zivick E, Holloway A, Nagpal A, Wellnitz K, Czech T, Even KM, Brunow de Carvalho W, Rodriguez IS, Schwartz SP, Walker TC, Campos-Miño S, Dervan LA, Geneslaw AS, Sewell TB, Pryce P, Silver WG, Lin JE, Vargas WS, Topjian A, Alcamo AM, McGuire JL, Domínguez Rojas JA, Muñoz JT, Hong SJ, Muller WJ, Doerfler M, Williams CN, Drury K, Bhagat D, Nelson A, Price D, Dapul H, Santos L, Kahoud R, Francoeur C, Appavu B, Guiliams KP, Agner SC, Walson KH, Rasmussen L, Janas A, Ferrazzano P, Farias-Moeller R, Snooks KC, Chang CH, Yun J, Schober ME; Global Consortium Study of Neurologic Dysfunction in COVID-19 (GCS-NeuroCOVID) Investigators. Prevalence and Risk Factors of Neurologic Manifestations in Hospitalized Children Diagnosed with Acute SARS-CoV-2 or MIS-C. Pediatr Neurol. 2022 Mar;128:33-44. doi: 10.1016/j.pediatrneurol.2021.12.010. Epub 2021 Dec 28. PMID: 35066369; PMCID: PMC8713420.

Farias-Moeller R, Jayakar A, Guerriero RM, Carpenter JL, **Wainwright MS**, Harrar DB. Pediatric Critical Care Neurologists in the United States and Canada: A Survey of Clinical Practice Experience. J Child Neurol. 2022 Mar;37(4):288-297. doi: 10.1177/08830738211070099. Epub 2022 Jan 17. PMID: 35037772.

Pediatric Neurology Faculty Publications 2014-2025

- Ko PY, Khalatbari H, Hatt D, Coufal N, Barry D, **Wainwright MS**, Khanna PC, **Amlie-Lefond C**. Hemorrhagic Transformation Following Childhood Cardioembolic Stroke Is Not Increased in Anticoagulated Patients. *J Child Neurol*. 2022 Mar;37(4):273-280. doi: 10.1177/08830738211025866. Epub 2022 Jan 13. PMID: 35021919.
- Steinman KJ**, Stone WL, Ibañez LV, Attar SM. Reducing Barriers to Autism Screening in Community Primary Care: A Pragmatic Trial Using Web-Based Screening. *Acad Pediatr*. 2022 Mar;22(2):263-270. doi: 10.1016/j.acap.2021.04.017. Epub 2021 Apr 23. PMID: 33901728; PMCID: PMC8536796.
- Price D, Dapul H, Santos L, Kahoud R, Francoeur C, Appavu B, Williams KP, Agner SC, Walson KH, Rasmussen L, Janas A, Ferrazzano P, Farias-Moeller R, Snooks KC, Chang CH, Yun J, Schober ME; Global Consortium Study of Neurologic Dysfunction in COVID-19 (GCS-NeuroCOVID) Investigators. Prevalence and Risk Factors of Neurologic Manifestations in Hospitalized Children Diagnosed with Acute SARS-CoV-2 or MIS-C. *Pediatr Neurol*. 2022 Mar;128:33-44. doi: 10.1016/j.pediatrneurol.2021.12.010. Epub 2021 Dec 28. PMID: 35066369; PMCID: PMC8713420.
- Kuo CH, Casimo K, Wu J, Collins K, Rice P, Chen BW, Yang SH, Lo YC, **Novotny EJ**, Weaver KE, Chen YY, Ojemann JG. Electrooculography to Investigate Age-Related Brain Lateralization on Pediatric Motor Inhibition. *Front Neurol*. 2022 Mar 7;13:747053. doi: 10.3389/fneur.2022.747053. PMID: 35330804; PMCID: PMC8940229.
- Tunc EM, **Otten CE**, Kodish IM, Kim SJ, Kochar A, **Novotny E**, Yoshida H. Seventeen-Year-Old Female With History of Depression Presented With Mania and Enuresis. *Pediatrics*. 2022 Mar 1;149(3):e2021051176. doi: 10.1542/peds.2021-051176. PMID: 35229119.
- Knupp KG, Coryell J, Singh RK, Gaillard WD, Shellhaas RA, Koh S, Mitchell WG, Harini C, Millichap JJ, May A, Dlugos D, Nickels K, Mytinger JR, Keator C, Yozawitz E, Singhal N, **Lockrow J**, Thomas JF, Juarez-Colunga E. Comparison of Cosyntropin, Vigabatrin, and Combination Therapy in New-Onset Infantile Spasms in a Prospective Randomized Trial. *J Child Neurol*. 2022 Mar;37(3):186-193. doi: 10.1177/08830738211073400. Epub 2022 Jan 19. PMID: 35044272.
- Harrar DB, **Benedetti GM**, Jayakar A, Carpenter JL, Mangum TK, Chung M, Appavu B; International Pediatric Stroke Study Group and Pediatric Neurocritical Care Research Group. Pediatric Acute Stroke Protocols in the United States and Canada. *J Pediatr*. 2022 Mar;242:220-227.e7. doi: 10.1016/j.jpeds.2021.10.048. Epub 2021 Nov 11. PMID: 34774972
- Bornstein R, James K, Stokes J, Park KY, Kayser EB, Snell J, Bard A, Chen Y, Kalume F, **Johnson SC**. Differential effects of mTOR inhibition and dietary ketosis in a mouse model of subacute necrotizing encephalomyelopathy. *Neurobiol Dis*. 2022 Feb;163:105594. doi: 10.1016/j.nbd.2021.105594. Epub 2021 Dec 20. PMID: 34933094; PMCID: PMC8770160
- Conway KM, Gedlinske A, Mathews KD, **Perlman S**, Johnson N, Butterfield R, Hung M, Bounsanga J, Matthews D, Oleszek J, Romitti PA. A population-based study of scoliosis among males diagnosed with a dystrophinopathy identified by the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet). *Muscle Nerve*. 2022 Feb;65(2):193-202. doi: 10.1002/mus.27464. Epub 2021 Dec 1. PMID: 34787322; PMCID: PMC8752499.
- Shalabi H, Nellan A, Shah NN, **Gust J**. Immunotherapy Associated Neurotoxicity in Pediatric Oncology. *Front Oncol*. 2022 Feb 21;12:836452. doi: 10.3389/fonc.2022.836452. PMID: 35265526; PMCID: PMC8899040.
- Madden LK, Rajajee V, Human T, **Wainwright MS**, Guanci M, Mainali S, Rowe S, McLaughlin D, Lunde J, Lele A, Fried H. Correction to: Neurocritical Care Society Guidelines Update: Lessons from a Decade of GRADE Guidelines. *Neurocrit Care*. 2022 Feb;36(1):333. doi: 10.1007/s12028-021-01411-0. Erratum for: *Neurocrit Care*. 2022 Feb;36(1):1-10. PMID: 34874505; PMCID: PMC9172880.
- Jung S, Kayser EB, **Johnson SC**, Li L, Worstman HM, Sun GX, Sedensky MM, Morgan PG. Tetraethylammonium chloride reduces anaesthetic-induced neurotoxicity in *Caenorhabditis elegans* and mice. *Br J Anaesth*. 2022 Jan;128(1):77-88. doi: 10.1016/j.bja.2021.09.036. Epub 2021 Nov 30. PMID: 34857359; PMCID: PMC8787783
- Sen K, Whitehead M, **Castillo Pinto C**, Caldovic L, Gropman A. Fifteen years of urea cycle disorders brain research: Looking back, looking forward. *Anal Biochem*. 2022 Jan 1;636:114343. doi: 10.1016/j.ab.2021.114343. Epub 2021 Oct 9. PMID: 34637785; PMCID: PMC8671367.
- Castillo-Pinto C**, Lamotte G, Mehta A, Sonti R, Di Maria G, Ruiz D, Kumar PN, Stermer AB, Denny MC. Healthcare Worker With Large Vessel Acute Ischemic Stroke Likely Related to Mild SARS-CoV-2 Infection. *Neurohospitalist*. 2022 Jan;12(1):48-56. doi: 10.1177/1941874420966845. Epub 2020 Nov 10. PMID: 34950386; PMCID: PMC8689530.

Pediatric Neurology Faculty Publications 2014-2025

Kirsch MP, LaRovere K, Balakrishnan B, Erklauer J, Francoeur C, Ganesan SL, Jayakar A, Lovett M, Luchette M, Press CA, Wolf M, Ferrazzano P, **Wainwright MS**, Appavu B; Pediatric Neurocritical Care Research Group (PNCRG). A Survey of Neuromonitoring Practices in North American Pediatric Intensive Care Units. *Pediatr Neurol*. 2022 Jan;126:125-130. doi: 10.1016/j.pediatrneurol.2021.11.002. Epub 2021 Nov 12. PMID: 34864306; PMCID: PMC9135309.

Stokes JC, Bornstein RL, James K, Park KY, Spencer K, Vo K, Snell JC, Johnson BM, Morgan PG, Sedensky MM, Baertsch N, **Johnson SC**: Leukocytes mediate disease pathogenesis in the Ndufs4(KO) mouse model of Leigh syndrome. *JCI Insight*. Jan 20, 2022. PMID: 35050903

Keene JC, Woods B, **Wainwright M**, King M, **Morgan LA**. Optimized Benzodiazepine Treatment of Pediatric Status Epilepticus Through a Standardized Emergency Medical Services Resuscitation Tool. *Pediatr Neurol*. 2022 Jan;126:50-55. doi: 10.1016/j.pediatrneurol.2021.10.001. Epub 2021 Oct 9. PMID: 34736064.

Shurtleff HA, Poliakov A, Barry D, Wright JN, Warner MH, **Novotny EJ**, Marashly A, Buckley R, Goldstein HE, Hauptman JS, Ojemann JG, Shaw DWW. A clinically applicable functional MRI memory paradigm for use with pediatric patients. *Epilepsy Behav*. 2022 Jan;126:108461. doi: 10.1016/j.yebeh.2021.108461. Epub 2021 Dec 9. PMID: 34896785.

Wainwright MS, Williams K, Kannan S, Simon DW, Tasker RC, Traube C, Pineda J; Pediatric Organ Dysfunction Information Update Mandate (PODIUM) Collaborative. Acute Neurologic Dysfunction in Critically Ill Children: The PODIUM Consensus Conference. *Pediatrics*. 2022 Jan 1;149(1 Suppl 1):S32-S38. doi: 10.1542/peds.2021-052888E. PMID: 34970681

Bemba MM, Agus M, Akcan-Arikan A, Alexander P, Basu R, Bennett TD, Bohn D, Brandão LR, Brown AM, Carcillo JA, Checchia P, Cholette J, Cheifetz IM, Cornell T, Doctor A, Eckerle M, Erickson S, Farris RWD, Faustino EVS, Fitzgerald JC, Fuhrman DY, Giuliano JS, Williams K, Gaies M, Gorga SM, Hall M, Hanson SJ, Hartman M, Hassinger AB, Irving SY, Jeffries H, Jouvet P, Kannan S, Karam O, Khemani RG, Kissoon N, Lacroix J, Laussen P, Leclerc F, Lee JH, Lefebvre S, Lobner K, McKiernan PJ, Menon K, Monagle P, Muszynski JA, Odetola F, Parker R, Pathan N, Pierce RW, Pineda J, Prince JM, Robinson KA, Rowan CM, Ryerson LM, Sanchez-Pinto LN, Schlapbach LJ, Selewski DT, Shekerdemian LS, Simon D, Smith LS, Squires JE, Squires RH, Sutherland SM, Ouellette Y, Spaeder MC, Srinivasan V, Steiner ME, Tasker RC, Thiagarajan R, Thomas N, Tissieres P, Traube C, Tucci M, Typpo KV, **Wainwright MS**, Ward SL, Watson RS, Weiss S, Whitney J, Willson D, Wynn JL, Yehya N, Zimmerman JJ. Pediatric Organ Dysfunction Information Update Mandate (PODIUM) Contemporary Organ Dysfunction Criteria: Executive Summary. *Pediatrics*. 2022 Jan 1;149(1 Suppl 1):S1-S12. doi: 10.1542/peds.2021-052888B. PMID: 34970673; PMCID: PMC9599725.

Tully HM, Doherty D, **Wainwright M**. Mortality in pediatric hydrocephalus. *Dev Med Child Neurol*. 2022 Jan;64(1):112-117. doi: 10.1111/dmcn.14975. Epub 2021 Jul 15. PMID: 34268734; PMCID : PMC8671148

2021

Faulhaber LD, Phuong AQ, Hartsuyker KJ, Cho Y, Mand KK, Harper SD, Olson AK, Garden GA, Shih AY, **Gust J**. Brain capillary obstruction during neurotoxicity in a mouse model of anti-CD19 chimeric antigen receptor T-cell therapy. *Brain Commun*. 2021 Dec 31;4(1):fcab309. doi: 10.1093/braincomms/fcab309. PMID: 35169706; PMCID: PMC8833245.

Castillo-Pinto C, Sen K, Gropman A. Neuromonitoring in Rare Disorders of Metabolism. *Yale J Biol Med*. 2021 Dec 29;94(4):645-655. PMID: 34970103; PMCID: PMC8686771.

Bornstein R, James K, Stokes J, Park KY, Snell J, Pan A, Bard A, Kalume F, **Johnson SC**: Differential effects of mTOR inhibition and dietary ketosis in a mouse model of subacute necrotizing encephalomyelopathy. *Neurobiology of Disease*. Dec 20, 2021. PMID: 34933094

Shurtleff HA, Poliakov A, Barry D, Wright JN, Warner MH, **Novotny EJ**, **Marashly A**, Buckley R, Goldstein HE, Hauptman JS, Ojemann JG, Shaw A clinically applicable functional MRI memory paradigm for use with pediatric patients. *DWW.Epilepsy Behav*. 2021 Dec 9;126:108461. doi: 10.1016/j.yebeh.2021.108461. Online ahead of print. PMID: 34896785

Wainwright MS. Editorial: The path to resilience and recovery: understanding the epidemiology, neuropathology and treatment of neurologic injury due to the SARS-CoV-2 virus in children. *Curr Opin Pediatr*. 2021 Dec 1;33(6):576-579. doi: 10.1097/MOP.0000000000001079. PMID: 34654051; PMCID: PMC8577292.

Pediatric Neurology Faculty Publications 2014-2025

Vasquez A, Farias-Moeller R, Sánchez-Fernández I, Abend NS, Amengual-Gual M, Anderson A, Arya R, Brenton JN, Carpenter JL, Chapman K, Clark J, Gaillard WD, Glauser T, Goldstein JL, Goodkin HP, Guerriero RM, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Riviello JJ, Sands TT, Sannagowdara K, Tasker RC, Tchapyjnikov D, Topjian A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group (pSERG). Super-Refractory Status Epilepticus in Children: A Retrospective Cohort Study. *Pediatr Crit Care Med*. 2021 Dec 1;22(12):e613-e625

Patel PB, Bearden D. Neuropathogenesis of severe acute respiratory syndrome coronavirus 2. *Curr Opin Pediatr*. 2021 Dec 1;33(6):597-602. doi: 10.1097/MOP.0000000000001068. PMID: 34734914; PMCID: PMC8577294.

Madden LK, Rajajee V, Human T, **Wainwright MS**, Guanci M, Mainali S, Rowe S, McLaughlin D, Lunde J, Lele A, Fried H. Neurocritical Care Society Guidelines Update: Lessons from a Decade of GRADE Guidelines. *Neurocrit Care*. 2021 Nov 2;1–10. doi: 10.1007/s12028-021-01375-1. Epub ahead of print. Erratum in: *Neurocrit Care*. 2021 Dec 7;; PMID: 34729676; PMCID: PMC8562933

Sheehan T, Amengual-Gual M, Vasquez A, Abend NS, Anderson A, Appavu B, Arya R, Barcia Aguilar C, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Gaínza-Lein M, Glauser TA, Goldstein JL, Goodkin HP, Guerriero RM, Huh L, Jackson M, Kapur K, Kahoud R, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Reece L, Riviello JJ, Sands TT, Sannagowdara K, Shellhaas R, Smith G, Tasker RC, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Zhang B, Loddenkemper T; Pediatric Status Epilepticus Research Group. Benzodiazepine administration patterns before escalation to second-line medications in pediatric refractory convulsive status epilepticus. *Epilepsia*. 2021 Nov;62(11):2766-2777. doi: 10.1111/epi.17043. Epub 2021 Aug 21. PMID: 34418087.

Conway KM, Gedlinske A, Mathews KD, **Perlman S**, Johnson N, Butterfield R, Hung M, Bounsanga J, Matthews D, Oleszek J, Romitti PA. A population-based study of scoliosis among males diagnosed with a dystrophinopathy identified by the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet). *Muscle Nerve*. 2021 Nov 17. doi: 10.1002/mus.27464. Epub ahead of print. PMID: 34787322.

Kirschen MP, LaRovere K, Balakrishnan B, Erklauer J, Francoeur C, Ganesan SL, Jayakar A, Lovett M, Luchette M, Press CA, Wolf M, Ferrazzano P, **Wainwright MS**, Appavu B; Pediatric Neurocritical Care Research Group (PNCRG). A Survey of Neuromonitoring Practices in North American Pediatric Intensive Care Units. *Pediatr Neurol*. 2021 Nov 12;126:125-130. doi: 10.1016/j.pediatrneurol.2021.11.002. Epub ahead of print. PMID: 34864306

Harrar DB, **Benedetti GM**, Jayakar A, Carpenter JL, Mangum TK, Chung M, Appavu B; Pediatric Acute Stroke Protocols in the United States and Canada. International Pediatric Stroke Study Group and Pediatric Neurocritical Care Research Group. *J Pediatr*. 2021 Nov 11:S0022-3476(21)01048-9. doi: 10.1016/j.jpeds.2021.10.048. Online ahead of print. PMID: 34774972

Stokes J, Freed A, Pan A, Sun G, Bornstein R, Snell J, Park KY, Morgan P, Sedensky MM, **Johnson SC**: Mechanisms underlying neonate specific metabolic effects of volatile anesthetics. *eLIFE*. Jul 12, 2021. PMID: 34254587.

Cheff DM, Muotri AR, Stockwell BR, Schmidt EE, Ran Q, Kartha RV, **Johnson SC**, Mittel P, Arner ESJ, Wigby KM, Hall MD, Rames SK: Development of Therapies for Rare Genetic Disorders of GPX4: Roadmap and Opportunities. *Orphanet Journal of Rare Diseases*. Epub 2021 Oct 23. PMID: 34688299.

Beatty CW, **Lockrow JP**, Gedela S, Gehred A, Ostendorf AP. The Missed Value of Underutilizing Pediatric Epilepsy Surgery: A Systematic Review. *Semin Pediatr Neurol*. 2021 Oct;39:100917. doi: 10.1016/j.spen.2021.100917. Epub 2021 Aug 19. PMID: 34620465.

Lee S, **Amlie-Lefond CM**. Target Practice: Aiming for Automated Perfusion in Childhood Stroke. *Stroke*. 2021 Oct;52(10):3305-3307. doi: 10.1161/STROKEAHA.121.036218. Epub 2021 Aug 18. PMID: 34404239 No abstract available.

Schober ME, Robertson CL, **Wainwright MS**, Roa JD, Fink EL. COVID-19 and the Pediatric Nervous System: Global Collaboration to Meet a Global Need. *Neurocrit Care*. 2021 Oct;35(2):283-290. doi: 10.1007/s12028-021-01269-2. Epub 2021 Jun 28. PMID: 34184177; PMCID: PMC8238033.

Keene JC, **Morgan LA**, Abend NS, Bates SV, Bauer Huang SL, Chang T, Chu CJ, Glass HC, Massey SL, Ostrander B, Pardo AC, Press CA, Soul JS, Shellhaas RA, Thomas C, **Natarajan N**. Treatment of Neonatal Seizures: Comparison of

Pediatric Neurology Faculty Publications 2014-2025

Treatment Pathways From 11 Neonatal Intensive Care Units. *Pediatr Neurol.* 2021 Oct 11:S0887-8994(21)00222-8. doi: 10.1016/j.pediatrneurol.2021.10.004. Online ahead of print.PMID: 34750046

Keene JC, Woods B, **Wainwright M**, King M, **Morgan LA**. Optimized Benzodiazepine Treatment of Pediatric Status Epilepticus Through a Standardized Emergency Medical Services Resuscitation Tool *Pediatr Neurol.* 2021 Oct 9;126:50-55. doi: 10.1016/j.pediatrneurol.2021.10.001. Online ahead of print.PMID: 34736064

Johnson JO, Chia R, Miller DE, Li R, Kumaran R, Abramzon Y, Alahmady N, Renton AE, Topp SD, Gibbs JR, Cookson MR, Sabir MS, Dalgard CL, Troakes C, Jones AR, Shatunov A, Iacoangeli A, Al Khleifat A, Ticozzi N, Silani V, Gellera C, Blair IP, Dobson-Stone C, Kwok JB, Bonkowski ES, Palvadeau R, Tienari PJ, Morrison KE, Shaw PJ, Al-Chalabi A, Brown RH Jr, Calvo A, Mora G, Al-Saif H, Gotkine M, Leigh F, Chang IJ, **Perlman SJ**, Glass I, Scott AI, Shaw CE, Basak AN, Landers JE, Chiò A, Crawford TO, Smith BN, Traynor BJ; FALS Sequencing Consortium; American Genome Center; International ALS Genomics Consortium; and ITALSGEN Consortium, Smith BN, Ticozzi N, Fallini C, Gkazi AS, Topp SD, Scotter EL, Kenna KP, Keagle P, Tiloca C, Vance C, Troakes C, Colombrita C, King A, Pensato V, Castellotti B, Baas F, Ten Asbroek ALMA, McKenna-Yasek D, McLaughlin RL, Polak M, Asress S, Esteban-Pérez J, Stevic Z, D'Alfonso S, Mazzini L, Comi GP, Del Bo R, Ceroni M, Gagliardi S, Querin G, Bertolin C, van Rheenen W, Rademakers R, van Blitterswijk M, Lauria G, Duga S, Corti S, Cereda C, Corrado L, Sorarù G, Williams KL, Nicholson GA, Blair IP, Leblond-Manry C, Rouleau GA, Hardiman O, Morrison KE, Veldink JH, van den Berg LH, Al-Chalabi A, Pall H, Shaw PJ, Turner MR, Talbot K, Taroni F, García-Redondo A, Wu Z, Glass JD, Gellera C, Ratti A, Brown RH Jr, Silani V, Shaw CE, Landers JE, Dalgard CL, Adeleye A, Soltis AR, Alba C, Viollet C, Bacikova D, Hupalo DN, Sukumar G, Pollard HB, Wilkerson MD, Martinez EM, Abramzon Y, Ahmed S, Arepalli S, Baloh RH, Bowser R, Brady CB, Brice A, Broach J, Campbell RH, Camu W, Chia R, Cooper-Knock J, Ding J, Drepper C, Drory VE, Duncley TL, Eicher JD, England BK, Faghri F, Feldman E, Floeter MK, Fratta P, Geiger JT, Gerhard G, Gibbs JR, Gibson SB, Glass JD, Hardy J, Harms MB, Heiman-Patterson TD, Hernandez DG, Jansson L, Kirby J, Kowall NW, Laaksovirta H, Landeck N, Landi F, Le Ber I, Lumbroso S, MacGowan DJL, Maragakis NJ, Mora G, Mouzat K, Murphy NA, Myllykangas L, Nalls MA, Orrell RW, Ostrow LW, Pamphlett R, Pickering-Brown S, Pioro EP, Pletnikova O, Pliner HA, Pulst SM, Ravits JM, Renton AE, Rivera A, Robberecht W, Rogaeva E, Rollinson S, Rothstein JD, Scholz SW, Sendtner M, Shaw PJ, Sidle KC, Simmons Z, Singleton AB, Smith N, Stone DJ, Tienari PJ, Troncoso JC, Valori M, Van Damme P, Van Deerlin VM, Van Den Bosch L, Zinman L, Landers JE, Chiò A, Traynor BJ, Angelocola SM, Ausiello FP, Barberis M, Bartolomei I, Battistini S, Bersano E, Bisogni G, Borghero G, Brunetti M, Cabona C, Calvo A, Canale F, Canosa A, Cantisani TA, Capasso M, Caponnetto C, Cardinali P, Carrera P, Casale F, Chiò A, Colletti T, Conforti FL, Conte A, Conti E, Corbo M, Cuccu S, Dalla Bella E, D'Errico E, DeMarco G, Dubbioso R, Ferrarese C, Ferraro PM, Filippi M, Fini N, Floris G, Fuda G, Gallone S, Gianferrari G, Giannini F, Grassano M, Greco L, Iazzolino B, Introna A, La Bella V, Lattante S, Lauria G, Liguori R, Logroscino G, Logullo FO, Lunetta C, Mandich P, Mandrioli J, Manera U, Manganelli F, Marangi G, Marinou K, Marrosu MG, Martinelli I, Messina S, Moglia C, Mora G, Mosca L, Murru MR, Origone P, Passaniti C, Petrelli C, Petrucci A, Pozzi S, Pugliatti M, Quattrini A, Ricci C, Riolo G, Riva N, Russo M, Sabatelli M, Salamone P, Salivetto M, Salvi F, Santarelli M, Sbaiz L, Sideri R, Simone I, Simonini C, Spataro R, Tanel R, Tedeschi G, Ticca A, Torriello A, Tranquilli S, Tremolizzo L, Trojsi F, Vasta R, Vacchiano V, Vita G, Volanti P, Zollino M, Zucchi E. Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurol.* 2021 Oct 1;78(10):1236-1248. doi: 10.1001/jamaneurol.2021.2598. PMID: 34459874; PMCID: PMC8406220.

Prendergast E, Allen KY, Mills MG, Moran T, Harris ZL, Malakooti M, Smith CM, **Wainwright MS**, McCarthy-Kowols M. Targeted Temperature Management Protocol in a Pediatric Intensive Care Unit: A Quality Improvement Project. *Crit Care Nurse.* 2021 Oct 1;41(5):41-50. doi: 10.4037/ccn2021554. PMID: 34595494.

Gaínza-Lein M, Barcia Aguilar C, Piantino J, Chapman KE, Sánchez Fernández I, Amengual-Gual M, Anderson A, Appavu B, Arya R, Brenton JN, Carpenter JL, Clark J, Farias-Moeller R, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Huh L, Kahoud R, Kapur K, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, Nayak A, **Novotny E Jr**, Ostendorf AP, Payne ET, Peariso K, Reece L, Riviello J, Sannagowdara K, Sands TT, Sheehan T, Tasker RC, Tchapyjnikov D, Vasquez A, **Wainwright MS**, Wilfong A, Williams K, Zhang B, Loddenkemper T; Factors associated with long-term outcomes in pediatric refractory status epilepticus. *Pediatric Status Epilepticus Research Group.Epilepsia.* 2021 Sep;62(9):2190-2204. doi: 10.1111/epi.16984. Epub 2021 Jul 12.PMID: 34251039

Park KB, Chapman T, Aldinger KA, Mirzaa GM, Zeiger J, Beck A, Glass IA, Hevner RF, Jansen AC, Marshall DA, Oegema R, Parrini E, **Saneto RP**, Curry CJ, Hall JG, Guerrini R, Leventer RJ, Dobyns WB. The spectrum of brain malformations and disruptions in twins. *Am J Med Genet A.* 2021 Sep;185(9):2690-2718. doi: 10.1002/ajmg.a.61972. Epub 2020 Nov 18. PMID: 33205886; PMCID: PMC8683564

Vitanza NA, Johnson AJ, Wilson AL, Brown C, Yokoyama JK, Künkele A, Chang CA, Rawlings-Rhea S, Huang W, Seidel K, Albert CM, Pinto N, **Gust J**, Finn LS, Ojemann JG, Wright J, Orentas RJ, Baldwin M, Gardner RA, Jensen MC, Park JR. Locoregional infusion of HER2-specific CAR T cells in children and young adults with recurrent or refractory CNS

Pediatric Neurology Faculty Publications 2014-2025

tumors: an interim analysis. *Nat Med*. 2021 Sep;27(9):1544-1552. doi: 10.1038/s41591-021-01404-8. Epub 2021 Jul 12. PMID: 34253928 Clinical Trial.

Sun LR, Harrar D, Drocton G, **Castillo-Pinto C**, Gailloud P, Pearl MS. Endovascular therapy for acute stroke in children: age and size technical limitations. *J Neurointerv Surg*. 2021 Sep;13(9):794-798. doi: 10.1136/neurintsurg-2021-017311. Epub 2021 Apr 8. PMID: 33832970.

Shurtleff HA, Roberts EA, Young CC, Barry D, Warner MH, **Saneto RP**, Buckley R, Firman T, Poliakov AV, Ellenbogen RG, Hauptman JS, Ojemann JG, **Marashly A**. Pediatric hemispherectomy outcome: Adaptive functioning, intelligence, and memory. *Epilepsy Behav*. 2021 Sep 16;124:108298. doi: 10.1016/j.yebeh.2021.108298. Epub ahead of print. PMID: 34537627.

Jung S, Kayser EB, **Johnson SC**, Li L, Worstman H, Sun G, Sedensky MM, Morgan PG: Tetraethylammonium chloride reduces anaesthetic-induced neurotoxicity in *Caenorhabditis elegans* and mice. *British Journal of Anaesthesia*. <https://doi.org/10.1016/j.bja.2021.09.036>, PMID: 34857359.

Miller DE, Sulovari A, Wang T, Loucks H, Hoekzema K, Munson KM, Lewis AP, Fuerte EPA, Paschal CR, Walsh T, Thies J, Bennett JT, Glass I, Dipple KM, Patterson K, Bonkowski ES, Nelson Z, Squire A, Sikes M, Beckman E, Bennett RL, Earl D, Lee W, Allikmets R, **Perlman SJ**, Chow P, Hing AV, Wenger TL, Adam MP, Sun A, Lam C, Chang I, Zou X, Austin SL, Huggins E, Safi A, Iyengar AK, Reddy TE, Majoros WH, Allen AS, Crawford GE, Kishnani PS; University of Washington Center for Mendelian Genomics, King MC, Cherry T, Chong JX, Bamshad MJ, Nickerson DA, Mefford HC, Doherty D, Eichler EE. Targeted long-read sequencing identifies missing disease-causing variation. *Am J Hum Genet*. 2021 Aug 5;108(8):1436-1449. doi: 10.1016/j.ajhg.2021.06.006. Epub 2021 Jul 2. PMID: 34216551; PMCID: PMC8387463.

Zhang J, Tang W, Bhatia NK, Xu Y, Paudyal N, Liu D, Kim S, Song R, XiangWei W, Shaulsky G, Myers SJ, Dobyns W, Jayaraman V, Traynelis SF, Yuan H, **Bozarth X**. A *de novo* *GRIN1* Variant Associated With Myoclonus and Developmental Delay: From Molecular Mechanism to Rescue Pharmacology. *Front Genet*. 2021 Aug 3;12:694312. doi: 10.3389/fgene.2021.694312. eCollection 2021. PMID: 34413877 Free PMC article.

Engelke UF, van Outersterp RE, Merx J, van Geenen FA, van Rooij A, Berden G, Huigen MC, Kluijtmans LA, Peters TM, Al-Shekaili HH, Leavitt BR, de Vrieze E, Broekman S, van Wijk E, Tseng LA, Kulkarni P, Rutjes FP, Mecinović J, Struys EA, Jansen LA, **Gospe SM Jr**, Mercimek-Andrews S, Hyland K, Willemsen MA, Bok LA, van Karnebeek CD, Wevers RA, Boltje TJ, Oomens J, Martens J, Coene KL. Untargeted metabolomics and infrared ion spectroscopy identify biomarkers for pyridoxine-dependent epilepsy *J Clin Invest*. 2021 Aug 2;131(15):e148272. doi: 10.1172/JCI148272. PMID: 34138754 Free PMC article.

Jennifer I Koop , Kevin Credille , Yang Wang , Michelle Loman , **Ahmad Marashly** , Irene Kim , Sean M Lew , Mohit Maheshwari Determination of language dominance in pediatric patients with epilepsy for clinical decision-making: Correspondence of intracarotid amobarbital procedure and fMRI modalities *Epilepsy Behav* . 2021 Aug;121(Pt A):108041.

Hannah M Tully, Dan Doherty, **Mark Wainwright**. Mortality in pediatric hydrocephalus *Dev Med Child Neurol*. 2021 Jul 15. doi: 10.1111/dmcn.14975. Online ahead of print.

Stokes J, Freed A, Pan A, Sun G, Bornstein R, Snell J, Park KY, Morgan P, Sedensky MM, **Johnson SC**: Mechanisms underlying neonate specific metabolic effects of volatile anesthetics. *eLIFE*. Jul 12, 2021. PMID: 34254587

Gospe SM Jr. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Mirzaa G, Amemiya A, editors. Pyridoxine-Dependent Epilepsy – *ALDH7A1* GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2021. 2001 Dec 7 [updated 2021 Jul 29].

Edmonds BD, Welch W, Sogawa Y, Mountz J, Bagić A, Patterson C. The Role of Magnetoencephalography and Single-Photon Emission Computed Tomography in Evaluation of Children With Drug-Resistant Epilepsy. *J Child Neurol*. 2021 Jul;36(8):673-679. doi: 10.1177/0883073821996558. Epub 2021 Mar 5. PMID: 33663250.

Sculier C, Barcia Aguilar C, Gaspard N, Gaínza-Lein M, Sánchez Fernández I, Amengual-Gual M, Anderson A, Arya R, Burrows BT, Brenton JN, Carpenter JL, Chapman KE, Clark J, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Gorman M, Lai YC, McDonough TL, Mikati MA, Nayak A, Peariso K, Riviello J, Rusie A, Sperberg K, Stredny CM, Tasker RC, Tchapyjnikov D, Vasquez A, **Wainwright MS**, Wilfong AA, Williams K, Loddenkemper T; Clinical presentation of new

Pediatric Neurology Faculty Publications 2014-2025

onset refractory status epilepticus in children (the pSERG cohort). *pSERG.Epilepsia*. 2021 Jul;62(7):1629-1642. doi: 10.1111/epi.16950. Epub 2021 Jun 6.

Barcia Aguilar C, Amengual-Gual M, Sánchez Fernández I, Abend NS, Anderson A, Appavu B, Arya R, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Gaínza-Lein M, Glauser T, Goldstein JL, Goodkin HP, Guerriero RM, Huh L, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf A, Payne ET, Peariso K, Piantino J, Riviello J, Sannagowdara K, Sheehan T, Sands TT, Tasker RC, Tchapyjnikov D, Topjian AA, Vasquez A, **Wainwright MS**, Wilfong AA, Williams K, Loddenkemper T; pSERG. Time to Treatment in Pediatric Convulsive Refractory Status Epilepticus: The Weekend Effect. *Pediatr Neurol*. 2021 Jul;120:71-79. doi: 10.1016/j.pediatrneurol.2021.03.009. Epub 2021 Mar 26

Saneto RP, **Patrick KE**, Perez FA. Homoplasmy of the m. 8993 T>G variant in a patient without MRI findings of Leigh syndrome, ataxia or retinal abnormalities Mitochondrion. 2021 Jul;59:58-62. doi: 10.1016/j.mito.2021.04.010. Epub 2021 Apr 22.

Tarazi RA, **Patrick KE**, Lampietro M, Apollonsky N. Hydroxyurea Use Associated with Nonverbal and Executive Skills in Sick Cell Anemia. *J Pediatr Psychol*. 2021 Jul 20;46(6):710-718. doi: 10.1093/jpepsy/jsab015

Vitanza NA, Johnson AJ, Wilson AL, Brown C, Yokoyama JK, Künkele A, Chang CA, Rawlings-Rhea S, Huang W, Seidel K, Albert CM, Pinto N, **Gust J**, Finn LS, Ojemann JG, Wright J, Orentas RJ, Baldwin M, Gardner RA, Jensen MC, Park Locoregional infusion of HER2-specific CAR T cells in children and young adults with recurrent or refractory CNS tumors: an interim analysis. *J R.Nat Med*. 2021 Jul 12. doi: 10.1038/s41591-021-01404-8. Online ahead of print.

Gaínza-Lein M, Barcia Aguilar C, Piantino J, Chapman KE, Sánchez Fernández I, Amengual-Gual M, Anderson A, Appavu B, Arya R, Brenton JN, Carpenter JL, Clark J, Farias-Moeller R, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Huh L, Kahoud R, Kapur K, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, Nayak A, **Novotny E Jr**, Ostendorf AP, Payne ET, Peariso K, Reece L, Riviello J, Sannagowdara K, Sands TT, Sheehan T, Tasker RC, Tchapyjnikov D, Vasquez A, **Wainwright MS**, Wilfong A, Williams K, Zhang B, Loddenkemper T; Factors associated with long-term outcomes in pediatric refractory status epilepticus. *Pediatric Status Epilepticus Research Group.Epilepsia*. 2021 Jul 12. doi: 10.1111/epi.16984. Online ahead of print.

Oesch G, Perez FA, **Wainwright MS**, Shaw DWW, **Amlie-Lefond C**. Focal Cerebral Arteriopathy of Childhood: Clinical and Imaging Correlates. *Stroke*. 2021 Jul;52(7):2258-2265. doi: 10.1161/STROKEAHA.120.031880. Epub 2021 May 27.

Davies OMT, Garzon MC, Frieden IJ, Cottrell CE, Gripp KW, **Saneto RP**, Shwayder T, Mirzaa GM, Drolet BA. Cutaneous vascular anomalies associated with a mosaic variant of AKT3: Genetic analysis continues to refine the diagnosis, nomenclature, and classification of vascular anomalies. *Am Acad Dermatol*. 2021 Jul 6:S0190-9622(21)02067-3. doi: 10.1016/j.jaad.2021.06.877. Online ahead of print.

Schober ME, Robertson CL, **Wainwright MS**, Roa JD, Fink EL COVID-19 and the Pediatric Nervous System: Global Collaboration to Meet a Global Need. *Neurocrit Care*. 2021 Jun 28:1-8. doi: 10.1007/s12028-021-01269-2. Online ahead of print.

O'Brien NF, Reuter-Rice K, **Wainwright MS**, Kaplan SL, Appavu B, Erklauer JC, Ghosh S, Kirschen M, Kozak B, Lidsky K, Lovett ME, Mehollin-Ray AR, Miles DK, Press CA, Simon DW, Tasker RC, LaRovere KL Practice Recommendations for Transcranial Doppler Ultrasonography in Critically Ill Children in the Pediatric Intensive Care Unit: A Multidisciplinary Expert Consensus Statement. *J Pediatr Intensive Care*. 2021 Jun;10(2):133-142.

Vasquez A, Farias-Moeller R, Sánchez-Fernández I, Abend NS, Amengual-Gual M, Anderson A, Arya R, Brenton JN, Carpenter JL, Chapman K, Clark J, Gaillard WD, Glauser T, Goldstein JL, Goodkin HP, Guerriero RM, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Riviello JJ, Sands TT, Sannagowdara K, Tasker RC, Tchapyjnikov D, Topjian A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group (pSERG) Super-Refractory Status Epilepticus in Children: A Retrospective Cohort Study. *Pediatr Crit Care Med*. 2021 Jun 14.

Erickson SL, Killien EY, **Wainwright M**, Mills B, Vavilala MS. Mean Arterial Pressure and Discharge Outcomes in Severe Pediatric Traumatic Brain Injury. *Neurocrit Care*. 2021 Jun;34(3):1017-1025. doi: 10.1007/s12028-020-01121-z. Epub 2020 Oct 27. PMID: 33108627; PMCID: PMC8076334.

Pediatric Neurology Faculty Publications 2014-2025

Paczesny S, Pasquini MC, Pavletic SZ, Agarwal A, Spellman S, Kean L, Bernatchez C, **Gust J**, Staedtke V, Perales MA. Blueprint for the discovery of biomarkers of toxicity and efficacy for CAR T cells and T-cell engagers. *Blood Adv*. 2021 Jun 8;5(11):2519-2522.

Shalabi H, **Gust J**, Taraseviciute A, Wolters PL, Leahy AB, Sandi C, Laetsch TW, Wiener L, Gardner RA, Nussenblatt V, Hill JA, Curran KJ, Olson TS, Annesley C, Wang HW, Khan J, Pasquini MC, Duncan CN, Grupp SA, Pulsipher MA, Shah NN. Beyond the storm - subacute toxicities and late effects in children receiving CAR T cells. *Nat Rev Clin Oncol*. 2021 Jun;18(6):363-378.

Ferrazzano P, Yeske B, Mumford J, Kirk G, Bigler ED, **Bowen K**, O'Brien N, Rosario B, Beers SR, Rathouz P, Bell MJ, Alexander AL. Brain Magnetic Resonance Imaging Volumetric Measures of Functional Outcome after Severe Traumatic Brain Injury in Adolescents. *J Neurotrauma*. 2021 Jun 1;38(13):1799-1808. doi: 10.1089/neu.2019.6918. Epub 2021 Feb 24.

Acsadi G, Crawford TO, Müller-Felber W, Shieh PB, Richardson R, **Natarajan N**, Castro D, Ramirez-Schrempp D, Gambino G, Sun P, Farwell W. Safety and efficacy of nusinersen in spinal muscular atrophy: The EMBRACE study. *Muscle Nerve*. 2021 May;63(5):668-677. doi: 10.1002/mus.27187. Epub 2021 Feb 16.

Kelly JP, Phillips JO, **Saneto RP**, Khalatbari H, Poliakov A, Tarczy-Hornoch K, Weiss AH. Cerebral Visual Impairment Characterized by Abnormal Visual Orienting Behavior With Preserved Visual Cortical Activation. *Invest Ophthalmol Vis Sci*. 2021 May 3;62(6):15.

Khalatbari H, Wright JN, Ishak GE, Perez FA, **Amlie-Lefond CM**, Shaw DWW. Deep medullary vein engorgement and superficial medullary vein engorgement: two patterns of perinatal venous stroke. *Pediatr Radiol*. 2021 May;51(5):675-685.

Ferdinandusse S, McWalter K, Te Brinke H, IJlst L, Mooijer PM, Ruiters JPN, van Lint AEM, Pras-Raves M, Wever E, Millan F, Guillen Sacoto MJ, Begtrup A, Tarnopolsky M, Brady L, Ladda RL, Sell SL, Nowak CB, Douglas J, Tian C, Ulm E, **Perlman S**, Drack AV, Chong K, Martin N, Brault J, Brokamp E, Toro C, Gahl WA, Macnamara EF, Wolfe L; Undiagnosed Diseases Network, Waisfisz Q, Zwiijnenburg PJG, Ziegler A, Barth M, Smith R, Ellingwood S, Gaebler-Spira D, Bakhtiari S, Kruer MC, van Kampen AHC, Wanders RJA, Waterham HR, Cassiman D, Vaz FM. An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. *Genet Med*. 2021 Apr;23(4):740-750. doi: 10.1038/s41436-020-01027-3. Epub 2020 Nov 26.

Saneto RP, Patrick KE, Perez FA Homoplasmy of the m. 8993 T>G variant in a patient without MRI findings of Leigh syndrome, ataxia or retinal abnormalities. *Mitochondrion*. 2021 Apr 22;59:58-62.

Barry M, Barry D, Kansagra AP, Hallam D, Abraham M, **Amlie-Lefond C**; Thrombolysis in Pediatric Stroke (TIPSTER) Investigators*. Higher-Quality Data Collection Is Critical to Establish the Safety and Efficacy of Pediatric Mechanical Thrombectomy. *Stroke*. 2021 Apr;52(4):1213-1221.

Lauzier DC, Galardi MM, Williams KP, Goyal MS, **Amlie-Lefond C**, Hallam DK, Kansagra AP. Pediatric Thrombectomy: Design and Workflow Lessons From Two Experienced Centers. *Stroke*. 2021 Apr;52(4):1511-1519.

Steinman KJ, Stone WL, Ibañez LV, Attar SM. Reducing Barriers to Autism Screening in Community Primary Care: A Pragmatic Trial using Web-Based Screening. *Acad Pediatr*. 2021 Apr 23:S1876-2859(21)00237-0.

Novotny EJ Diverse genetic causes of polymicrogyria with epilepsy. Epilepsy Phenome/Genome Project, Epi4K Consortium. *Epilepsia*. 2021 Apr;62(4):973-983. doi: 10.1111/epi.16854. Epub 2021 Apr 5.

Simpson TS, Peterson RL, **Patrick KE**, Forster JE, McNally KA. Concussion Symptom Treatment and Education Program: A Feasibility Study *J Head Trauma Rehabil*. 2021 Mar-Apr 01;36(2):E79-E88.

Weiss KE, **Steinman KJ**, Kodish I, Sim L, Yurs S, Steggall C, Fobian AD. Functional Neurological Symptom Disorder in Children and Adolescents within Medical Settings. *J Clin Psychol Med Settings*. 2021 Mar;28(1):90-101. doi: 10.1007/s10880-020-09736-2.

Barcia Aguilar C, Amengual-Gual M, Sánchez Fernández I, Abend NS, Anderson A, Appavu B, Arya R, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Gaínza-Lein M, Glauser T, Goldstein JL, Goodkin HP, Guerriero RM, Huh L, Lai YC, McDonough TL, Mikati MA, **Morgan LA, Novotny EJ**, Ostendorf A, Payne ET, Peariso K, Piantino J, Riviello J, Sannagowdara K, Sheehan T, Sands TT, Tasker RC, Tchapyjnikov D, Topjian AA, Vasquez A,

Pediatric Neurology Faculty Publications 2014-2025

- Wainwright MS**, Wilfong AA, Williams K, Loddenkemper T; pSERG. Time to Treatment in Pediatric Convulsive Refractory Status Epilepticus: The Weekend Effect. *Pediatr Neurol*. 2021 Mar 26;120:71-79
- Mingbunjerdasuk D, Blume H**, Browd S, Samii A. Intraventricular Baclofen Following Deep Brain Stimulation in a Child with Refractory Status Dystonicus. *Mov Disord Clin Pract*. 2021 Mar 19;8(3):456-459
- Madurski C, Jarvis JM, Beers SR, Houtrow AJ, Wagner AK, Fabio A, Wang C, Smith CM, Doughty L, Janesko-Feldman K, Rubin P, Pollon D, Treble-Barna A, Kochanek PM, Fink EL; PICU-Rehabilitation Study Group (**Mark Wainwright**). Serum Biomarkers of Regeneration and Plasticity are Associated with Functional Outcome in Pediatric Neurocritical Illness: An Exploratory Study *Neurocrit Care*. 2021 Mar 4.
- Kamins J, Richards R, Barney BJ, Locandro C, Pacchia CF, Charles AC, Cook LJ, Gioia G, Giza CC, **Blume HK**. Evaluation of Posttraumatic Headache Phenotype and Recovery Time After Youth Concussion. *JAMA Netw Open*. 2021 Mar 1;4(3):e211312. doi: 10.1001/jamanetworkopen.2021.1312.
- Gust J**, Annesley CE, Gardner RA, **Bozarth X**. EEG Correlates of Delirium in Children and Young Adults With CD19-Directed CAR T Cell Treatment-Related Neurotoxicity. *J Clin Neurophysiol*. 2021 Mar 1;38(2):135-142.
- Payal B Patel**, Andrew Belden, Ryan Handoko, Thanyawee Puthanakit, Stephen Kerr, Pope Kosalaraksa, Pradthana Ounchanum, Suparat Kanjanavanit, Linda Aupibul, Chaiwat Ngampiyasakul, Wicharn Luesomboon, Claude A Mellins, Kathleen Malee, Jintanat Ananworanich, Robert Paul. Behavioral impairment and cognition in Thai adolescents affected by HIV. *Glob Ment Health (Camb)*. 2021 Feb 9;8:e3. doi: 10.1017/gmh.2021.1. eCollection 2021.
- Harini Sarva, Gustavo Patino, Mehmood Rashid, **James WM Owens**, Matthew Robbins, and Stefano Sandrone (2021) The status of neurology fellowships in the United States: clinical needs, educational barriers, and future outlooks. *BMC Medical Education*. 2021 Feb 17: 21(1)108-117.
- Mingbunjerdasuk D**, Wong M, **Bozarth X**, Sun A. Co-occurrence of Metachromatic Leukodystrophy in Phelan-McDermid Syndrome. *J Child Neurol*. 2021 Feb;36(2):148-151.
- Peter Ferrazzano, Benjamin Yeske, Jeanette Mumford, Gregory Kirk, Erin D Bigler, **Katherine Bowen**, Nicole O'Brien, Bedda Rosario, Sue R Beers, Paul Rathouz, Michael J Bell, Andrew L Alexander, and the ADAPT MRI Biomarkers Investigators. Brain Magnetic Resonance Imaging Volumetric Measures of Functional Outcome after Severe Traumatic Brain Injury in Adolescents. *J Neurotrauma* 2021 Feb 24
- Patel PB**, Belden A, Handoko R, Puthanakit T, Kerr S, Kosalaraksa P, Ounchanum P, Kanjanavanit S, Aupibul L, Ngampiyasakul C, Luesomboon W, Mellins CA, Malee K, Ananworanich J, Paul R. Behavioral impairment and cognition in Thai adolescents affected by HIV. *Glob Ment Health (Camb)*. 2021 Feb 9;8:e3. doi: 10.1017/gmh.2021.1. PMID: 34026234; PMCID: PMC8127634.
- Amlie-Lefond C, Wainwright MS**. Childhood Stroke: Thinking Locally, Acting Globally? *Stroke*. 2021 Jan;52(1):162-163.
- Murphy OC, Messacar K, Benson L, Bove R, Carpenter JL, Crawford T, Dean J, DeBiasi R, Desai J, Elrick MJ, Farias-Moeller R, Gombolay GY, Greenberg B, Harmelink M, Hong S, Hopkins SE, Oleszek J, **Otten C**, Sadowsky CL, Schreiner TL, Thakur KT, Van Haren K, Carballo CM, Chong PF, Fall A, Gowda VK, Helfferich J, Kira R, Lim M, Lopez EL, Wells EM, Yeh EA, Pardo CA; AFM working group. Acute flaccid myelitis: cause, diagnosis, and management *Lancet*. 2021 Jan 23;397(10271):334-346.
- Acsadi G, Crawford TO, Müller-Felber W, Shieh PB, Richardson R, **Natarajan N**, Castro D, Ramirez-Schrempp D, Gambino G, Sun P, Farwell W. Safety and efficacy of nusinersen in spinal muscular atrophy: the EMBRACE study. *Muscle Nerve*. 2021 Jan 26.
- Crow YJ, Marshall H, Rice GI, Seabra L, Jenkinson EM, Baranano K, Battini R, Berger A, Blair E, Blauwblomme T, Bolduc F, Boddaert N, Buckard J, Burnett H, Calvert S, Caumes R, Ng AC, Chiang D, Clifford DB, Cordelli DM, de Burca A, Demic N, Desguerre I, De Waele L, Di Fonzo A, Dunham SR, Dyack S, Elmslie F, Ferrand M, Fisher G, Karimiani EG, Ghomid J, Gibbon F, Goel H, Hilmarson HT, Hughes I, Jacob A, Jones EA, Kumar R, Leventer RJ, MacDonald S, Maroofian R, Mehta SG, Metz I, Monfrini E, Neumann D, Noetzel M, O'Driscoll M, Öunap K, Panzer A, Parikh S, Prabhakar P, Ramond F, Sandford R, **Saneto R**, Soh C, Stutterd CA, Subramanian GM, Talbot K, Thomas RH, Toro C, Touraine R, Wakeling E, Wassmer E, Whitney A, Livingston JH, O'Keefe RT, Badrock AP. Leukoencephalopathy with

Pediatric Neurology Faculty Publications 2014-2025

calcifications and cysts: Genetic and phenotypic spectrum. Am J Med Genet A. 2021 Jan;185(1):15-25. doi: 10.1002/ajmg.a.61907. Epub 2020 Oct 7.

Malbari F, Partap S, **Gust J**, Duke E, **Sato A**, Khakoo Y, Ullrich NJ. Neuro-Oncology Training for the Child Neurology Resident. J Child Neurol. 2021 Jan;36(1):79-82

Weaver JJ, Hallam DK, Chick JFB, Vaidya S, Shin DS, **Natarajan N**, Rad N, Reis J, Koo KSH, Shivaram GM, Thibodeau A, Apkon S, Monroe EJ. Transforaminal intrathecal delivery of nusinersen for older children and adults with spinal muscular atrophy and complex spinal anatomy: an analysis of 200 consecutive injections. J Neurointerv Surg. 2021 Jan;13(1):75-78.

Grinspan ZM, Patel AD, Shellhaas RA, Berg AT, Axeen ET, Bolton J, Clarke DF, Coryell J, Gaillard WD, Goodkin HP, Koh S, Kukla A, Mbwana JS, **Morgan LA**, Singhal NS, Storey MM, Yozawitz EG, Abend NS, Fitzgerald MP, Fridinger SE, Helbig I, Massey SL, Prelack MS, Buchhalter J; Pediatric Epilepsy Learning Healthcare System. Design and implementation of electronic health record common data elements for pediatric epilepsy: Foundations for a learning health care system. Epilepsia. 2021 Jan;62(1):198-216.

Ko PY, Glass IA, Crandall S, Weiss A, Dorschner MO, Kelly JP, Phillips JO, **Lopez J**. Two Missense CACNA1A Variants in a Single Family with Variable Neurobehavioral, Cerebellar, Epileptic, and Oculomotor Features. Neuropediatrics. 2021 Jan 14.

Coughlin CR 2nd, Tseng LA, Abdenur JE, Ashmore C, Boemer F, Bok LA, Boyer M, Buhas D, Clayton PT, Das A, Dekker H, Evangelidou A, Feillet F, Footitt EJ, **Gospe SM Jr**, Hartmann H, Kara M, Kristensen E, Lee J, Lilje R, Longo N, Lunsing RJ, Mills P, Papadopoulou MT, Pearl PL, Piazzon F, Plecko B, Saini AG, Santra S, Sjarif DR, Stockler-Ipsiroglu S, Striano P, Van Hove JLK, Verhoeven-Duif NM, Wijburg FA, Zuberi SM, van Karnebeek CDM. Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to α -aminoacidic semialdehyde dehydrogenase deficiency. J Inher Metab Dis. 2021 Jan;44(1):178-192.

Dixon SM, Binkley MM, **Gospe SM Jr**, Guerriero RM. Child Neurology Applicants Place Increasing Emphasis on Quality of Life Factors. Pediatr Neurol. 2021 Jan;114:42-46.

2020

Lai YC, Muscal E, Wells E, Shukla N, Eschbach K, Hyeong Lee K, Kaliakatsos M, Desai N, Wickström R, Viri M, Freri E, Granata T, Nangia S, Dilella R, Brunklaus A, **Wainwright MS**, Gorman MP, Stredny CM, Asiri A, Hundallah K, Doja A, Payne E, Wirrell E, Koh S, Carpenter JL, Riviello. Anakinra usage in febrile infection related epilepsy syndrome: an international cohort. J. Ann Clin Transl Neurol. 2020 Dec;7(12):2467-2474

Cronin M, Wainwright MS. Plasma Exchange for Treatment of Refractory Demyelination. Pediatr Neurol Briefs. 2020 Dec 9;34:16.

Bass DI, **Shurtleff H, Warner M**, Knott D, Poliakov A, Friedman S, Collins MJ, **Lopez J, Lockrow JP, Novotny EJ**, Ojemann JG, Hauptman JS. Awake Mapping of the Auditory Cortex during Tumor Resection in an Aspiring Musical Performer: A Case Report. Pediatr Neurosurg. 2020;Dec 1 155(6):351-358

Gust J, Ponce R, Liles WC, Garden GA, Turtle CJ. Cytokines in CAR T Cell-Associated Neurotoxicity. Front Immunol. 2020 Dec 16;11:577027.

Weisleder P, **Gospe SM Jr** Withholding Childhood Immunizations: A Parent's Right or a Child's Neglect? Pediatr Neurol. 2020 Dec;113:80-81.

Oesch G, **Bozarth XL**. Rufinamide efficacy and association with phenotype and genotype in children with intractable epilepsy: A retrospective single center study. Epilepsy Res. 2020 Dec;168:106211.

Vasquez A, Gaínza-Lein M, Abend NS, Amengual-Gual M, Anderson A, Arya R, Brenton JN, Carpenter JL, Chapman K, Clark J, Farias-Moeller R, Gaillard WD, Glauser T, Goldstein JL, Goodkin HP, Guerriero RM, Kapur K, Lai YC, McDonough TL, Mikati MA, **Morgan LA, Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Riviello JJ, Sannagowdara K, Tasker RC, Tchapyjnikov D, Topjian A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T;

Pediatric Neurology Faculty Publications 2014-2025

Pediatric Status Epilepticus Research Group (pSERG). First-line medication dosing in pediatric refractory status epilepticus. *Neurology*. 2020 Nov 10;95(19):e2683-e2696

Park KB, Chapman T, Aldinger KA, Mirzaa GM, Zeiger J, Beck A, Glass IA, Hevner RF, Jansen AC, Marshall DA, Oegema R, Parrini E, **Saneto RP**, Curry CJ, Hall JG, Guerrini R, Leventer RJ, Dobyns WB. The spectrum of brain malformations and disruptions in twins. *Am J Med Genet A*. 2020 Nov 18. Online ahead of print. PMID: 33205886

Powell WT, Schaaf CP, Rech ME, **Wrede J**. Polysomnographic characteristics and sleep-disordered breathing in Schaaf-Yang syndrome. *Pediatr Pulmonol*. 2020 Nov;55(11):3162-3167.

Jeon TY, Poliakov AV, Friedman SD, **Bozarth XL**, **Novotny EJ**, Hauptman JS, Moon SH, Shaw DWW. Structural MRI and tract-based spatial statistical analysis of diffusion tensor imaging in children with hemimegalencephaly. *Neuroradiology*. 2020 Nov;62(11):1467-1474.

Lewis H, Samanta D, Örsell JL, Bosanko KA, Rowell A, Jones M, Dale RC, Taravath S, Hahn CD, Krishnakumar D, Chagnon S, Keller S, Hagebeuk E, Pathak S, Bebin EM, Arndt DH, **Alexander JJ**, Mainali G, Coppola G, Maclean J, Sparagana S, McNamara N, Smith DM, Raggio V, Cruz M, Fernández-Jaén A, Kava MP, Emrick L, Fish JL, Vanderver A, Helman G, Pierson TM, Zarate YA. Epilepsy and Electroencephalographic Abnormalities in SATB2-Associated Syndrome. *Pediatr Neurol*. 2020 Nov;112:94-100.

Ferdinandusse S, McWalter K, Te Brinke H, IJlst L, Mooijer PM, Ruiter JPN, van Lint AEM, Pras-Raves M, Wever E, Millan F, Guillen Sacoto MJ, Begtrup A, Tarnopolsky M, Brady L, Ladda RL, Sell SL, Nowak CB, Douglas J, Tian C, Ulm E, **Perlman S**, Drack AV, Chong K, Martin N, Brault J, Brokamp E, Toro C, Gahl WA, Macnamara EF, Wolfe L; Undiagnosed Diseases Network, Waisfisz Q, Zwijsen P, Ziegler A, Barth M, Smith R, Ellingwood S, Gaebler-Spira D, Bakhtiari S, Kruer MC, van Kampen AHC, Wanders RJA, Waterham HR, Cassiman D, Vaz FM. An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. *Genet Med*. 2020 Nov 26. Online ahead of print. PMID: 33239752

Cronin M, **Wainwright MS**. Underpowered and Too Heterogenous: A Humbling Assessment of the Literature Supporting Neuroprognostication After Pediatric Cardiac Arrest. *Pediatr Crit Care Med*. 2020 Oct;21(10):915-916.

Weisenberg JLZ, Fitzgerald RT, Constantino JN, Winawer MR, Thio LL; EPGP Investigators (**Novotny EJ**). Familial aggregation of status epilepticus in generalized and focal epilepsies. *Neurology*. 2020 Oct 13;95(15):e2140-e2149.

Boos MD, **Bozarth XL**, Sidbury R, Cooper AB, Perez F, Chon C, Paras G, **Amlie-Lefond C**. Forehead location and large segmental pattern of facial port-wine stains predict risk of Sturge-Weber syndrome. *J Am Acad Dermatol*. 2020 Oct;83(4):1110-1117.

Ebrahimi-Fakhari D, Teinert J, Behne R, Wimmer M, D'Amore A, Eberhardt K, Brechmann B, Ziegler M, Jensen DM, Nagabhyrava P, Geisel G, Carmody E, Shamshad U, Dies KA, Yuskaitis CJ, Salussolia CL, Ebrahimi-Fakhari D, Pearson TS, Saffari A, Ziegler A, Kölker S, Volkmann J, Wiesener A, Bearden DR, Lakhani S, Segal D, Udwadia-Hegde A, Martinuzzi A, Hirst J, **Perlman S**, Takiyama Y, Xiomerisiou G, Vill K, Walker WO, Shukla A, Dubey Gupta R, Dahl N, Aksoy A, Verhelst H, Delgado MR, Kremlikova Pourouva R, Sadek AA, Elkhateeb NM, Blumkin L, Brea-Fernández AJ, Dacruz-Álvarez D, Smol T, Ghomid J, Miguel D, Heine C, Schlump JU, Langen H, Baets J, Bulk S, Darvish H, Bakhtiari S, Kruer MC, Lim-Melia E, Aydinli N, Alanay Y, El-Rashidy O, Nampoothiri S, Patel C, Beetz C, Bauer P, Yoon G, Guillot M, Miller SP, Bourinaris T, Houlden H, Robelin L, Anheim M, Alamri AS, Mahmoud AAH, Inaloo S, Habibzadeh P, Faghihi MA, Jansen AC, Brock S, Roubertie A, Darras BT, Agrawal PB, Santorelli FM, Gleeson J, Zaki MS, Sheikh SI, Bennett JT, Sahin M. Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. *Brain*. 2020 Oct 1;143(10):2929-2944.

Sun LR, Harrar D, Drocton G, **Castillo-Pinto C**, Felling R, Carpenter JL, Wernovsky G, McDougall CG, Gailloud P, Pearl MS. Mechanical Thrombectomy for Acute Ischemic Stroke: Considerations in Children. *Stroke*. 2020 Oct;51(10):3174-3181. doi: 10.1161/STROKEAHA.120.029698. Epub 2020 Sep 11. PMID: 32912096

Kim SH, Green-Snyder L, Lord C, Bishop S, **Steinman KJ**, Bernier R, Hanson E, Goin-Kochel RP, Chung WK. Language characterization in 16p11.2 deletion and duplication syndromes. *Am J Med Genet B Neuropsychiatr Genet*. 2020 Sep;183(6):380-391.

Saneto RP. Mitochondrial diseases: expanding the diagnosis in the era of genetic testing. *J Transl Genet Genom*. 2020;4:384-428. Epub 2020 Sep 29. PMID: 33426505

Pediatric Neurology Faculty Publications 2014-2025

Vossler DG, Bainbridge JL, Boggs JG, **Novotny EJ**, Loddenkemper T, Faught E, Amengual-Gual M, Fischer SN, Gloss DS, Olson DM, Towne AR, Naritoku D, Welty TE. Treatment of Refractory Convulsive Status Epilepticus: A Comprehensive Review by the American Epilepsy Society Treatments Committee. *Epilepsy Curr.* 2020 Sep;20(5):245-264.

Bornstein R, Gonzalez B, **Johnson SC**. Mitochondrial pathways in human health and aging. *Mitochondrion.* 2020 Sep 54:72-84. DOI: 10.1016/j.mito.2020.07.007. Epub 2020 Jul 30. PMID: 32738358.

Law EF, **Blume H**, Palermo TM. Longitudinal Impact of Parent Factors in Adolescents With Migraine and Tension-Type Headache. *Headache.* 2020 Sep;60(8):1722-1733.

Pearl PL, Mink JW, Cohen BH, Bamford N, Bass N, Jordan L, **Wainwright MS**, Larson RB. The President, Past President, Executive Director, and the Board of the Child Neurology Society Denounce Racism and Inequality. *Ann Neurol.* 2020 Aug;88(2):209-210.

Patrick KE, Schultheis MT, Agate FT, McCurdy MD, Daly BP, Tarazi RA, Chute DL, Hurewitz F. Executive function "drives" differences in simulated driving performance between young adults with and without autism spectrum disorder. *Child Neuropsychol.* 2020 Jul;26(5):649-665.

Simpson TS, Peterson RL, **Patrick KE**, Forster JE, McNally KA. J Concussion Symptom Treatment and Education Program: A Feasibility Study. *Head Trauma Rehabil.* 2020 Jul 22.

Fernández IS, Abend NS, Amengual-Gual M, Anderson A, Arya R, Aguilar CB, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Gaillard WD, Gaínza-Lein M, Glauser T, Goldstein J, Goodkin HP, Guerriero RM, Lai YC, McDonough T, Mikati MA, **Morgan LA**, **Novotny E Jr**, Payne E, Peariso K, Piantino J, Ostendorf A, Sands TT, Sannagowdara K, Tasker RC, Tchapyjnikov D, Topjian AA, Vasquez A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; pSERG. Association of guideline publication and delays to treatment in pediatric status epilepticus. *Neurology.* 2020 Jul 1

Crotty EE, Leary SES, Geyer JR, Olson JM, Millard NE, **Sato AA**, Ermoian RP, Cole BL, Lockwood CM, Paulson VA, Browd SR, Ellenbogen RG, Hauptman JS, Lee A, Ojemann JG, Vitanza NA. Children with DIPG and high-grade glioma treated with temozolomide, irinotecan, and bevacizumab: the Seattle Children's Hospital experience. *J Neurooncol.* 2020 Jul;148(3):607-617.

Nambot S, Faivre L, Mirzaa G, Thevenon J, Bruel AL, Mosca-Boidron AL, Masurel-Paulet A, Goldenberg A, Le Meur N, Charollais A, Mignot C, Petit F, Rossi M, Metreau J, Layet V, Amram D, Boute-Bénéjean O, Bhoj E, Cousin MA, Kruisselbrink TM, Lanpher BC, Klee EW, Fiala E, Grange DK, Meschino WS, Hiatt SM, Cooper GM, Oivié H, Smith WE, Dumas M, Lehman A; CAUSES Study, Inglese C, Nizon M, Guerrini R, Vetro A, Kaplan ES, Miramar D, Van Gils J, Fergelot P, Bodamer O, Herkert JC, Pajusalu S, Öunap K, Filiano JJ, Smol T, Piton A, Gérard B, Chantot-Bastaraud S, Bienvenu T, Li D, Juusola J, Devriendt K, Bilan F, Poé C, Chevarin M, Jouan T, Tisserant E, Rivière JB, Tran Mau-Them F, Philippe C, Duffourd Y, **Dobyns WB**, Hevner R, Thauvin-Robinet C. De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *Eur J Hum Genet.* 2020 Jun;28(6):770-782.

Brock S, Vanderhasselt T, Vermaning S, Keymolen K, Régál L, Romaniello R, Wiczorek D, Storm TM, Schaeferhoff K, Hehr U, Kuechler A, Krägeloh-Mann I, Haack TB, Kasteleijn E, Schot R, Mancini GMS, Webster R, Mohammad S, Leventer RJ, Mirzaa G, **Dobyns WB**, Bahi-Buisson N, Meuwissen M, Jansen AC, Stouffs K. Defining the phenotypical spectrum associated with variants in *TUBB2A*. *J Med Genet.* 2020 Jun 22:jmedgenet-2019-106740

Benedetti GM, Vartanian RJ, McCaffery H, Shellhaas RA. Early Electroencephalogram Background Could Guide Tailored Duration of Monitoring for Neonatal Encephalopathy Treated with Therapeutic Hypothermia. *J Pediatr.* 2020 Jun;211:81-87.e1

Marashly A, Koop J, Loman M, Kim I, Maheshwari M, Lew SM. Multiple hippocampal transections for refractory pediatric mesial temporal lobe epilepsy: seizure and neuropsychological outcomes. *J Neurosurg Pediatr.* 2020 Jun 26:1-10.

Pediatric Neurology Faculty Publications 2014-2025

Rafay MF, Shapiro KA, Surmava AM, deVeber GA, Kirton A, Fullerton HJ, **Amlie-Lefond C**, Weschke B, Dlamini N, Carpenter JL, Mackay MT, Rivkin M, Linds A, Bernard TJ; International Pediatric Stroke Study (IPSS) Group. Spectrum of cerebral arteriopathies in children with arterial ischemic stroke. *Neurology*. 2020 Jun 9;94(23):e2479-e2490

Holland EL, **Saneto RP**, Knipper EK. Hypermetabolic Syndrome and Dyskinesia After Neurologic Surgery for Labrune Syndrome: A Case Report. *A A Pract*. 2020 May;14(7):e01212..

Lennox AL, Hoyer ML, Jiang R, Johnson-Kerner BL, Suit LA, Venkataramanan S, Sheehan CJ, Alsina FC, Fregeau B, Aldinger KA, Moey C, Lobach I, Afejar A, Babovic-Vuksanovic D, Bézieau S, Blackburn PR, Bunt J, Burglen L, Campeau PM, Charles P, Chung BHY, Cogné B, Curry C, D'Agostino MD, Di Donato N, Faivre L, Héron D, Innes AM, Isidor B, Keren B, Kimball A, Klee EW, Kuentz P, Küry S, Martin-Coignard D, Mirzaa G, Mignot C, Miyake N, Matsumoto N, Fujita A, Nava C, Nizon M, Rodriguez D, Blok LS, Thauvin-Robinet C, Thevenon J, Vincent M, Ziegler A, **Dobyns W**, Richards LJ, Barkovich AJ, Floor SN, Silver DL, Sherr EH. Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*. 2020 May 6;106(3):404-420.e8

Weaver JJ, Hallam DK, Chick JFB, Vaidya S, Shin DS, **Natarajan N**, Rad N, Reis J, Koo KSH, Shivaram GM, Thibodeau A, Apkon S, Monroe EJ. Transforaminal intrathecal delivery of nusinersen for older children and adults with spinal muscular atrophy and complex spinal anatomy: an analysis of 200 consecutive injections. *J Neurointerv Surg*. 2020 May

Blumkin L, Leibovitz Z, Kraiden-Haratz K, Arad A, Yosovich K, Gindes L, Zerem A, Ben-Sira L, Lev D, Nissenkorn A, Kidron D, **Dobyns WB**, Malinger G, Bahi-Buisson N, Leventer RJ, Lerman-Sagie T. Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features *Eur J Paediatr Neurol*. 2020 May;26:46-60.

Miller I, Scheffer IE, Gunning B, Sanchez-Carpintero R, Gil-Nagel A, Perry MS, **Saneto RP**, Checketts D, Dunayevich E, Knappertz V; GWPCARE2 Study Group. Dose-Ranging Effect of Adjunctive Oral Cannabidiol vs Placebo on Convulsive Seizure Frequency in Dravet Syndrome: A Randomized Clinical Trial. *JAMA Neurol*. 2020 May 1;77(5):613-621.

Tsai MH, Muir AM, Wang WJ, Kang YN, Yang KC, Chao NH, Wu MF, Chang YC, Porter BE, Jansen LA, Sebire G, Deconinck N, Fan WL, Su SC, Chung WH, Almanza Fuerte EP, Mehaffey MG; University of Washington Center for Mendelian Genomics, Ng CC, Chan CK, Lim KS, Leventer RJ, Lockhart PJ, Riney K, Damiano JA, Hildebrand MS, Mirzaa GM, **Dobyns WB**, Berkovic SF, Scheffer IE, Tsai JW, Mefford HC. Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron*. 2020 Apr 22;106(2):237-245.e8.

Sandrone S, Berthaud JV, Carlson C, Cios J, Dixit N, Farheen A, Kraker J, **Owens JWM**, Patino G, Sarva H, Weber D, Schneider LD. Active Learning in Psychiatry Education: Current Practices and Future Perspectives. *Front Psychiatry*. 2020 Apr 23;11:211.

Nambot S, Hevner RF, **Dobyns WB**. Reply to Hsueh YP et al. *Eur J Hum Genet*. 2020 Apr 9.

Mineyko A, Kirton A, Billingham L, Tatishvili NN, Wintermark M, deVeber G, Fox C; SIPS Investigators. Kirton A, Abdalla A, Zafeiriou D, Friedman N, Aprasidze T, Kolk A, Armstrong J, Ichord R, **Amlie-Lefond C**, deVeber G, Kovacevic G, Hernandez Chavez M, Mackay M, Titomanlio L, Williams K, Elbers J, Fullerton H, Benedict S, Dowling M, Jordan L, Pergami P. Seizures and Outcome One Year After Neonatal and Childhood Cerebral Sinovenous Thrombosis. *Pediatr Neurol*. 2020 Apr;105:21-26.

Johnson SC, Kayser EB, Bornstein R, Stokes J, Bitto A, Park KY, Pan A, Sun G, Raftery D, Kaerberlein M, Sedensky MM, Morgan PG. Regional metabolic signatures in the Ndufs4(KO) mouse brain implicate defective glutamate/ α -ketoglutarate metabolism in mitochondrial disease. *Molecular Genetics and Metabolism*. 2020 Jun;130(2):118-132. doi: 10.1016/j.ymgme.2020.03.007. Epub 2020 Apr 3. PMID: 32331968 27.

Perez FA, Oesch G, **Amlie-Lefond CM** MRI Vessel Wall Enhancement and Other Imaging Biomarkers in Pediatric Focal Cerebral Arteriopathy-Inflammatory Subtype. *Stroke*. 2020 Mar;51(3):853-859.

Friederich MW, Perez FA, Knight KM, Van Hove RA, Yang SP, **Saneto RP**, Van Hove JKL. Pathogenic variants in NUBPL result in failure to assemble the matrix arm of complex I and cause a complex leukoencephalopathy with thalamic involvement. *Mol Genet Metab*. 2020 Mar;129(3):236-242.

Gonzalez-Perez P, Smith C, Sebetka WL, Gedlinske A, **Perlman S**, Mathews KD. Clinical and electrophysiological evaluation of myasthenic features in an alpha-dystroglycanopathy cohort (FKRP-predominant). *Neuromuscul Disord*. 2020 Mar;30(3):213-218.

Pediatric Neurology Faculty Publications 2014-2025

Barca E, Long Y, Cooley V, Schoenaker R, Emmanuele V, DiMauro S, Cohen BH, Karaa A, Vladutiu GD, Haas R, Van Hove JKL, Scaglia F, Parikh S, Bedoyan JK, DeBrosse SD, Gavrilova RH, **Saneto RP**, Enns GM, Stacpoole PW, Ganesh J, Larson A, Zolkipli-Cunningham Z, Falk MJ, Goldstein AC, Tarnopolsky M, Gropman A, Camp K, Krotoski D, Engelstad K, Rosales XQ, Kriger J, Grier J, Buchsbaum R, Thompson JLP, Hirano M. Mitochondrial diseases in North America: An analysis of the NAMDC Registry. *Neurol Genet.* 2020 Mar 2;6(2):e402.. eCollection 2020 Apr

Vabres P, Sorlin A, Kholmanskikh SS, Demeer B, St-Onge J, Duffourd Y, Kuentz P, Courcet JB, Carmignac V, Garret P, Bessis D, Boute O, Bron A, Captier G, Carmi E, Devauchelle B, Geneviève D, Gondry-Jouet C, Guibaud L, Lafon A, Mathieu-Dramard M, Thevenon J, **Dobyns WB**, Bernard G, Polubothu S, Faravelli F, Kinsler VA, Thauvin C, Faivre L, Ross ME, Rivière JB. Author Correction: Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. *Nat Genet.* 2020 Mar;52(3):353.

Lennox AL, Hoye ML, Jiang R, Johnson-Kerner BL, Suit LA, Venkataramanan S, Sheehan CJ, Alsina FC, Fregeau B, Vabres P, Sorlin A, Kholmanskikh SS, Demeer B, St-Onge J, Duffourd Y, Kuentz P, Courcet JB, Carmignac V, Garret P, Bessis D, Boute O, Bron A, Captier G, Carmi E, Devauchelle B, Geneviève D, Gondry-Jouet C, Guibaud L, Lafon A, Mathieu-Dramard M, Thevenon J, **Dobyns WB**, Bernard G, Polubothu S, Faravelli F, Kinsler VA, Thauvin C, Faivre L, Ross ME, Rivière JB. Author Correction: Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. *Nat Genet.* 2020 Mar;52(3):353. doi: 10.1038/s41588-019-0565-x.

Blumkin L, Leibovitz Z, Kraiden-Haratz K, Arad A, Yosovich K, Gindes L, Zerem A, Ben-Sira L, Lev D, Nissenkorn A, Kidron D, **Dobyns WB**, Malinger G, Bahi-Buisson N, Leventer RJ, Lerman-Sagie T. Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. *Eur J Paediatr Neurol.* 2020 Mar 4. pii: S1090-3798(20)30044-1.

DelRosso LM, Yi T, **Chan JHM**, **Wrede JE**, **Lockhart CT**, Ferri R. Determinants of ferritin response to oral iron supplementation in children with sleep movement disorders. *Sleep.* 2020 Mar 12;43(3). pii: zsz234..

Neighbors A, Moss T, Holloway L, Yu SH, Annese F, Skinner S, **Saneto R**, Steet R. Functional analysis of a novel mutation in the TIMM8A gene that causes deafness-dystonia-optic neuropathy syndrome. *Mol Genet Genomic Med.* 2020 Mar;8(3):e1121.

Aldinger KA, Moey C, Lobach I, Afenjar A, Babovic-Vuksanovic D, Bézieau S, Blackburn PR, Bunt J, Burglen L, Campeau PM, Charles P, Chung BHY, Cogné B, Curry C, D'Agostino MD, Di Donato N, Faivre L, Héron D, Innes AM, Isidor B, Keren B, Kimball A, Klee EW, Kuentz P, Küry S, Martin-Coignard D, Mirzaa G, Mignot C, Miyake N, Matsumoto N, Fujita A, Nava C, Nizon M, Rodriguez D, Blok LS, Thauvin-Robinet C, Thevenon J, Vincent M, Ziegler A, **Dobyns W**, Richards LJ, Barkovich AJ, Floor SN, Silver DL, Sherr EH. Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron.* 2020 Feb 28.

Delrosso LM, **Lockhart C**, **Wrede JE**, Chen ML, Samson M, Reed J, Martin-Washo S, Arp M, Ferri R. Comorbidities in children with elevated periodic limb movement index during sleep. *Sleep.* 2020 Feb 13;43(2).

Amlie-Lefond C, Shaw DWW, Cooper A, **Wainwright MS**, Kirton A, Felling RJ, Abraham MG, Mackay MT, Dowling MM, Torres M, Rivkin MJ, Grabowski EF, Lee S, Kurz JE, McMillan HJ, Barry D, **Lee-Eng J**, Ichord RN. Risk of Intracranial Hemorrhage Following Intravenous tPA (Tissue-Type Plasminogen Activator) for Acute Stroke Is Low in Children. *Stroke.* 2020 Feb;51(2):542-548.

Amlie-Lefond C, **Wainwright MS**. Response by Amlie-Lefond and Wainwright to Letter Regarding Article, "Organizing for Acute Arterial Ischemic Stroke in Children". *Stroke.* 2020 Feb;51(2):e37.

Tsai MH, Muir AM, Wang WJ, Kang YN, Yang KC, Chao NH, Wu MF, Chang YC, Porter BE, Jansen LA, Sebire G, Deconinck N, Fan WL, Su SC, Chung WH, Almanza Fuerte EP, Mehaffey MG; University of Washington Center for Mendelian Genomics, Ng CC, Chan CK, Lim KS, Leventer RJ, Lockhart PJ, Riney K, Damiano JA, Hildebrand MS, Mirzaa GM, **Dobyns WB**, Berkovic SF, Scheffer IE, Tsai JW, Mefford HC. Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron.* 2020 Feb 10.

Press CA, Kirschen M, LaRovere K, Risen S, Williams KP, Chung M, Griffith J, Erklauer J, Peariso K, Ducharme-Crevier L, Shah SS, Hall M, **Wainwright MS**. Variation in Treatment and Outcomes of Children With Acute Disseminated Encephalomyelitis. *Hosp Pediatr.* 2020 Feb;10(2):159-165..

Pediatric Neurology Faculty Publications 2014-2025

Patel, P, Belden A, Handoko R, Paul, R, et al. Behavioral Impairment and Cognition in Thai Adolescents Affected by HIV. *Global Mental Health*. 9;8:e3. Feb 2020. PMID: 34026234

Nelson BR, Roby JA, **Dobyns WB**, Rajagopal L, Gale M Jr, Adams Waldorf KM. Immune Evasion Strategies Used by Zika Virus to Infect the Fetal Eye and Brain. *Viral Immunol*. 2020 Jan/Feb;33(1):22-37.

Marashly A, Koop J, Loman M, Lee YW, Lew SM. Examining the Utility of Resective Epilepsy Surgery in Children With Electrical Status Epilepticus in Sleep: Long Term Clinical and Electrophysiological Outcomes. *Front Neurol*. 2020 Jan 15;10:1397.

LaRovere KL, Tasker RC, **Wainwright M**, Reuter-Rice K, Appavu B, Miles D, Lidsky K, Vittner P, Gundersen D, O'Brien NF Transcranial Doppler Ultrasound During Critical Illness in Children: Survey of Practices in Pediatric Neurocritical Care Centers. *Pediatric Neurocritical Care Research Group (PNCRG)*. *Pediatr Crit Care Med*. 2020 Jan;21(1):67-74.

Wainwright MS. PIRSE: Philosophical Investigations in Refractory Status Epilepticus. *Pediatr Crit Care Med*. 2020 Jan;21(1):98-99.

Mak CCY, Doherty D, Lin AE, Vegas N, Cho MT, Viot G, Dimartino C, Weisfeld-Adams JD, Lessel D, Joss S, Li C, Gonzaga-Jauregui C, Zarate YA, Ehmke N, Horn D, Troyer C, Kant SG, Lee Y, Ishak GE, Leung G, Barone Pritchard A, Yang S, Bend EG, Filippini F, Roadhouse C, Lebrun N, Mehaffey MG, Martin PM, Apple B, Millan F, Puk O, Hoffer MJV, Henderson LB, McGowan R, Wentzensen IM, Pei S, Zahir FR, Yu M, Gibson WT, Seman A, Steeves M, Murrell JR, Luettgen S, Francisco E, Strom TM, Amlie-Wolf L, Kaindl AM, Wilson WG, Halbach S, Basel-Salmon L, Lev-El N, Denecke J, Vissers LELM, Radtke K, Chelly J, Zackai E, Friedman JM, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Reid RR, Devriendt K, Chae JH, Stolerman E, McDougall C, Powis Z, Bienvenu T, Tan TY, Orenstein N, **Dobyns WB**, Shieh JT, Choi M, Waggoner D, Gripp KW, Parker MJ, Stoler J, Lyonnet S, Cormier-Daire V, Viskochil D, Hoffman TL, Amiel J, Chung BHY, Gordon CT. MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis *Brain*. 2020 Jan 1;143(1):55-68.

Goeggel Simonetti B, Rafay MF, Chung M, Lo WD, Beslow LA, Billingham LL, Fox CK, Pagnamenta A, Steinlin M, Mackay MT; IPSS Study Group. Collaborators (98) Ashwal S, deVeber G, Ferriero D, Fullerton H, Ichord R, Kirkham F, Lynch JK, O'Callaghan F, Pavlakis S, Sebire G, Willan A, deVeber G, Dowling M, Mackay M, Hernandez Chavez M, Bernard T, Nowak-Gottl U, Benedict S, Rafay M, Yager J, Lo W, Parakh M, Kirton A, Ichord R, Steinlin M, Carpenter J, Rivkin M, Wiznitzer M, O'Callaghan F, Kan L, Zafeiriou D, Fullerton H, Bjornson B, Jordan L, Ashwal S, Torres M, Kolk A, Troncoso M, Whelan H, Pavlakis S, Chan A, Kopyta I, Abdalla A, Gerard T, Golomb M, Humphreys P, Noetzel M, Heyer G, **Amlie-Lefond C**, Yeh A, Pergami P, Cardona F, Kovacevic G, Ganesan V, Elbers J, Friedman N, Wong V, Tatishvili N, Sultan S, Abraham L, Gonzalez V, Titomanlio L, Saengpatrachai M, Lerner NB, Crosswell H, Oliviera C, Andrade Alveal L, Felling R, Hunfeld M, Altuna D, Zecavati N, Buckley D, Deray M, Hassanein S, Brankovic-Sreckovic V, Hegde A, Raybagkar D, Sebire G, Miteff C, Condie J, Khoury C, Carpenter S, Holzhauer S, Garcia Soler P, Nassif El-Hakam L, Conto L, Francisco Vilavedra J, Kosofsky B, Guang Y, Taylor JM, Goldenberg N, Toma A, Beslow LA, **Wainwright M**, Xavier F, Grabowski E, Nass R, Wilson J. Comparative study of posterior and anterior circulation stroke in childhood: Results from the International Pediatric Stroke Study. *Neurology*. 2020 Jan 28;94(4):e337-e344.

Nambot S, Faivre L, Mirzaa G, Thevenon J, Bruel AL, Mosca-Boidron AL, Masurel-Paulet A, Goldenberg A, Le Meur N, Charollais A, Mignot C, Petit F, Rossi M, Metreau J, Layet V, Amram D, Boute-Bénéjean O, Bhoj E, Cousin MA, Kruisselbrink TM, Lanpher BC, Klee EW, Fiala E, Grange DK, Meschino WS, Hiatt SM, Cooper GM, Olivié H, Smith WE, Dumas M, Lehman A; CAUSES Study, Inglese C, Nizon M, Guerrini R, Vetro A, Kaplan ES, Miramar D, Van Gils J, Fergelot P, Bodamer O, Herkert JC, Pajusalu S, Öunap K, Filiano JJ, Smol T, Piton A, Gérard B, Chantot-Bastaraud S, Bienvenu T, Li D, Juusola J, Devriendt K, Bilan F, Poé C, Chevarin M, Jouan T, Tisserant E, Rivière JB, Tran Mau-Them F, Philippe C, Duffourd Y, **Dobyns WB**, Hevner R, Thauvin-Robinet C. De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *Eur J Hum Genet*. 2020 Jan 31.

Monteiro FP, Curry CJ, Hevner R, Elliott S, Fisher JH, Turocy J, **Dobyns WB**, Costa LA, Freitas E, Kitajima JP, Kok F. Biallelic loss of function variants in ATP1A2 cause hydrops fetalis, microcephaly, arthrogryposis and extensive cortical malformations *Eur J Med Genet*. 2020 Jan;63(1):103624.

Pediatric Neurology Faculty Publications 2014-2025

Sandrone S, Berthaud JV, Carlson C, Cios J, Dixit N, Farheen A, Kraker J, **Owens JWM**, Patino G, Sarva H, Weber D, Schneider LD. Strategic Considerations for Applying the Flipped Classroom to Neurology Education. *Ann Neurol*. 2020 Jan;87(1):4-9.

2019

Kahn-Kirby AH, Amagata A, Maeder CI, Mei JJ, Sideris S, Kosaka Y, Hinman A, Malone SA, Bruegger JJ, Wang L, Kim V, Shrader WD, Hoff KG, Latham JC, Ashley EA, Wheeler MT, Bertini E, Carrozzo R, Martinelli D, Dionisi-Vici C, Chapman KA, Enns GM, Gahl W, Wolfe L, Saneto RP, **Johnson SC**, Trimmer JK, Klein MB, Holst CR. Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. *PLoS One*. 2019. 14(3):e0214250. PMID: 30921410.

Johnson SC, Pan A, Sun GX, Freed A, Stokes JC, Bornstein R, Witkowski M, Li L, Ford JM, Howard CRA, Sedensky MM, Morgan PG. Relevance of experimental paradigms of anesthesia induced neurotoxicity in the mouse. *PLoS One*. 2019. 14(3):e0213543. PMID: 30897103.

Griffiths KK, Morgan PG, **Johnson SC**, Nambyiah P, Soriano SG, Johnson K, Xu J, Garber C, Maxwell L, Saraiya N. A Summary of Preclinical Poster Presentations at the Sixth Biennial Pediatric Anesthesia Neurodevelopment Assessment (PANDA) Symposium. *J Neurosurg Anesthesiol*. 2019. 31(1):163-165. PMID: 30767942

Johnson, SC, Pan A, Li L, Sedensky M, Morgan P: Neurotoxicity of Anesthetics: Mechanisms and Meaning from Mouse Intervention Studies. *Neurotoxicology and Teratology*. 2019. 71:22-31. PMID: 30472095.

Johnson, SC, Martinez F, Bitto A, Gonzalez B, Tazaerslan C, Cohen C, Delaval L, Timsit J, Knebelmann B, Terzi F, Mahal T, Zhu Y, Morgan P, Sedensky M, Kaeberlein M, Legendre C, Suh Y, Canaud G: mTOR inhibitors may benefit kidney transplant recipients with mitochondrial diseases. *Kidney International*. 2019. 95(2):455-466. PMID: 30471880

Amlie-Lefond C, Wainwright IMS. Organizing for Acute Arterial Ischemic Stroke in Children. *Stroke*. 2019 Dec;50(12):3662-3668.

Gust J, Annesley CE, Gardner RA, **Bozarth X**. EEG Correlates of Delirium in Children and Young Adults With CD19-Directed CAR T Cell Treatment-Related Neurotoxicity. *J Clin Neurophysiol*. 2019 Dec 13.

Bozarth XL, McGuire J, **Novotny E**. Current Status of Continuous Electroencephalographic Monitoring in Critically Ill Children. *Pediatr Neurol*. 2019 Dec;101:11-17

Friederich MW, Perez FA, Knight KM, Van Hove RA, Yang SP, **Saneto RP**, Van Hove JLK. Pathogenic variants in NUBPL result in failure to assemble the matrix arm of complex I and cause a complex leukoencephalopathy with thalamic involvement. *Mol Genet Metab*. 2019 Dec 30.

Farhadian SF, Mistry H, Kirchwey T, Chiarella J, Calvi R, Chintanaphol M, **Patel P**, Landry ML, Robertson K, Spudich. Markers of CNS Injury in Adults Living With HIV With CSF HIV Not Detected vs Detected. *Open Forum Infect Dis*. 2019 Dec 14;6(12):ofz528.

RA, Ceppi F, Rivers J, Annesley C, Summers C, Taraseviciute A, **Gust J**, Leger KJ, Tarlock K, Cooper TM, Finney OC, Brakke H, Li DH, Park JR, Jensen MC. Preemptive mitigation of CD19 CAR T-cell cytokine release syndrome without attenuation of antileukemic efficacy. *Gardner Blood*. 2019 Dec 12;134(24):2149-2158. doi: 10.1182/blood.2019001463.

Ravanpay AC, **Gust J**, Johnson AJ, Rolczynski LS, Cecchini M, Chang CA, Høglund VJ, Mukherjee R, Vitanza NA, Orentas RJ, Jensen MC. EGFR806-CAR T cells selectively target a tumor-restricted EGFR epitope in glioblastoma. *Oncotarget*. 2019 Dec 17;10(66):7080-7095.

Bearer C, Agostoni C, Anand KJS, Ambalavanan N, Bhandari V, Bliss JM, Bloomfield F, Bonifacio SL, Buhimschi I, Cilio MR, Coppes M, Czinn SJ, El-Khuffash A, Embleton N, Felderhoff-Müser U, Ferriero DM, Florin T, Fuentes-Afflick E, Gardner W, **Gospe SM Jr**, Gunn A, Gressens P, Guissani D, Haiden N, Hauptman M, Kim KS, Klebanoff M, Lachman P, Lanphear B, Ozen S, Roehr C, Roland D, Rosenblum N, Schwarz M, Staiano A, Stroustrup A, Valente EM, Wilson-Costello D, Wynn J, Molloy E. Toward the elimination of bias in Pediatric Research. *Pediatr Res*. 2019 Dec;86(6):680-681.

Pediatric Neurology Faculty Publications 2014-2025

Gardner RA, Ceppi F, Rivers J, Annesley C, Summers C, Taraseviciute A, **Gust J**, Leger KJ, Tarlock K, Cooper TM, Finney OC, Brakke H, Li DH, Park JR, Jensen MC. Preemptive mitigation of CD19 CAR T-cell cytokine release syndrome without attenuation of antileukemic efficacy. *Blood*. 2019 Dec 12;134(24):2149-2158. doi: 10.1182/blood.2019001463

Wilson JL, **Amlie-Lefond C**, Abruzzo T, Orbach DB, Rivkin MJ, deVeber GA, Pergami P. Survey of practice patterns and preparedness for endovascular therapy in acute pediatric stroke. *Childs Nerv Syst*. 2019 Dec;35(12):2371-2378.

Parikh S, Galioto R, Lapin B, Haas R, Hirano M, Koenig MK, **Saneto RP**, Zolkipli-Cunningham Z, Goldstein A, Karaa A. Fatigue in primary genetic mitochondrial disease: No rest for the weary. *Neuromuscul Disord*. 2019 Nov;29(11):895-902.

Lockrow JP, Wright JN, **Saneto RP**, **Amlie-Lefond C**. Epileptic Spasms Predict Poor Epilepsy Outcomes After Perinatal Stroke. *J Child Neurol*. 2019 Nov;34(13):830-836.

Kochanek PM, Grenvik AN, Tasker RC, Carney N, Totten AM, Adelson PD, Selden NR, Davis-O'Reilly C, Hart EL, Bell MJ, Bratton SL, Grant GA, Kissoon N, Reuter-Rice KE, Vavilala MS, **Wainwright MS**. The authors reply. *Pediatr Crit Care Med*. 2019 Nov;20(11):1105-1107. .

Rummey C, Corben LA, Delatycki MB, Subramony SH, Bushara K, Gomez CM, Hoyle JC, Yoon G, Ravina B, Mathews KD, Wilmot G, Zesiewicz T, **Perlman S**, Farmer JM, Lynch DR. Psychometric properties of the Friedreich Ataxia Rating Scale. *Neurol Genet*. 2019 Oct 29;5(6):371

Wood AM, Geddes GC, **Marashly A**. Language regression, hemichorea and focal subclinical seizures in a 6-year-old girl with GLUT-1 deficiency. *Epilepsy Behav Rep*. 2019 Oct 24;14:100340.

Gust J, Ishak GE. Chimeric Antigen Receptor T-Cell Neurotoxicity Neuroimaging: More Than Meets the Eye. *AJNR Am J Neuroradiol*. 2019 Oct;40(10):E50-E51.

Oegema R, Barkovich AJ, Mancini GMS, Guerrini R, **Dobyns WB**. Subcortical heterotopic gray matter brain malformations: Classification study of 107 individuals. *Neurology*. 2019 Oct 1;93(14):e1360-e1373

Law EF, Powers SW, **Blume H**, Palermo TM. Screening Family and Psychosocial Risk in Pediatric Migraine and Tension-Type Headache: Validation of the Psychosocial Assessment Tool (PAT). *Headache*. 2019 Oct;59(9):1516-1529.

Chan JHM, **Owens JW**, **Wrede JE**. Case of an In-Laboratory Vagal Nerve Stimulator Titration for Vagal Nerve Stimulator-Induced Pediatric Obstructive Sleep Apnea. *J Clin Sleep Med*. 2019 Oct 15;15(10):1539-1542.

Patel PB, Apornpong T, Puthanakit T, Thongpibul K, Kosalaraksa P, Hansudewechakul R, Kanjanavanit S, Ngampiyaskul C, Luesomboon W, Wongsawat J, Sun LP, Chettra K, Saphonn V, Mellins CA, Malee K, Spudich S, Ananworanich J, Kerr SJ, Paul R; PREDICT/Resilience Study Trajectory Analysis of Cognitive Outcomes in Children With Perinatal HIV. Group. *Pediatr Infect Dis J*. 2019 Oct;38(10):1038-1044

DelRosso LM, **Lockhart C**, **Wrede JE**, Chen ML, Samson M, Reed J, Martin-Washo S, Arp M, Ferri R. Comorbidities in children with elevated periodic limb movement index during sleep. *Sleep*. 2019 Sep 26.

DelRosso LM, Yi T, Chan JHM, **Wrede JE**, **Lockhart CT**, Ferri R. Determinants of ferritin response to oral iron supplementation in children with sleep movement disorders. *Sleep*. 2019 Sep 29. pii: zsz234.

Dines JN, Liu YJ, Neufeld-Kaiser W, Sawyer T, Ishak GE, **Tully HM**, Racobaldo M, Sanchez-Valle A, Disteche CM, Juusola J, Torti E, McWalter K, Doherty D, Dipple KM. Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for FOXA2 in 20p11.2 deletion syndrome. *Am J Med Genet A*. 2019 Sep;179(9):1783-1790

Gripp KW, Morse LA, Axelrad M, Chatfield KC, Chidekel A, **Dobyns W**, Doyle D, Kerr B, Lin AE, Schwartz DD, Sibbles BJ, Siegel D, Shankar SP, Stevenson DA, Thacker MM, Weaver KN, White SM, Rauen KA. Costello syndrome: Clinical phenotype, genotype, and management guidelines. *Am J Med Genet A*. 2019 Sep;179(9):1725-1744.

Sansevere AJ, Arya R, Sánchez Fernández I, Gaillard WD, Tasker RC, Lai YC, Anderson AE, Tchapyjnikov D, Chapman KE, Brenton JN, Carpenter JL, Gaínza-Lein M, Goldstein JL, Goodkin HP, Jackson MC, Kapur K, Mikati MA, Peariso K, Glauser TA, Topjian AA, **Wainwright M**, Wilfong AA, Williams KL, Loddenkemper T, Abend NS; De Novo Variants in

Pediatric Neurology Faculty Publications 2014-2025

WDR37 Are Associated with Epilepsy Pediatric Status Epilepticus Research Group (pSERG). J Clin Neurophysiol. 2019 Sep;36(5):365-370.

Luat AF, Juhász C, Loeb JA, Chugani HT, Falchek SJ, Jain B, Greene-Roethke C, **Amlie-Lefond C**, Ball KL, Davis A, Pinto A. Neurological Complications of Sturge-Weber Syndrome: Current Status and Unmet Needs. Pediatr Neurol. 2019 Sep;98:31-38

Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC; Undiagnosed Diseases Network, Chung WK, Markello TC, **Dobyns WB**, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. Am J Hum Genet. 2019 Aug 1;105(2):413-424. doi: 10.1016/j.ajhg.2019.06.014. Epub 2019 Jul 18. Erratum in: Am J Hum Genet. 2019 Sep 5;105(3):672-674.

Berg AT, Wusthoff C, Shellhaas RA, Loddenkemper T, Grinspan ZM, **Saneto RP**, Knupp KG, Patel A, Sullivan JE, Kossoff EH, Chu CJ, Massey S, Valencia I, Keator C, Wirrell EC, Coryell J, Millichap JJ, Gaillard WD. Immediate outcomes in early life epilepsy: A contemporary account. Epilepsy Behav. 2019 Aug;97:44-50.

Topjian AA, de Caen A, **Wainwright MS**, Abella BS, Abend NS, Atkins DL, Bembea MM, Fink EL, Guerguerian AM, Haskell SE, Kilgannon JH, Lasa JJ, Hazinski MF. Pediatric Post-Cardiac Arrest Care: A Scientific Statement From the American Heart Association. 2019 Aug 6;140(6):e194-e233.

Patel PB. HIV-Associated Neurocognitive Disorders. Medlink Neurology. 2019 August;

Sánchez Fernández I, Gaínza-Lein M, Abend NS, Amengual-Gual M, Anderson A, Arya R, Brenton JN, Carpenter JL, Chapman KE, Clark J, Farias-Moeller R, Davis Gaillard W, Glauser TA, Goldstein J, Goodkin HP, Guerriero RM, Hecox K, Jackson M, Kapur K, Kelley SA, Kossoff EHW, Lai YC, McDonough TL, Mikati MA, **Morgan LA**, **Novotny EJ**, Ostendorf AP, Payne ET, Peariso K, Piantino J, Riviello JJ Jr, Sannagowdara K, Stafstrom CE, Tasker RC, Tchapyjnikov D, Topjian AA, Vasquez A, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; The onset of pediatric refractory status epilepticus is not distributed uniformly during the day. Pediatric Status Epilepticus Research Group. Seizure. 2019 Aug;70:90-96.

Holt RJ, Young RM, Crespo B, Ceroni F, Curry CJ, Bellacchio E, Bax DA, Ciolfi A, Simon M, Fagerberg CR, van Binsbergen E, De Luca A, Memo L, **Dobyns WB**, Mohammed AA, Clokie SJH, Zazo Seco C, Jiang YH, Sørensen KP, Andersen H, Sullivan J, Powis Z, Chassevent A, Smith-Hicks C, Petrovski S, Antoniadis T, Shashi V, Gelb BD, Wilson SW, Gerrelli D, Tartaglia M, Chassaing N, Calvas P, Ragge NK. De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. Am J Hum Genet. 2019 Jul 23. .

Ng BG, Sosicka P, Agadi S, Almannai M, Bacino CA, Barone R, Botto LD, Burton JE, Carlston C, Chung BH, Cohen JS, Coman D, Dipple KM, Dorrani N, **Dobyns WB**, Elias AF, Epstein L, Gahl WA, Garozzo D, Hammer TB, Haven J, Héron D, Herzog M, Hoganson GE, Hunter JM, Jain M, Juusola J, Lakhani S, Lee H, Lee J, Lewis K, Longo N, Lourenço CM, Mak CCY, McKnight D, Mendelsohn BA, Mignot C, Mirzaa G, Mitchell W, Muhle H, Nelson SF, Olczak M, Palmer CGS, Partikian A, Patterson MC, Pierson TM, Quinonez SC, Regan BM, Ross ME, Guillen Sacoto MJ, Scaglia F, Scheffer IE, Segal D, Singhal NS, Striano P, Sturiale L, Symonds JD, Tang S, Vilain E, Willis M, Wolfe LA, Yang H, Yano S, Powis Z, Suchy SF, Rosenfeld JA, Edmondson AC, Grunewald S, Freeze HH. SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Hum Mutat. 2019 Jul;40(7):908-925.

Gust J, Finney OC, Li D, Brakke HM, Hicks RM, Futrell RB, Gamble DN, Rawlings-Rhea SD, Khalatbari HK, Ishak GE, Duncan VE, Hevner RF, Jensen MC, Park JR, Gardner RA. Glial injury in neurotoxicity after pediatric CD19-directed chimeric antigen receptor T cell therapy. Ann Neurol. 2019 Jul;86(1):42-54

Lockrow JP, Wright JN, **Saneto RP**, **Amlie-Lefond C**. Epileptic Spasms Predict Poor Epilepsy Outcomes After Perinatal Stroke J Child Neurol. 2019 Jul 24:

Wainwright MS. Clinician, Heal Thyself": Decisional Conflict, Value Judgments, and Their Role in Practice Variation in Traumatic Brain Injury Pediatr Crit Care Med. 2019 Jul;20(7):679-680.

Sandrone S, Berthaud JV, Carlson C, Cios J, Dixit N, Farheen A, Kraker J, **Owens JWM**, Patino G, Sarva H, Weber D, Schneider LD. Education Research: Flipped classroom in neurology: Principles, practices, and perspectives Neurology. 2019 Jul 2;93(1):e106-e111.

Pediatric Neurology Faculty Publications 2014-2025

Kochanek PM, Tasker RC, Carney N, Totten AM, Adelson PD, Selden NR, Davis-O'Reilly C, Hart EL, Bell MJ, Bratton SL, Grant GA, Kissoon N, Reuter-Rice KE, Vavilala MS, **Wainwright MS**. Guidelines for the Management of Pediatric Severe Traumatic Brain Injury, Third Edition: Update of the Brain Trauma Foundation Guidelines, Executive Summary Neurosurgery. 2019 Jun 1;84(6):1169-1178.

Ibañez LV, Stoep AV, Myers K, Zhou C, Dorsey S, **Steinman KJ**, Stone WL. Promoting early autism detection and intervention in underserved communities: study protocol for a pragmatic trial using a stepped-wedge design. Psychiatry. 2019 Jun 7;19(1):169.

Barry M, Hallam DK, Bernard TJ, **Amlie-Lefond C** What is the Role of Mechanical Thrombectomy in Childhood Stroke? Pediatr Neurol. 2019 Jun;95:19-25.

Chelse AB, Kurz JE, Gorman KM, Epstein LG, Balmert LC, Ciolino JD, **Wainwright MS**. Remote poststroke headache in children: Characteristics and association with stroke recurrence. Neurol Clin Pract. 2019 Jun;9(3):194-200.

McKeever JD, Schultheis MT, Sim T, Goykhman J, **Patrick K**, Ehde DM, Woods SP. Selective reminding of prospective memory in Multiple Sclerosis Neuropsychol Rehabil. 2019 Jun;29(5):675-690.

Karasozen Y, Osbun JW, Parada CA, Busald T, Tatman P, Gonzalez-Cuyar LF, Hale CJ, Alcantara D, O'Driscoll M, **Dobyns WB**, Murray M, Kim LJ, Byers P, Dorschner MO, Ferreira M Jr. Somatic PDGFRB Activating Variants in Fusiform Cerebral Aneurysms Am J Hum Genet. 2019 May 2;104(5):968-976.

Marsillio LE, Manghi T, Carroll MS, Balmert LC, **Wainwright MS** Heart rate variability as a marker of recovery from critical illness in children. PLoS One. 2019 May 17;14(5)

Amengual-Gual M, Sánchez Fernández I, **Wainwright MS**. Novel drugs and early polypharmacotherapy in status epilepticus Seizure. 2019 May;68:79-88. doi: 10.1016/j.seizure.2018.08.004. Epub 2018 Aug 7. Review.

Guilliams KP, Kirkham FJ, Holzhauer S, Pavlakis S, Philbrook B, **Amlie-Lefond C**, Noetzel MJ, Dlamini N, Sharma M, Carpenter JL, Fox CK, Torres M, Ichord RN, Jordan LC, Dowling MM. Arteriopathy Influences Pediatric Ischemic Stroke Presentation, but Sickle Cell Disease Influences Stroke Management. Stroke. 2019 May;50(5):1089-1094

Messacar K, Sillau S, Hopkins SE, **Otten C**, Wilson-Murphy M, Wong B, Santoro JD, Treister A, Bains HK, Torres A, Zabrocki L, Glanternik JR, Hurst AL, Martin JA, Schreiner T, Makhani N, DeBiasi RL, Kruer MC, Tremoulet AH, Van Haren K, Desai J, Benson LA, Gorman MP, Abzug MJ, Tyler KL, Dominguez SR. Safety, tolerability, and efficacy of fluoxetine as an antiviral for acute flaccid myelitis. Neurology. 2019 Apr 30;92(18):e2118-e2126.

Kochanek PM, Tasker RC, Carney N, Totten AM, Adelson PD, Selden NR, Davis-O'Reilly C, Hart EL, Bell MJ, Bratton SL, Grant GA, Kissoon N, Reuter-Rice KE, Vavilala MS, **Wainwright MS**. Guidelines for the Management of Pediatric Severe Traumatic Brain Injury, Third Edition: Update of the Brain Trauma Foundation Guidelines. Pediatr Crit Care Med. 2019 Mar;20(3S Suppl 1):S1-S82. Erratum in: Pediatr Crit Care Med. 2019 Apr;20(4):404.

Vandervore LV, Schot R, Kasteleijn E, Oegema R, Stouffs K, Gheldof A, Grochowska MM, van der Sterre MLT, van Unen LMA, Wilke M, Elfferich P, van der Spek PJ, Heijnsman D, Grandone A, Demmers JAA, Dekkers DHW, Slotman JA, Kremers GJ, Schaaf GJ, Masius RG, van Essen AJ, Rump P, van Haeringen A, Peeters E, Altunoglu U, Kalayci T, Poot RA, **Dobyns WB**, Bahi-Buisson N, Verheijen FW, Jansen AC, Mancini GMS. Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics. Brain. 2019 Apr 1;142(4):867-884

Kahn-Kirby AH, Amagata A, Maeder CI, Mei JJ, Sideris S, Kosaka Y, Hinman A, Malone SA, Bruegger JJ, Wang L, Kim V, Shrader WD, Hoff KG, Latham JC, Ashley EA, Wheeler MT, Bertini E, Carrozzo R, Martinelli D, Dionisi-Vici C, Chapman KA, Enns GM, Gahl W, Wolfe L, **Saneto RP**, **Johnson SC**, Trimmer JK, Klein MB, Holst CR. Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy PLoS One. 2019 Mar 28;14(3):

Wainwright MS. Getting Excited About Paroxysms: Why Treating Sympathetic Hyperarousal After Traumatic Brain Injury May Be More Important Than We Appreciated Pediatr Crit Care Med. 2019 Mar;20(3):295-296.

Pediatric Neurology Faculty Publications 2014-2025

Kochanek PM, Tasker RC, Carney N, Totten AM, Adelson PD, Selden NR, Davis-O'Reilly C, Hart EL, Bell MJ, Bratton SL, Grant GA, Kissoon N, Reuter-Rice KE, Vavilala MS, **Wainwright MS**. Guidelines for the Management of Pediatric Severe Traumatic Brain Injury, Third Edition: Update of the Brain Trauma Foundation Guidelines, Executive Summary. *Pediatr Crit Care Med*. 2019 Mar;20(3):280-289

Kochanek PM, Tasker RC, Bell MJ, Adelson PD, Carney N, Vavilala MS, Selden NR, Bratton SL, Grant GA, Kissoon N, Reuter-Rice KE, **Wainwright MS**. Management of Pediatric Severe Traumatic Brain Injury: 2019 Consensus and Guidelines-Based Algorithm for First and Second Tier Therapies. *Pediatr Crit Care Med*. 2019 Mar;20(3):269-279.

Dlamini N, Muthusami P, **Amlie-Lefond C**. Childhood Moyamoya: Looking Back to the Future. *Pediatr Neurol*. 2019 Feb;91:11-19.

Recober A, **Patel PB**, Thibault DP, Hill AW, Kaiser EA, Willis AW. Sociodemographic Factors Associated With Hospital Care for Pediatric Migraine: A National Study Using the Kids' Inpatient Dataset. *Pediatr Neurol*. **2019 Feb**;91:34-40. doi: 10.1016/j.pediatrneurol.2017.10.023.

Wenger TL, Hopper RA, Rosen A, **Tully HM**, Cunningham ML, Lee A. A genotype-specific surgical approach for patients with Pfeiffer syndrome due to W290C pathogenic variant in FGFR2 is associated with improved developmental outcomes and reduced mortality. *Genet Med*. 2019 Feb;21(2):471-476.

Piao CS, Holloway AL, Hong-Routson S, **Wainwright MS**. Depression following traumatic brain injury in mice is associated with down-regulation of hippocampal astrocyte glutamate transporters by thrombin *J Cereb Blood Flow Metab*. 2019 Jan;39(1):58-73.

2018

Pediatric Neurology Faculty Publications 2014-2025

Global, Regional, and Country-Specific Lifetime Risks of Stroke, 1990 and 2016. GBD 2016 Lifetime Risk of Stroke Collaborators, Feigin VL, Nguyen G, Cercy K, Johnson CO, Alam T, Parmar PG, Abajobir AA, Abate KH, Abd-Allah F, Abejie AN, Abyu GY, Ademi Z, Agarwal G, Ahmed MB, Akinyemi RO, Al-Raddadi R, Aminde LN, **Amlie-Lefond C**, Ansari H, Asayesh H, Asgedom SW, Atey TM, Ayele HT, Banach M, Banerjee A, Barac A, Barker-Collo SL, Bärnighausen T, Barregard L, Basu S, Bedi N, Behzadifar M, Béjot Y, Bennett DA, Bensenor IM, Berhe DF, Boneya DJ, Brainin M, Campos-Nonato IR, Caso V, Castañeda-Orjuela CA, Rivas JC, Catalá-López F, Christensen H, Criqui MH, Damasceno A, Dandona L, Dandona R, Davletov K, de Courten B, deVeber G, Dokova K, Edessa D, Endres M, Faraon EJA, Farvid MS, Fischer F, Foreman K, Forouzanfar MH, Gall SL, Gebrehiwot TT, Geleijnse JM, Gillum RF, Giroud M, Goulart AC, Gupta R, Gupta R, Hachinski V, Hamadeh RR, Hankey GJ, Hareri HA, Havmoeller R, Hay SI, Hegazy MI, Hibstu DT, James SL, Jeemon P, John D, Jonas JB, Jóźwiak J, Kalani R, Kandel A, Kasaeian A, Kengne AP, Khader YS, Khan AR, Khang YH, Khubchandani J, Kim D, Kim YJ, Kivimaki M, Kokubo Y, Kolte D, Kopec JA, Kosen S, Kravchenko M, Krishnamurthi R, Kumar GA, Lafranconi A, Lavados PM, Legesse Y, Li Y, Liang X, Lo WD, Lorkowski S, Lotufo PA, Loy CT, Mackay MT, Abd El Razek HM, Mahdavi M, Majeed A, Malekzadeh R, Malta DC, Mamun AA, Mantovani LG, Martins SCO, Mate KK, Mazidi M, Mehata S, Meier T, Melaku YA, Mendoza W, Mensah GA, Meretoja A, Mezgebe HB, Miazgowski T, Miller TR, Ibrahim NM, Mohammed S, Mokdad AH, Moosazadeh M, Moran AE, Musa KI, Negoi RI, Nguyen M, Nguyen QL, Nguyen TH, Tran TT, Nguyen TT, Anggraini Ningrum DN, Norrving B, Noubiap JJ, O'Donnell MJ, Olagunju AT, Onuma OK, Owolabi MO, Parsaeian M, Patton GC, Piradov M, Pletcher MA, Pourmalek F, Prakash V, Qorbani M, Rahman M, Rahman MA, Rai RK, Ranta A, Rawaf D, Rawaf S, Renzaho AM, Robinson SR, Sahathevan R, Sahebkar A, Salomon JA, Santalucia P, Santos IS, Sartorius B, Schutte AE, Sepanlou SG, Shafieesabet A, Shaikh MA, Shamsizadeh M, Sheth KN, Sisay M, Shin MJ, Shiue I, Silva DAS, Sobngwi E, Soljak M, Sorensen RJD, Sposato LA, Stranges S, Suliankatchi RA, Tabarés-Seisdedos R, Tanne D, Nguyen CT, Thakur JS, Thrift AG, Tirschwell DL, Topor-Madry R, Tran BX, Nguyen LT, Truelsen T, Tsilimparis N, Tyrovolas S, Ukwaja KN, Uthman OA, Varakin Y, Vasankari T, Venketasubramanian N, Vlassov VV, Wang W, Werdecker A, Wolfe CDA, Xu G, Yano Y, Yonemoto N, Yu C, Zaidi Z, El Sayed Zaki M, Zhou M, Ziaeian B, Zipkin B, Vos T, Naghavi M, Murray CJL, Roth GA. *N Engl J Med*. 2018 Dec 20;379(25):2429-2437.

Venot Q, Blanc T, Rabia S, Berteloot L, Ladraa S, Duong J, Blanc E, **Johnson SC**, Huguin C, Boccara O, Sarnacki S, Boddaert N, Pannier S, Martinez F, Magassa S, Yamaguchi J, Knebelmann B, Merville P, Grenier N, Joly D, Cormier-Daire V, Michot C, Bole-Feyssot C, Picard A, Soupre V, Lyonnet S, Sadoine J, Slimani L, Chaussain C, Laroche-Raynaud C, Guibaud L, Broissand C, Amiel J, Legendre C, Terzi F, Canaud G: Targeted therapy in patients with PIK3CA-related overgrowth syndrome. *Nature*. 2018. 558:p540–546. PMID: 29899452

Blume HK. Posttraumatic headache in pediatrics: an update and review. **Curr Opin Pediatr**. 2018 Dec;30(6):755-763. Rhombencephalosynapsis: Fused cerebellum, confused geneticists. Aldinger KA, Dempsey JC, **Tully HM**, Grout ME, Mehaffey MG, **Dobyns WB**, Doherty D. *Am J Med Genet C Semin Med Genet*. 2018 Dec;178(4):432-439. doi: 10.1002/ajmg.c.31666. Review.

An update on oculocerebrocutaneous (Delleman-Oorthuys) syndrome. Moog U, **Dobyns WB**. *Am J Med Genet C Semin Med Genet*. 2018 Dec;178(4):414-422

Gust J, Taraseviciute A, Turtle CJ. Neurotoxicity Associated with CD19-Targeted CAR-T Cell Therapies. **CNS Drugs**. 2018 Dec;32(12):1091-1101

MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. Dobyns WB, Aldinger KA, Ishak GE, Mirzaa GM, Timms AE, Grout ME, Dremmen MHG, Schot R, Vandervore L, van Slegtenhorst MA, Wilke M, Kasteleijn E, Lee AS, Barry BJ, Chao KR, Szczaluba K, Kobori J, Hanson-Kahn A, Bernstein JA, Carr L, D'Arco F, Miyana K, Okazaki T, Saito Y, Sasaki M, Das S, Wheeler MM, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics; Center for Mendelian Genomics at the Broad Institute of MIT and Harvard, Engle EC, Verheijen FW, Doherty D, Mancini GMS. *Am J Hum Genet*. 2018 Dec 6;103(6):1009-1021.

Bozarth X, Dines JN, Cong Q, Mirzaa GM, Foss K, Lawrence Merritt J 2nd, Thies J, Mefford HC, **Novotny E**. Expanding clinical phenotype in CACNA1C related disorders: From neonatal onset severe epileptic encephalopathy to late-onset epilepsy. *Am J Med Genet A*. 2018 Dec;176(12):2733-2739.

Benedetti GM, Silverstein FS. Targeted Temperature Management in Pediatric Neurocritical Care. *Pediatr Neurol*. 2018 Nov;88:12-24.

Messacar K, Sillau S, Hopkins SE, **Otten C**, Wilson-Murphy M, Wong B, Santoro JD, Treister A, Bains HK, Torres A, Zabrocki L, Glanternik JR, Hurst AL, Martin JA, Schreiner T, Makhani N, DeBiasi RL, Kruer MC, Tremoulet AH, Van Haren K, Desai J, Benson LA, Gorman MP, Abzug MJ, Tyler KL, Dominguez SR. Safety, tolerability, and efficacy of fluoxetine as

Pediatric Neurology Faculty Publications 2014-2025

an antiviral for acute flaccid myelitis. *Neurology*. 2018 Nov 9.

Amlie-Lefond C, Flanagan J, Kanter J, **Dobyns WB**. The Genetic Landscape of Cerebral Steno-Occlusive Arteriopathy and Stroke in Sickle Cell Anemia. *J Stroke Cerebrovasc Dis*. 2018 Nov;27(11):2897-2904.

Bozarth X, Foss K, Mefford HC. A de novo in-frame deletion of CASK gene causes early onset infantile spasms and supratentorial cerebral malformation in a female patient *Am J Med Genet A*. 2018 Nov;176(11):2425-2429.

Schanze I, Bunt J, Lim JWC, Schanze D, Dean RJ, Alders M, Blanchet P, Attié-Bitach T, Berland S, Boogert S, Boppudi S, Bridges CJ, Cho MT, **Dobyns WB**, Donnai D, Douglas J, Earl DL, Edwards TJ, Faivre L, Fregeau B, Genevieve D, Gérard M, Gatinois V, Holder-Espinasse M, Huth SF, Izumi K, Kerr B, Lacaze E, Lakeman P, Mahida S, Mirzaa GM, Morgan SM, Nowak C, Peeters H, Petit F, Pilz DT, Puechberty J, Reinstein E, Rivière JB, Santani AB, Schneider A, Sherr EH, Smith-Hicks C, Wieland I, Zackai E, Zhao X, Gronostajski RM, Zenker M, Richards LJ. NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly *Am J Hum Genet*. 2018 Nov 1;103(5):752-768.

Piacitelli AM, Jensen DM, Brandling-Bennett H, Gray MM, Batra M, **Gust J**, Thaker A, Paschal C, Tsuchiya K, Pritchard CC, Perkins J, Mirzaa GM, Bennett JT. Characterization of a severe case of PIK3CA-related overgrowth at autopsy by droplet digital polymerase chain reaction and report of PIK3CA sequencing in 22 patients. *Am J Med Genet A*. 2018 Nov;176(11):2301-2308.

Marashly A, Loman MM, Lew SM Stereotactic laser ablation for nonlesional cingulate epilepsy: case report. *J Neurosurg Pediatr*. 2018 Nov 1;22(5):481-488

Fullerton HJ, Stence N, Hills NK, Jiang B, **Amlie-Lefond C**, Bernard TJ, Friedman NR, Ichord R, Mackay MT, Rafay MF, Chabrier S, Steinlin M, Elkind MSV, deVeber GA, Wintermark M; VIPS Investigators. Focal Cerebral Arteriopathy of Childhood: Novel Severity Score and Natural History. *Stroke*. 2018 Nov;49(11):2590-2596.

Shurtleff H, Barry D, Chanprasert S, Firman T, **Warner M**, **Saneto RP**. Cognitive characteristics of mitochondrial diseases in children. *Epilepsy Behav*. 2018 Nov;88:235-243

Paganoni S, Nicholson K, **Leigh F**, Swoboda K, Chad D, Drake K, Haley K, Cudkowicz M, Berry JD. Developing multidisciplinary clinics for neuromuscular care and research *Muscle Nerve*. 2017 Nov;56(5):848-858.

Leigh F, Ferlini A, Biggar D, Bushby K, Finkel R, Morgenroth LP, Wagner KR. Neurology Care, Diagnostics, and Emerging Therapies of the Patient With Duchenne Muscular Dystrophy. *Pediatrics*. 2018 Oct;142(Suppl 2):S5-S16.

Lockrow J, **Tully H**, **Saneto RP**. Epileptic spasms as the presenting seizure type in a patient with a new "O" of TORCH, congenital Zika virus infection *Epilepsy Behav Case Rep*. 2018 Oct 18;11:1-3.

Ramos EM, Carecchio M, Lemos R, Ferreira J, Legati A, Sears RL, Hsu SC, Panteghini C, Magistrelli L, Salsano E, Esposito S, Taroni F, Richard AC, Tranchant C, Anheim M, Aygnac X, Goizet C, Vidailhet M, Maltete D, Wallon D, Frebourg T, Pimentel L, Geschwind DH, Vanakker O, Galasko D, Fogel BL, Innes AM, Ross A, **Dobyns WB**, Alcantara D, O'Driscoll M, Hannequin D, Champion D; French PFBC study group, Oliveira JR, Garavaglia B, Coppola G, Nicolas G. Primary brain calcification: an international study reporting novel variants and associated phenotypes. *Eur J Hum Genet*. 2018 Oct;26(10):1462-1477

Benedetti GM, Silverstein FS, Rau SM, Lester SG, Benedetti MH, Shellhaas RA. Sedation and Analgesia Influence Electroencephalography Monitoring in Pediatric Neurocritical Care. *Pediatr Neurol*. 2018 Oct;87:57-64.

Coryell J, Gaillard WD, Shellhaas RA, Grinspan ZM, Wirrell EC, Knupp KG, Wusthoff CJ, Keator C, Sullivan JE, Lodenkemper T, Patel A, Chu CJ, Massey S, **Novotny EJ Jr**, **Saneto RP**, Berg AT. Pediatrics. Neuroimaging of Early Life Epilepsy 2018 Sep;142(3).

Riley JS, Quarato G, Cloix C, **Lopez J**, O'Prey J, Pearson M, Chapman J, Sesaki H, Carlin LM, Passos JF, Wheeler AP, Oberst A, Ryan KM, Tait SW. Mitochondrial inner membrane permeabilisation enables mtDNA release during apoptosis. *EMBO J*. 2018 Sep 3;37(17).

Pediatric Neurology Faculty Publications 2014-2025

Grassia F, Poliakov AV, **Poliachik SL**, Casimo K, Friedman SD, **Shurtleff H**, Giussani C, **Novotny EJ**, Ojemann JG, Hauptman JS. Changes in resting-state connectivity in pediatric temporal lobe epilepsy. *J Neurosurg Pediatr*. 2018 Sep;22(3):270-275.

López-Chiriboga AS, Klein C, Zekeridou A, McKeon A, Dubey D, Flanagan EP, Lennon VA, Tillemma JM, Wirrell EC, Patterson MC, Gadoth A, Aaen JG, Brenton JN, Bui JD, Moen A, **Otten C**, Piquet A, Pittock SJ. LGI1 and CASPR2 neurological autoimmunity in children. *Ann Neurol*. 2018 Sep;84(3):473-480.

Wainwright MS. Exposing and Engaging Unknown Unknowns in Traumatic Brain Injury. *Crit Care Med*. 2018 Sep;46(9):1556-1557.

Vasquez A, Gaínza-Lein M, Sánchez Fernández I, Abend NS, Anderson A, Brenton JN, Carpenter JL, Chapman K, Clark J, Gaillard WD, Glauser T, Goldstein J, Goodkin HP, Lai YC, Loddenkemper T, McDonough TL, Mikati MA, Nayak A, Payne E, Riviello J, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Tasker RC; Pediatric Status Epilepticus Research Group (pSERG). Hospital Emergency Treatment of Convulsive Status Epilepticus: Comparison of Pathways From Ten Pediatric Research Centers. *Pediatr Neurol*. 2018 Sep;86:33-41.

Vora SB, **Amlie-Lefond C**, Perez FA, Melvin AJ. Varicella-Associated Stroke. *J Pediatr*. 2018 Aug;199:281-281.e1.

Brock S, Stouffs K, Scalais E, D'Hooghe M, Keymolen K, Guerrini R, **Dobyns WB**, Di Donato N, Jansen AC. Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. *Eur J Hum Genet*. 2018 Aug;26(8):1132-1142.

Fat, Pharmacokinetics, and Fosphenytoin: Bigger Doses May Be Better. **Wainwright MS**. *Pediatr Crit Care Med*. 2018 Aug;19(8):784-785.

Wrede JE, Parsons EC, Watson NF. A Novel Treatment for Nasolacrimal Air Regurgitation Into the Eye With CPAP: The Total Face Mask. *J Clin Sleep Med*. 2018 Aug 15;14(8):1415-1417.

Chan P, **Patel P**, Hellmuth J, Colby DJ, Kroon E, Sacdalan C, Pinyakorn S, Jagodzinski L, Krebs S, Ananworanich J, Valcour V, Spudich S; Distribution of Human Immunodeficiency Virus (HIV) Ribonucleic Acid in Cerebrospinal Fluid and Blood Is Linked to CD4/CD8 Ratio During Acute HIV. RV254/SEARCH 010 Study Team. *J Infect Dis*. 2018 Aug 14;218(6):937-945.

Schaffer AE, Breuss MW, Caglayan AO, Al-Sanaa N, Al-Abdulwahed HY, Kaymakçalan H, Yılmaz C, Zaki MS, Rosti RO, Copeland B, Baek ST, Musaev D, Scott EC, Ben-Omran T, Kariminejad A, Kayserili H, Mojahedi F, Kara M, Cai N, Silhavy JL, Elsharif S, Fenercioglu E, Barshop BA, Kara B, Wang R, Stanley V, James KN, Nachnani R, Kalur A, Megahed H, Incecik F, Danda S, Alanay Y, Fageih E, Melikishvili G, Mansour L, Miller I, Sukhudyann B, Chelly J, **Dobyns WB**, Bilguvar K, Jamra RA, Gunel M, Gleeson JG. Biallelic loss of human CTNNA2, encoding α N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. *Nat Genet*. 2018 Aug;50(8):1093-1101.

Nickels KC, Zaccariello MJ, **Hamiwka LD**, Wirrell EC. Cognitive and neurodevelopmental comorbidities in paediatric epilepsy. *Nat Rev Neurol*. 2016 Aug;12(8):465-76.

Amengual-Gual M, Sánchez Fernández I, **Wainwright MS**. Novel drugs and early polypharmacotherapy in status epilepticus. *Seizure*. 2018 Aug 7.

Patrick KE, Hurewitz F, McCurdy MD, Agate FT, Daly BP, Tarazi RA, Chute DL, Schultheis MT. Driving Comparisons Between Young Adults with Autism Spectrum Disorder and Typical Development. *J Dev Behav Pediatr*. 2018 Jul/Aug;39(6):451-460.

The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. Coughlin CR 2nd, Swanson MA, Spector E, Meeks NJL, Kronquist KE, Aslamy M, Wempe MF, van Karnebeek CDM, **Gospe SM Jr**, Aziz VG, Tsai BP, Gao H, Nagy PL, Hyland K, van Dooren SJM, Salomons GS, Van Hove JLK. *J Inher Metab Dis*. 2018 Jul 24.

Piacitelli AM, Jensen DM, Brandling-Bennett H, Gray MM, Batra M, **Gust J**, Thaker A, Paschal C, Tsuchiya K, Pritchard CC, Perkins J, Mirzaa GM, Bennett JT. Characterization of a severe case of PIK3CA-related overgrowth at autopsy by droplet digital polymerase chain reaction and report of PIK3CA sequencing in 22 patients. *Am J Med Genet A*. 2018 Jul 31.

Pediatric Neurology Faculty Publications 2014-2025

- Gospe SM Jr.** Developmental outcome in pyridoxine-dependent epilepsy: Better late (onset) than early Eur J Paediatr Neurol. 2018 Jul;22(4):575-576.
- Harms MB, **Shannon Bowen KE**, Hanson JL, Pollak Instrumental learning and cognitive flexibility processes are impaired in children exposed to early life stress. Dev Sci. 2018 Jul;21(4):e12596.
- Oesch G, Maga AM, Friedman SD, Poliachik SL, Budech CB, Wright JN, Bok LA, **Gospe SM Jr.** Geometric morphometrics reveal altered corpus callosum shape in pyridoxine-dependent epilepsy Neurology. 2018 Jul 3;91(1):e78-e86.
- PAR3 dysfunction in conjunction with dynamic HIPPO signaling drives cortical enlargement with massive heterotopia. Liu WA, Chen S, Li Z, Lee CH, Mirzaa G, **Dobyns WB**, Ross ME, Zhang J, Shi SH. Genes Dev. 2018 Jun 1;32(11-12):763-780
- Wenger TL, Hopper RA, Rosen A, **Tully HM**, Cunningham ML, Lee A. A genotype-specific surgical approach for patients with Pfeiffer syndrome due to W290C pathogenic variant in FGFR2 is associated with improved developmental outcomes and reduced mortality Genet Med. 2018 Jun 18.
- Sánchez Fernández I, Gaínza-Lein M, Abend NS, Anderson AE, Arya R, Brenton JN, Carpenter JL, Chapman KE, Clark J, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Helseth AR, Jackson MC, Kapur K, Lai YC, McDonough TL, Mikati MA, Nayak A, Peariso K, Riviello JJ Jr, Tasker RC, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group (pSERG): **Morgan, LA**. Factors associated with treatment delays in pediatric refractory convulsive status epilepticus. Neurology. 2018 May 8;90(19):e1692-e1701.
- Shukla A, **Saneto RP**, Hebbar M, Mirzaa G, Girisha KM. A neurodegenerative mitochondrial disease phenotype due to biallelic loss-of-function variants in PNPLA8 encoding calcium-independent phospholipase A2γ. Am J Med Genet A. 2018 May;176(5):1232-1237.
- Kannan N, Wang J, Mink RB, **Wainwright MS**, Groner JI, Bell MJ, Giza CC, Zatzick DF, Ellenbogen RG, Boyle LN, Mitchell PH, Rivara FP, Rowhani-Rahbar A, Vavilala MS; PEGASUS (Pediatric Guideline Adherence Outcomes) Study. Timely Hemodynamic Resuscitation and Outcomes in Severe Pediatric Traumatic Brain Injury: Preliminary Findings. Pediatr Emerg Care. 2018 May;34(5):325-329.
- Leibovitz Z, Mandel H, Falik-Zaccai TC, Ben Harouch S, Savitzki D, Krajden-Haratz K, Gindes L, Tamarkin M, Lev D, **Dobyns WB**, Lerman-Sagie T. Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. Eur J Paediatr Neurol. 2018 May;22(3):525-531.
- Heinzen EL, O'Neill AC, Zhu X, Allen AS, Bahlo M, Chelly J, Chen MH, **Dobyns WB**, Freytag S, Guerrini R, Leventer RJ, Poduri A, Robertson SP, Walsh CA, Zhang M; Epi4K Consortium; Epilepsy Phenome/Genome Project. De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genet. 2018 May 8;14(5):.
- Di Donato N, Timms AE, Aldinger KA, Mirzaa GM, Bennett JT, Collins S, Olds C, Mei D, Chiari S, Carvill G, Myers CT, Rivière JB, Zaki MS; University of Washington Center for Mendelian Genomics, Gleeson JG, Rump A, Conti V, Parrini E, Ross ME, Ledbetter DH, Guerrini R, **Dobyns WB**. Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genet Med. 2018 Apr 19.
- Grinspan ZM, Shellhaas RA, Coryell J, Sullivan JE, Wirrell EC, Mytinger JR, Gaillard WD, Kossoff EH, Valencia I, Knupp KG, Wusthoff C, Keator C, Ryan N, Loddenkemper T, Chu CJ, **Novotny EJ Jr**, Millichap J, Berg AT. Comparative Effectiveness of Levetiracetam vs Phenobarbital for Infantile Epilepsy. JAMA Pediatr. 2018 Apr 1;172(4):352-360.
- Gaínza-Lein M, Sánchez Fernández I, Jackson M, Abend NS, Arya R, Brenton JN, Carpenter JL, Chapman KE, Gaillard WD, Glauser TA, Goldstein JL, Goodkin HP, Kapur K, Mikati MA, Peariso K, Tasker RC, Tchapyjnikov D, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group. Association of Time to Treatment With Short-term Outcomes for Pediatric Patients With Refractory Convulsive Status Epilepticus. JAMA Neurol. 2018 Apr 1;75(4):410-418.
- Patel PB**, Spudich, S. Global Health Neurology: HIV/AIDS. Semin Neurol. 2018 Apr;38(2):238-246.
- Weaver JJ, **Natarajan N**, Shaw DWW, Apkon SD, Koo KSH, Shivaram GM, Monroe EJ. Transforaminal intrathecal delivery of nusinersen using cone-beam computed tomography for children with spinal muscular atrophy and extensive surgical instrumentation: early results of technical success and safety Pediatr Radiol. 2018 Mar;48(3):392-397.
- Patel P**, Ross A, Henretig FM, Liu G, Harding B, Panzer J. Clinical Reasoning: A 12-year-old girl with headache and

Pediatric Neurology Faculty Publications 2014-2025

change in mental status. *Neurology*. 2018 Mar 13;90(11):524-529.

Monroe EJ, **Amlie-Lefond CM**. Cone beam computed tomography-guided transpterygoidal aspiration of a carotid space abscess in Lemierre's syndrome.. *Radiol Case Rep*. 2018 Mar 30;13(3):618-621. doi: 10.1016/j.radcr.2018.03.007. eCollection 2018 Jun.

Ruggeri G, Timms AE, Cheng C, Weiss A, **Kollros P**, Chapman T, **Tully H**, Mirzaa GM. Bi-allelic mutations of CCDC88C are a rare cause of severe congenital hydrocephalus. *Am J Med Genet A*. 2018 Mar;176(3):676-681.

Adams Waldorf KM, Nelson BR, Stencel-Baerenwald JE, Studholme C, Kapur RP, Armistead B, Walker CL, Merillat S, Vornhagen J, Tisoncik-Go J, Baldessari A, Coleman M, Dighe MK, Shaw DWW, Roby JA, Santana-Ufret V, Boldenow E, Li J, Gao X, Davis MA, Swannstrom JA, Jensen K, Widman DG, Baric RS, Medwid JT, Hanley KA, Ogle J, Gough GM, Lee W, English C, Durning WM, Thiel J, Gatenby C, Dewey EC, Fairgrieve MR, Hodge RD, Grant RF, Kuller L, **Dobyns WB**, Hevner RF, Gale M Jr, Rajagopal L. Congenital Zika virus infection as a silent pathology with loss of neurogenic output in the fetal brain. *Nat Med*. 2018 Mar;24(3):368-374.

Berg AT, Chakravorty S, Koh S, Grinspan ZM, Shellhaas RA, **Saneto RP**, Wirrell EC, Coryell J, Chu CJ, Mytinger JR, Gaillard WD, Valencia I, Knupp KG, Loddenkemper T, Sullivan JE, Poduri A, Millichap JJ, Keator C, Wusthoff C, Ryan N, **Dobyns WB**, Hegde M. Why West? Comparisons of clinical, genetic and molecular features of infants with and without spasms. *PLoS One*. 2018 Mar 8;13(3):

Natarajan N, Beatty CW, Gust J, Hamiwka L. Provider Practices of Phenobarbital Discontinuation in Neonatal Seizures *J Child Neurol*. 2018 Feb;33(2):153-157.

Evaluation and Acute Management of Ischemic Stroke in Infants and Children. **Amlie-Lefond C**. *Continuum (Minneapolis, Minn)*. 2018 Feb;24(1, Child Neurology):150-170. doi: 10.1212. Review.

PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Reijnders MRF, Janowski R, Alvi M, Self JE, van Essen TJ, Vreeburg M, Rouhl RPW, Stevens SJC, Stegmann APA, Schieving J, Pfundt R, van Dijk K, Smeets E, Stumpel CTRM, Bok LA, Cobben JM, Engelen M, Mansour S, Whiteford M, Chandler KE, Douzgou S, Cooper NS, Tan EC, Foo R, Lai AHM, Rankin J, Green A, Lönnqvist T, Isohanni P, Williams S, **Ruhoy I**, Carvalho KS, Dowling JJ, Lev DL, Sterbova K, Lassuthova P, Neupauerová J, Waugh JL, Keros S, Clayton-Smith J, Smithson SF, Brunner HG, van Hoeckel C, Anderson M, Clowes VE, Siu VM, Ddd Study T, Selber P, Leventer RJ, Nellaker C, Niessing D, Hunt D, Baralle D. *J Med Genet*. 2018 Feb;55(2):104-113

Weaver KE, Poliakov A, **Novotny EJ**, Olson JD, Grabowski TJ, Ojemann JG. Electrocorticography and the early maturation of high-frequency suppression within the default mode network. *J Neurosurg Pediatr*. 2018 Feb;21(2):133-140.

Phelps IG, Dempsey JC, Grout ME, Isabella CR, **Tully HM**, Doherty D, Bachmann-Gagescu R. Interpreting the clinical significance of combined variants in multiple recessive disease genes: systematic investigation of Joubert syndrome yields little support for oligogenicity. *Genet Med*. 2018 Feb;20(2):223-233.

Wainwright MS. Neurologic Complications in the Pediatric Intensive Care Unit. *Continuum (Minneapolis, Minn)*. 2018 Feb;24(1, Child Neurology):288-299.

Fry AE, Fawcett KA, Zelnik N, Yuan H, Thompson BAN, Shemer-Meir L, Cushion TD, Mugalaasi H, Sims D, Stoodley N, Chung SK, Rees MI, Patel CV, Brueton LA, Layet V, Giuliano F, Kerr MP, Banne E, Meiner V, Lerman-Sagie T, Helbig KL, Kofman LH, Knight KM, Chen W, Kannan V, Hu C, Kusumoto H, Zhang J, Swanger SA, Shaulsky GH, Mirzaa GM, Muir AM, Mefford HC, **Dobyns WB**, Mackenzie AB, Mullins JGL, Lemke JR, Bahi-Buisson N, Traynelis SF, Iago HF, Pilz DT. De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. *Brain*. 2018 Jan 22. doi:10.1093/brain/awx358.

Wang AC, Ibrahim GM, Poliakov AV, Wang PI, Fallah A, Mathern GW, Buckley RT, Collins K, Weil AG, **Shurtleff HA, Warner MH**, Perez FA, Shaw DW, Wright JN, **Saneto RP, Novotny EJ**, Lee A, Browd SR, Ojemann JG. Corticospinal tract atrophy and motor fMRI predict motor preservation after functional cerebral hemispherectomy. *J Neurosurg Pediatr*. 2018 Jan;21(1):81-89

Steiner JE, McCoy GN, Hess CP, **Dobyns WB**, Metry DW, Drolet BA, Maheshwari M, Siegel DH. Structural malformations of the brain, eye, and pituitary gland in PHACE syndrome. *Am J Med Genet A*. 2018 Jan;176(1):48-55. doi:10.1002/ajmg.a.38523

Pediatric Neurology Faculty Publications 2014-2025

Patel P., Ross, A., Lui, G., Harding, B., Henretig, F., Panzer, J. Clinical Reasoning: A 12 year old Female with Headache and Change in Mental Status. *Neurology*. 90(11): 524-529, 2018

Chan P, Hellmuth J, Colby D, Kroon E, Sacdalan C, Fletcher J, **Patel P**, Pinyakorn S, Valcour V, Ananworanich J, Spudich S.J Safety of lumbar puncture procedure in an international research setting during acute HIV infection. *Virus Erad*. 2018 Jan 1;4(1):16-20.

2017

Johnson SC, Gonzalez B, Zhang Q, Milholland B, Zhang Z, Suh Y: Network analysis of mitonuclear GWAS reveals functional networks and tissue expression profiles of disease-associated genes. *Human Genetics*. 2017. 136: p55-65. PMID: 27704213

Amlie-Lefond CM, Pavlakis SG. Preventing cognitive decline in sickle cell disease: A good night's sleep. *Neurology*. 2017 Dec 12;89(24):2402-2403. doi: 10.1212/WNL.0000000000004754

Wallace AS, Hudac CM, **Steinman KJ**, Peterson JL, DesChamps TD, Duyzend MH, Nuttle X, Eichler EE, Bernier RA. Longitudinal report of child with de novo 16p11.2 triplication. *Clin Case Rep*. 2017 Dec 6;6(1):147-154. eCollection 2018 Jan. PMID: 29375855

Gust J, Hay KA, Hanafi LA, Li D, Myerson D, Gonzalez-Cuyar LF, Yeung C, Liles WC, Wurfel M, Lopez JA, Chen J, Chung D, Harju-Baker S, Özpölat T, Fink KR, Riddell SR, Maloney DG, Turtle CJ. Endothelial Activation and Blood-Brain Barrier Disruption in Neurotoxicity after Adoptive Immunotherapy with CD19 CAR-T Cells. *Cancer Discov*. 2017 Dec;7(12):1404-1419. doi: 10.1158/2159-8290.CD-17-0698.

Demarest ST, Shellhaas RA, Gaillard WD, Keator C, Nickels KC, Hussain SA, Loddenkemper T, Patel AD, **Saneto RP**, Wirrell E, Sánchez Fernández I, Chu CJ, Grinspan Z, Wusthoff CJ, Joshi S, Mohamed IS, Stafstrom CE, Stack CV, Yozawitz E, Bluvstein JS, Singh RK, Knupp KG; Pediatric Epilepsy Research Consortium. The impact of hypsarrhythmia on infantile spasms treatment response: Observational cohort study from the National Infantile Spasms Consortium. *Epilepsia*. 2017 Dec;58(12):2098-2103. doi: 10.1111/epi.13937.

Parikh S, Goldstein A, Karaa A, Koenig MK, Anselm I, Brunel-Guitton C, Christodoulou J, Cohen BH, Dimmock D, Enns GM, Falk MJ, Feigenbaum A, Frye RE, Ganesh J, Griesemer D, Haas R, Horvath R, Korson M, Kruer MC, Mancuso M, McCormack S, Raboisson MJ, Reimschisel T, Salvarinova R, **Saneto RP**, Scaglia F, Shoffner J, Stacpoole PW, Sue CM, Tarnopolsky M, Van Karnebeek C, Wolfe LA, Cunningham ZZ, Rahman S, Chinnery PF. Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genet Med*. 2017 Dec;19(12). doi: 10.1038/gim.2017.107.

Hay KA, Hanafi LA, Li D, **Gust J**, Liles WC, Wurfel MM, López JA, Chen J, Chung D, Harju-Baker S, Cherian S, Chen X, Riddell SR, Maloney DG, Turtle CJ. Kinetics and biomarkers of severe cytokine release syndrome after CD19 chimeric antigenreceptor-modified T-cell therapy. *Blood*. 2017 Nov 23;130(21):2295-2306. doi:10.1182/blood-2017-06-793141. Epub 2017 Sep 18. PubMed PMID: 28924019; PubMed

Farhadian S, **Patel P**, Spudich S. Neurological Complications of HIV Infection. *Curr Infect Dis Rep*. 2017 Nov 21;19(12):50. doi: 10.1007/s11908-017-0606-5. PMID: 29164407 Review. *Global Health Neurology: HIV/AIDS*

Shellhaas RA, Berg AT, Grinspan ZM, Wusthoff CJ, Millichap JJ, Loddenkemper T, Coryell J, **Saneto RP**, Chu CJ, Joshi SM, Sullivan JE, Knupp KG, Kossoff EH, Keator C, Wirrell EC, Mytinger JR, Valencia I, Massey S, Gaillard WD. Initial Treatment for Nonsyndromic Early-Life Epilepsy: An Unexpected Consensus. *Pediatr Neurol*. 2017 Oct;75:73-79. doi: 10.1016/j.pediatrneurol.2017.06.011.

Kirk V, Baughn J, D'Andrea L, Friedman N, Galion A, Garetz S, Hassan F, **Wrede J**, Harrod CG, Malhotra RK. American Academy of Sleep Medicine Position Paper for the Use of a Home Sleep Apnea Test for the Diagnosis of OSA in Children. *J Clin Sleep Med*. 2017 Oct 15;13(10):1199-1203. doi: 0.5664/jcsm.6772.

Pediatric Neurology Faculty Publications 2014-2025

Rilinger JF, Smith CM, deRegnier RAO, Goldstein JL, Mills MG, Reynolds M, Backer CL, Burrowes DM, Mehta P, Piantino J, **Wainwright MS**. Transcranial Doppler Identification of Neurologic Injury during Pediatric Extracorporeal Membrane Oxygenation Therapy. *J Stroke Cerebrovasc Dis*. 2017 Oct;26(10):2336-2345. doi:10.1016/j.jstrokecerebrovasdis.2017.05.022

De Mori R, Romani M, D'Arrigo S, Zaki MS, Loreface E, Tardivo S, Biagini T, Stanley V, Musaev D, Fluss J, Micalizzi A, Nuovo S, Illi B, Chiapparini L, Di Marcotullio L, Issa MY, Anello D, Casella A, Ginevrino M, Leggins AS, Roosing S, Alfonsi R, Rosati J, Schot R, Mancini GMS, Bertini E, **Dobyns WB**, Mazza T, Gleeson JG, Valente EM. Hypomorphic Recessive Variants in *SUFU* Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. *Am J Hum Genet*. 2017 Oct 5;101(4):552-563. doi: 10.1016/j.ajhg.2017.08.017. Epub 2017 Sep 28

Alcantara D, Timms AE, Gripp K, Baker L, Park K, Collins S, Cheng C, Stewart F, Mehta SG, Saggat A, Sztriha L, Zombor M, Caluseriu O, Mesterman R, Van Allen MI, Jacquinet A, Ygberg S, Bernstein JA, Wenger AM, Guturu H, Bejerano G, Gomez-Ospina N, Lehman A, Alfei E, Pantaleoni C, Conti V, Guerrini R, Moog U, Graham JM Jr, Hevner R, **Dobyns WB**, O'Driscoll M, Mirzaa GM. Mutations of *AKT3* are associated with a wide spectrum of developmental disorders including extreme megalencephaly. *Brain*. 2017 Oct 1;140(10):2610-2622. doi: 10.1093/brain/awx203

Saneto RP. Genetics of Mitochondrial Disease. *Adv Genet*. 2017;98:63-116. doi: 10.1016/bs.adgen.2017.06.002. Epub 2017 Sep

Berg AT, Coryell J, **Saneto RP**, Grinspan ZM, **Alexander JJ**, Kekis M, Sullivan JE, Wirrell EC, Shellhaas RA, Mytinger JR, Gaillard WD, Kossoff EH, Valencia I, Knupp KG, Wusthoff C, Keator C, **Dobyns WB**, Ryan N, Loddenkemper T, Chu CJ, **Novotny EJ Jr**, Koh S. Early-Life Epilepsies and the Emerging Role of Genetic Testing. *JAMA Pediatr*. 2017 Sep 1;171(9):863-871. doi:10.1001/jamapediatrics.2017.1743..

Rosenthal ES, Claassen J, **Wainwright MS**, Husain AM, Vaitkevicius H, Raines S, Hoffmann E, Colquhoun H, Doherty JJ, Kaness SJ. Brexanolone as adjunctive therapy in super-refractory status epilepticus. *Ann Neurol*. 2017 Sep;82(3):342-352. doi: 10.1002/ana.25008.

Syrbe S, Harms FL, Parrini E, Montomoli M, Mütze U, Helbig KL, Polster T, Albrecht B, Bernbeck U, van Binsbergen E, Biskup S, Burglen L, Denecke J, Heron B, Heyne HO, Hoffmann GF, Hornemann F, Matsushige T, Matsuura R, Kato M, Korenke GC, Kuechler A, Lämmer C, Merckenschlager A, Mignot C, Ruf S, Nakashima M, Saitsu H, Stamberger H, Pisano T, Tohyama J, Weckhuysen S, Werckx W, Wickert J, Mari F, Verbeek NE, Møller RS, Koeleman B, Matsumoto N, **Dobyns WB**, Battaglia D, Lemke JR, Kutsche K, Guerrini R. Delineating *SPTAN1* associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*. 2017 Sep 1;140(9):2322-2336.

Bonwitt J, Poel A, DeBolt C, Gonzales E, Lopez A, Routh J, Rietberg K, Linton N, Reggin J, Sejvar J, Lindquist S, **Otten CE**. Acute Flaccid Myelitis Among Children - Washington, September-November 2016. *MMWR Morb Mortal Wkly Rep*. 2017 Aug 11;66(31):826-829.

Oegema R, Baillat D, Schot R, van Unen LM, Brooks A, Kia SK, Hoogeboom AJM, Xia Z, Li W, Cesaroni M, Lequin MH, van Slegtenhorst M, **Dobyns WB**, de Coe IFM, van den Berg D, Verheijen FW, Kremer A, van der Spek PJ, Heijnsman D, Wagner EJ, Fornerod M, Mancini GMS. Correction: Human mutations in integrator complex subunits link transcriptome integrity to brain development. *PLoS Genet*. 2017 Aug 1;13(8):e1006923. doi: 10.1371/journal.pgen.1006923. eCollection 2017 Aug. PubMed

McAdams RM, Fleiss B, Traudt C, Schwendimann L, Snyder JM, Haynes RL, **Natarajan N**, Gressens P, Juul SE. Long-Term Neuropathological Changes Associated with Cerebral Palsy in a Nonhuman Primate Model of Hypoxic-Ischemic Encephalopathy. *Dev Neurosci*. 2017;39(1-4):124-140.

Novotny EJ Jr. Early genetic testing for neonatal epilepsy: When, why, and how? *Neurology*. 2017 Aug 29;89(9):880-881.

Platzer K, Yuan H, Schütz H, Winschel A, Chen W, Hu C, Kusumoto H, Heyne HO, Helbig KL, Tang S, Willing MC, Tinkle BT, Adams DJ, Depienne C, Keren B, Mignot C, Frengen E, Strømme P, Biskup S, Döcker D, Strom TM, Mefford HC, Myers CT, Muir AM, LaCroix A, Sadleir L, Scheffer IE, Brilstra E, van Haelst MM, van der Smagt JJ, Bok LA, Møller RS, Jensen UB, Millichap JJ, Berg AT, Goldberg EM, De Bie I, Fox S, Major P, Jones JR, Zackai EH, Abou Jamra R, Rolfs A, Leventer RJ, Lawson JA, Roscioli T, Jansen FE, Ranza E, Korff CM, Lehesjoki AE, Courage C, Linnankivi T, Smith DR, Stanley C, Mintz M, McKnight D, Decker A, Tan WH, Tarnopolsky MA, Brady LI, Wolff M, Dondit L, Pedro HF, Parisotto SE, Jones KL, Patel AD, Franz DN, Vanzo R, Marco E, Ranells JD, Di Donato N, **Dobyns WB**, Laube B, Traynelis SF,

Pediatric Neurology Faculty Publications 2014-2025

Lemke JR. GRIN2B encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *J Med Genet*. 2017 Jul;54(7):460-470. doi: 10.1136/jmedgenet-2016-104509.

Van De Weghe JC, Rusterholz TDS, Latour B, Grout ME, Aldinger KA, Shaheen R, Dempsey JC, Maddirevula S, Cheng YH, Phelps IG, Gesemann M, Goel H, Birk OS, Alanzi T, Rawashdeh R, Khan AO; University of Washington Center for Mendelian Genomics, Bamshad MJ, Nickerson DA, Neuhauss SCF, **Dobyns WB**, Alkuraya FS, Roepman R, Bachmann-Gagescu R, Doherty D. Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. *Am J Hum Genet*. 2017 Jul 6;101(1):23-36. doi: 10.1016/j.ajhg.2017.05.010. Epub 2017 Jun 15. PubMed PMID: 28625504; PubMed Central PMCID: PMC5501774.

AJ, Guerrini R, **Dobyns WB**. Lissencephaly: Expanded imaging and clinical classification. *Am J Med Genet A*. 2017 Jun;173(6):1473-1488. doi:10.1002/ajmg.a.38245. Epub 2017 Apr 25.

Keros S, Buraniqi E, Alex B, Antonetty A, Fialho H, Hafeez B, Jackson MC, Jawahar R, Kjelleren S, Stewart E, **Morgan LA, Wainwright MS**, Sogawa Y, Patel AD, Loddenkemper T, Grinspan ZM. Increasing Ketamine Use for Refractory Status Epilepticus in US Pediatric Hospitals. *J Child Neurol*. 2017 Jun;32(7):638-646. doi: 10.1177/0883073817698629

Garavelli L, Ivanovski I, Caraffi SG, Santodirocco D, Pollazzon M, Cordelli DM, Abdalla E, Accorsi P, Adam MP, Baldo C, Bayat A, Belligni E, Bonvicini F, Breckpot J, Callewaert B, Cocchi G, Cuturilo G, Devriendt K, Dinulos MB, Djuric O, Epifanio R, Faravelli F, Formisano D, Giordano L, Grasso M, Grønborg S, Iodice A, Iughetti L, Lacombe D, Maggi M, Malbora B, Mammi I, Moutton S, Møller R, Muschke P, Napoli M, Pantaleoni C, Pascarella R, Pellicciari A, Poch-Olive ML, Raviglione F, Rivieri F, Russo C, Savasta S, Scarano G, Selicorni A, Silengo M, Sorge G, Tarani L, Tone LG, Toutain A, Trimouille A, Valera ET, Vergano SS, Zanotta N, Zollino M, **Dobyns WB**, Paciorkowski AR. Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. *Genet Med*. 2017 Jun;19(6):691-700. doi: 10.1038/gim.2016.176. Epub 2016 Nov 10

Kurz JE, Smith CM, **Wainwright MS**. Thermoregulate, autoregulate and ventilate: brain-directed critical care for pediatric cardiac arrest. *Curr Opin Pediatr*. 2017 Jun;29(3):259-265. Doi: Di Donato N, Chiari S, Mirzaa GM, Aldinger K, Parrini E, Olds C, Barkovich AJ, Guerrini R, **Dobyns WB**. Lissencephaly: Expanded imaging and clinical classification. *Am J Med Genet A*. 2017 Jun;173(6):1473-1488. doi:10.1002/ajmg.a.38245. Epub 2017 Apr 25. 1097/MOP.0000000000000482.

Marashly A, Lew S, Koop J. Successful surgical management of New Onset Refractory Status Epilepticus (NORSE) presenting with gelastic seizures in a 3 year old girl. *Epilepsy Behav Case Rep*. 2017 May 22;8:18-26. doi: 10.1016/

Ducharme-Crevier L, Press CA, Kurz JE, Mills MG, Goldstein JL, **Wainwright MS**. Early Presence of Sleep Spindles on Electroencephalography Is Associated With Good Outcome After Pediatric Cardiac Arrest. *Pediatr Crit Care Med*. 2017 May;18(5):452-460. doi: 10.1097/PCC.0000000000001137

Dempsey JC, Phelps IG, Bachmann-Gagescu R, Glass IA, **Tully HM**, Doherty D. Mortality in Joubert syndrome. *Am J Med Genet A*. 2017 May;173(5):1237-1242. doi: 10.1002/ajmg.a.38158.

Oegema R, Baillat D, Schot R, van Unen LM, Brooks A, Kia SK, Hoogeboom AJM, Xia Z, Li W, Cesaroni M, Lequin MH, van Slegtenhorst M, **Dobyns WB**, de Coo IFM, Verheijen FW, Kremer A, van der Spek PJ, Heijnsman D, Wagner EJ, Fornerod M, Mancini GMS. Human mutations in integrator complex subunits link transcriptome integrity to brain development. *PLoS Genet*. 2017 May 25;13(5):e1006809. doi: 10.1371/journal.pgen.1006809. eCollection 2017 May. Erratum in: *PLoS Genet*. 2017 Aug 1;13(8):e1006923.

Carter JC, Wrede JE. Overview of Sleep and Sleep Disorders in Infancy and Childhood. *Pediatr Ann*. 2017 Apr 1;46(4):e133-e138.

Mahdi J, Shah AC, **Sato A**, Morris SM, McKinstry RC, Listernick R, Packer RJ, Fisher MJ, Gutmann DH. A multi-institutional study of brainstem gliomas in children with neurofibromatosis type 1. *Neurology*. 2017 Apr 18;88(16):1584-1589.

Blume HK. Neurological Disorders in Primary Care Pediatrics. *Pediatr Ann*. 2017 Apr 1;46(4):e131-e132.

Lee S, Mirsky DM, Beslow LA, **Amlie-Lefond C**, Danehy AR, Lehman L, Stence NV, Vossough A, Wintermark M, Rivkin MJ; International Paediatric Stroke Study Neuroimaging Consortium and the Paediatric Stroke Neuroimaging Consortium. Pathways for Neuroimaging of Neonatal Stroke. *Pediatr Neurol*. 2017 Apr;69:37-48. doi:10.1016/j.pediatrneurol.2016.12.008.

Pediatric Neurology Faculty Publications 2014-2025

Gospe SM Jr. Pyridoxine-Dependent Epilepsy. 2001 Dec 7 [updated 2017 Apr 13]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018.

Randle SC. Tuberous Sclerosis Complex: A Review. *Pediatr Ann.* 2017 Apr 1;46(4):e166-e171.

Moore CA, Staples JE, **Dobyns WB**, Pessoa A, Ventura CV, Fonseca EB, Ribeiro EM, Ventura LO, Neto NN, Arena JF, Rasmussen SA. Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. *JAMA Pediatr.* 2017 Mar 1;171(3):288-295. doi: 10.1001/jamapediatrics.2016.3982. Review

Natarajan N, Pardo AC. Challenges in neurologic prognostication after neonatal brain injury. *Semin Perinatol.* 2017 Mar;41(2):117-123. doi:10.1053/j.semperi.2016.11.008. Epub 2017 Jan 28. Review. PubMed PMID: 28139254.

Amie-Lefond C, Ojemann JG. Neonatal Hemorrhagic Stroke. *JAMA Pediatr.* 2017 Mar 1;171(3):220-221. doi: 10.1001/jamapediatrics.2016.4466

Lardelli RM, Schaffer AE, Eggens VR, Zaki MS, Grainger S, Sathe S, Van Nostrand EL, Schlachetzki Z, Rosti B, Akizu N, Scott E, Silhavy JL, Heckman LD, Rosti RO, Dikoglu E, Gregor A, Guemez-Gamboa A, Musaev D, Mande R, Widjaja A, Shaw TL, Markmiller S, Marin-Valencia I, Davies JH, de Meirleir L, Kayserili H, Altunoglu U, Freckmann ML, Warwick L, Chitayat D, Blaser S, Çağlayan AO, Bilguvar K, Per H, Fagerberg C, Christesen HT, Kibaek M, Aldinger KA, Manchester D, Matsumoto N, Muramatsu K, Saitsu H, Shiina M, Ogata K, Foulds N, **Dobyns WB**, Chi NC, Traver D, Spaccini L, Bova SM, Gabriel SB, Gunel M, Valente EM, Nassogne MC, Bennett EJ, Yeo GW, Baas F, Lykke-Andersen J, Gleeson JG. Biallelic mutations in the 3' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nat Genet.* 2017 Mar;49(3):457-464. doi: 10.1038/ng.3762. Epub 2017 Jan 16.

Bordini BJ, **Monrad P.** Differentiating Familial Neuropathies from Guillain-Barré Syndrome. *Pediatr Clin North Am.* 2017 Feb;64(1):231-252.

McDonald CL, **Saneto RP**, Carmant L, Sotero de Menezes MA. Focal Seizures in Patients With SCN1A Mutations. *J Child Neurol.* 2017 Feb;32(2):170-176. doi: 10.1177/0883073816672379

Beatty CW, Wrede JE, Blume HK. Diagnosis, treatment, and outcomes of infantile spasms in the Trisomy 21 population. *Seizure.* 2017 Feb;45:184-188. doi: 10.1016/j.seizure.2016.12.016..

Tuschl K, Clayton PT, **Gospe SM Jr**, Mills PB. Dystonia/Parkinsonism, Hypermanganesemia, Polycythemia, and Chronic Liver Disease. 2012 Aug 30 [updated 2017 Feb 9]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018.

Wenger TL, Chow P, **Randle SC**, Rosen A, Birgfeld C, **Wrede J**, Javid P, King D, Manh V, Hing AV, Albers E. Novel findings of left ventricular non-compaction cardiomyopathy, microform cleft lip and poor vision in patient with SMC1A-associated Cornelia de Lange syndrome. *Am J Med Genet A.* 2017 Feb;173(2):414-420.

Piao CS, Holloway AL, Hong-Routson S, **Wainwright MS.** Depression following traumatic brain injury in mice is associated with down-regulation of hippocampal astrocyte glutamate transporters by thrombin. *J Cereb Blood Flow Metab.* 2017 Jan 1:271678X17742792. doi: 10.

Brun BN, Mockler SR, Laubscher KM, Stephan CM, Wallace AM, Collison JA, Zimmerman MB, **Dobyns WB**, Mathews KD. Comparison of brain MRI findings with language and motor function in the dystroglycanopathies. *Neurology.* 2017 Feb 14;88(7):623-629. doi: 10.1212/WNL.0000000000003609. Epub 2017 Jan 13. .

Kannan N, Quistberg A, Wang J, Groner JI, Mink RB, **Wainwright MS**, Bell MJ, Giza CC, Zatzick DF, Ellenbogen RG, Boyle LN, Mitchell PH, Vavilala MS. Frequency of and factors associated with emergency department intracranial pressure monitor placement in severe paediatric traumatic brain injury. *Brain Inj.* 2017;31(13-14):1745-1752. doi: 10.1080/02699052.2017.

Gorman KM, **Wainwright MS.** Adult Stroke Screening Tool in Childhood Ischemic Stroke. *Pediatr Neurol Briefs.* 2017 Jan;31(1):3. doi:10.15844/pedneurbriefs-31-1-3.

Sánchez Fernández I, Jackson MC, Abend NS, Arya R, Brenton JN, Carpenter JL, Chapman KE, Gaillard WD, Gaínza-Lein M, Glauser TA, Goldstein JL, Goodkin HP, Helseth A, Kapur K, McDonough TL, Mikati MA, Peariso K, Riviello J Jr, Tasker RC, Topjian AA, **Wainwright MS**, Wilfong A, Williams K, Loddenkemper T; pediatric Status Epilepticus Research

Pediatric Neurology Faculty Publications 2014-2025

Group (pSERG). Refractory status epilepticus in children with and without prior epilepsy or status epilepticus. *Neurology*. 2017 Jan 24;88(4):386-394. doi: 10.1212/WNL.0000000000003550.

Press CA, Morgan L, Mills M, Stack CV, Goldstein JL, Alonso EM, **Wainwright MS**. Spectral Electroencephalogram Analysis for the Evaluation of Encephalopathy Grade in Children With Acute Liver Failure. *Pediatr Crit Care Med*. 2017 Jan;18(1):64-72. doi: 10.1097/PCC.0000000000001016

Haldipur P, Dang D, Aldinger KA, Janson OK, Guimiot F, Adle-Biasette H, **Dobyns WB**, Siebert JR, Russo R, Millen KJ. Phenotypic outcomes in Mouse and HumanFoxc1 dependent Dandy-Walker cerebellar malformation suggest shared mechanisms. *Elife*. 2017 Jan 16;6. pii: e20898. doi: 10.7554/eLife.20898

Byers HM, Adam MP, LaCroix A, Leary SE, Cole B, **Dobyns WB**, Mefford HC. Description of a new oncogenic mechanism for atypical teratoid rhabdoid tumors in patients with ring chromosome 22. *Am J Med Genet A*. 2017 Jan;173(1):245-249. doi: 10.1002/ajmg.a.37993. Epub 2016 Oct 12

Patel, P., Baier J, Baranov, et al. Health Beliefs Regarding Pediatric Cerebral Palsy Among Caregivers in Botswana: A Qualitative Study. *Child Care Health Dev*. 43(6):861-868, 2017

Farhadian S., **Patel P.**, Spudich S., Neurological Complications of HIV. *Curr Infect Dis Rep*. 19(12): 50, 2017

2016

Ducharme-Crevier L, Mills MG, Mehta PM, Smith CM, **Wainwright MS**. Use of Transcranial Doppler for Management of Central Nervous System Infections in Critically Ill Children. *Pediatr Neurol*. 2016 Dec;65:52-58.e2. doi: 10.1016/j.pediatrneurol.2016.08.027.

Johnson SC, Kaeberlein M: Rapamycin in aging and disease: maximizing efficacy while minimizing side effects. *Oncotarget*. 2016; 7:p44876-44878. PMID: 27384492.

Teumer A, Qi Q, Nethander M, Aschard H, Bandinelli S, Beekman M, Berndt SI, Bidlingmaier M, Broer L; CHARGE Longevity Working Group, Cappola A, Ceda GP, Chanock S, Chen MH, Chen TC, Chen YI, Chung J, Del Greco Miglianico F, Eriksson J, Ferrucci L, Friedrich N, Gnewuch C, Goodarzi MO, Grarup N, Guo T, Hammer E, Hayes RB, Hicks AA, Hofman A, Houwing-Duistermaat JJ, Hu F, Hunter DJ, Husemoen LL, Isaacs A, Jacobs KB, Janssen JA, Jansson JO, Jehmlich N, **Johnson S**, Juul A, Karlsson M, Kilpelainen TO, Kovacs P, Kraft P, Li C, Linneberg A, Liu Y, Loos RJ; Body Composition Genetics Consortium, Lorentzon M, Lu Y, Maggio M, Magi R, Meigs J, Mellström D, Nauck M, Newman AB, Pollak MN, Pramstaller PP, Prokopenko I, Psaty BM, Reincke M, Rimm EB, Rotter JI, Saint Pierre A, Schurmann C, Seshadri S, Sjögren K, Slagboom PE, Strickler HD, Stumvoll M, Suh Y, Sun Q, Zhang C, Svensson J, Tanaka T, Tare A, Tönjes A, Uh HW, van Duijn CM, van Heemst D, Vandenput L, Vasan R, Völker U, Willems SM, Ohlsson C, Wallaschofski H, Kaplan R: Genomewide meta-analysis identifies loci associated with IGF-I and IGFBP-3 levels with impact on age-related traits. *Aging Cell*. 2016. 15(5):p811-24. PMID: 27329260.

Mohammad S, Wolfe LA, Stöbe P, Biskup S, **Wainwright MS**, Melin-Aldana H, Malladi P, Muenke M, Gahl WA, Whittington PF. Infantile Cirrhosis, Growth Impairment, and Neurodevelopmental Anomalies Associated with Deficiency of PPP1R15B. *J Pediatr*. 2016 Dec;179:144-149.e2. doi: 10.1016/j.jpeds.2016.08.043

Leto K, Arancillo M, Becker EB, Buffo A, Chiang C, Ding B, **Dobyns WB**, Dusart I, Haldipur P, Hatten ME, Hoshino M, Joyner AL, Kano M, Kilpatrick DL, Koibuchi N, Marino S, Martinez S, Millen KJ, Millner TO, Miyata T, Parmigiani E, Schilling K, Sekerková G, Sillitoe RV, Sotelo C, Uesaka N, Wefers A, Wingate RJ, Hawkes R. Consensus Paper: Cerebellar Development. *Cerebellum*. 2016 Dec;15(6):789-828.Review

Rivkin MJ, Bernard TJ, Dowling MM, **Amlie-Lefond C**. Corrigendum to 'Guidelines for Urgent Management of Stroke in Children' [Pediatric Neurology 56 (2016)8-17]. *Pediatr Neurol*. 2016 Nov;64:105. doi: 10.1016/j.pediatrneurol.2016.08.019. Epub 2016 Sep 1.

Steinman KJ, Spence SJ, Ramocki MB, Proud MB, Kessler SK, Marco EJ, Green Snyder L, D'Angelo D, Chen Q, Chung WK, Sherr EH; Simons VIP Consortium. 16p11.2 deletion and duplication: Characterizing neurologic phenotypes in a large clinically ascertained cohort. *Am J Med Genet A*. 2016 Nov;170(11):2943-2955. doi:10.1002/ajmg.a.37820.

Pediatric Neurology Faculty Publications 2014-2025

Kim J, Liao YH, **Ionita C**, Bale AE, Darras B, Acsadi G. Mitochondrial Membrane Protein-Associated Neurodegeneration Mimicking Juvenile Amyotrophic Lateral Sclerosis. *Pediatr Neurol*. 2016 Nov;64:83-86. doi:10.1016/j.pediatrneurol.2016.08.013.

Ravenscroft G, Di Donato N, Hahn G, Davis MR, Craven PD, Poke G, Neas KR, Neuhann TM, **Dobyns WB**, Laing NG. Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. *Neuromuscul Disord*. 2016 Nov;26(11):744-748. doi: 10.1016/j.nmd.2016.09.009. Epub 2016 Sep 19.

Adams Waldorf KM, Stencel-Baerenwald JE, Kapur RP, Studholme C, Boldenow E, Vornhagen J, Baldessari A, Dighe MK, Thiel J, Merillat S, Armistead B, Tisoncik-Go J, Green RR, Davis MA, Dewey EC, Fairgrieve MR, Gatenby JC, Richards T, Garden GA, Diamond MS, Juul SE, Grant RF, Kuller L, Shaw DW, Ogle J, Gough GM, Lee W, English C, Hevner RF, **Dobyns WB**, Gale M Jr, Rajagopal L. Fetal brain lesions after subcutaneous inoculation of Zika virus in a pregnant nonhuman primate. *Nat Med*. 2016 Nov;22(11):1256-1259. doi: 10.1038/nm.4193. Epub 2016 Sep

Ma M, Adams HR, Seltzer LE, **Dobyns WB**, Paciorkowski AR. Phenotype Differentiation of FOXP1 and MECP2 Disorders: A New Method for Characterization of Developmental Encephalopathies. *J Pediatr*. 2016 Nov;178:233-240.e10. doi:10.1016/j.jpeds.2016.08.032. Epub 2016 Sep 15.

Di Donato N, Jean YY, Maga AM, Krewson BD, Shupp AB, Avrutsky MI, Roy A, Collins S, Olds C, Willert RA, Czaja AM, Johnson R, Stover JA, Gottlieb S, Bartholdi D, Rauch A, Goldstein A, Boyd-Kyle V, Aldinger KA, Mirzaa GM, Nissen A, Brigatti KW, Puffenberger EG, Millen KJ, Strauss KA, **Dobyns WB**, Troy CM, Jinks RN. Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. *Am J Hum Genet*. 2016 Nov 3;99(5):1117-1129. doi: 10.1016/j.ajhg.2016.09.010. Epub 2016 Oct 20

Tully HM, Wenger TL, Kukull WA, Doherty D, **Dobyns WB**. Anatomical configurations associated with posthemorrhagic hydrocephalus among premature infants with intraventricular hemorrhage. *Neurosurg Focus*. 2016 Nov;41(5):E5.

Garzon MC, Epstein LG, Heyer GL, Frommelt PC, Orbach DB, Baylis AL, Blei F, Burrows PE, Chamlin SL, Chun RH, Hess CP, Joachim S, Johnson K, Kim W, Liang MG, Maheshwari M, McCoy GN, Metry DW, **Monrad PA**, Pope E, Powell J, Shwayder TA, Siegel DH, Tollefson MM, Vadivelu S, Lew SM, Frieden IJ, Drolet BA. PHACE Syndrome: Consensus-Derived Diagnosis and Care Recommendations. *J Pediatr*. 2016 Nov;178:24-33.e2.

Bernard TJ, Beslow LA, Manco-Johnson MJ, Armstrong-Wells J, Boada R, Weitzenkamp D, Hollatz A, Poisson S, **Amalie-Lefond C**, Lo W, deVeber G, Goldenberg NA, Dowling MM, Roach ES, Fullerton HJ, Benseler SM, Jordan LC, Kirton A, Ichord RN. Inter-Rater Reliability of the CASCADE Criteria: Challenges in Classifying Arteriopathies. *Stroke*. 2016 Oct;47(10):2443-9. doi:10.1161/STROKEAHA.116.013544. Epub 2016 Sep 15.

Tasker RC, Goodkin HP, Sánchez Fernández I, Chapman KE, Abend NS, Arya R, Brenton JN, Carpenter JL, Gaillard WD, Glauser TA, Goldstein J, Helseth AR, Jackson MC, Kapur K, Mikati MA, Peariso K, **Wainwright MS**, Wilfong AA, Williams K, Loddenkemper T; Pediatric Status Epilepticus Research Group. Refractory Status Epilepticus in Children: Intention to Treat With Continuous Infusions of Midazolam and Pentobarbital. *Pediatr Crit Care Med*. 2016 Oct;17(10):968-975

Buckley RT, Wang AC, Miller JW, **Novotny EJ**, Ojemann JG. Stereotactic laser ablation for hypothalamic and deep intraventricular lesions. *Neurosurg Focus*. 2016 Oct;41(4):E10.

Ostahowski PJ, Kannan N, **Wainwright MS**, Qiu Q, Mink RB, Groner JI, Bell MJ, Giza CC, Zatzick DF, Ellenbogen RG, Boyle LN, Mitchell PH, Vavilala MS; PEGASUS (Pediatric Guideline Adherence and Outcomes) Study. Variation in seizure prophylaxis in severe pediatric traumatic brain injury. *J Neurosurg Pediatr*. 2016 Oct;18(4):499-506.

Weisleder P, **Gospe SM Jr**, Ng YT, Sahin M. The Pediatric Neurology Trainee Publication Award for 2015. *Pediatr Neurol*. 2016 Oct;63:1-2. doi: 10.1016/j.pediatrneurol.2016.07.004.

Gripp KW, Aldinger KA, Bennett JT, Baker L, Tusi J, Powell-Hamilton N, Stabley D, Sol-Church K, Timms AE, **Dobyns WB**. A novel rasopathy caused by recurrent de novo missense mutations in PPP1CB closely resembles Noonan syndrome with loose anagen hair. *Am J Med Genet A*. 2016 Sep;170(9):2237-47. doi:10.1002/ajmg.a.37781.

Parrini E, Conti V, **Dobyns WB**, Guerrini R. Genetic Basis of Brain Malformations. *Mol Syndromol*. 2016 Sep;7(4):220-233. Epub 2016 Aug 27. Review.

Pediatric Neurology Faculty Publications 2014-2025

Natarajan N, Tully HM, Chapman T. Prenatal presentation of pyruvate dehydrogenase complex deficiency. *Pediatr Radiol*. 2016 Aug; 46(9):1354-7.

Tuschl K, Clayton PT, **Gospe SM Jr**, Gulab S, Ibrahim S, Singhi P, Aulakh R, Ribeiro RT, Barsottini OG, Zaki MS, Del Rosario ML, Dyack S, Price V, Rideout A, Gordon K, Wevers RA, Chong WK, Mills PB. Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10, a Manganese Transporter in Man. *Am J Hum Genet*. 2016 Aug 4;99(2):521. doi: 10.1016/j.ajhg.2016.07.015.

Tsuchida TN, Acharya JN, Halford JJ, **Kuratani JD**, Sinha SR, Stecker MM, Tatum WO 4th, Drislane FW. American Clinical Neurophysiology Society: EEG Guidelines Introduction. *J Clin Neurophysiol*. 2016 Aug;33(4):301-2. doi: 10.1097/WNP.0000000000000315.

Mossa-Basha M, de Havenon A, Becker KJ, Hallam DK, Levitt MR, Cohen WA, Hippe DS, Alexander MD, Tirschwell DL, Hatsukami T, **Amlie-Lefond C**, Yuan C. Added Value of Vessel Wall Magnetic Resonance Imaging in the Differentiation of Moyamoya Vasculopathies in a Non-Asian Cohort. *Stroke*. 2016 Jul;47(7):1782-8. doi: 10.1161/STROKEAHA.116.013320

Amlie-Lefond C, Gilden D. Varicella Zoster Virus: A Common Cause of Stroke in Children and Adults. *J Stroke Cerebrovasc Dis*. 2016 Jul;25(7):1561-1569. doi:10.1016/j.jstrokecerebrovasdis.2016.03.052. Epub 2016 Apr 29. Review

Mirzaa GM, Campbell CD, Solovieff N, Goold C, Jansen LA, Menon S, Timms AE, Conti V, Biag JD, Adams C, Boyle EA, Collins S, Ishak G, Poliachik S, Girisha KM, Yeung KS, Chung BHY, Rahikkala E, Gunter SA, McDaniel SS, Macmurdo CF, Bernstein JA, Martin B, Leary R, Mahan S, Liu S, Weaver M, Doerschner M, Jhangiani S, Muzny DM, Boerwinkle E, Gibbs RA, Lupski JR, Shendure J, **Saneto RP, Novotny EJ**, Wilson CJ, Sellers WR, Morrissey M, Hevner RF, Ojemann JG, Guerrini R, Murphy LO, Winckler W, **Dobyns WB**. Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurol*. 2016 Jul 1;73(7):836-845. doi: 10.1001/jamaneurol.2016.0363.

Casimo K, Darvas F, Wander J, Ko A, Grabowski TJ, **Novotny E**, Poliakov A, Ojemann JG, Weaver KE. Regional Patterns of Cortical Phase Synchrony in the Resting State. *Brain Connect*. 2016 Jul;6(6):470-81. doi: 10.1089/brain.2015.0362. Epub 2016 May 2.

Parikh S, Karaa A, Goldstein A, Ng YS, Gorman G, Feigenbaum A, Christodoulou J, Haas R, Tarnopolsky M, Cohen BK, Dimmock D, Feyma T, Koenig MK, Mundy H, Niyazov D, **Saneto RP, Wainwright MS**, Wusthoff C, McFarland R, Scaglia F. Solid organ transplantation in primary mitochondrial disease: Proceed with caution. *Mol Genet Metab*. 2016 Jul;118(3):178-84. doi: 10.1016/j.ymgme.2016.04.009

Saneto RP. Alpers-Huttenlocher syndrome: the role of a multidisciplinary health care team. *J Multidiscip Healthc*. 2016 Jul 26;9:323-33. doi:10.2147/JMDH.S84900. eCollection 2016. Review.

Kurz JE, Poloyac SM, Abend NS, Fabio A, Bell MJ, **Wainwright MS**; Investigators for the Approaches and Decisions in Acute Pediatric TBI Trial. Variation in Anticonvulsant Selection and Electroencephalographic Monitoring Following Severe Traumatic Brain Injury in Children-Understanding Resource Availability in Sites Participating in a Comparative Effectiveness Study. *Pediatr Crit Care Med*. 2016 Jul;17(7):649-57. doi: 10.1097/PCC.0000000000000765.

Byers HM, **Beatty CW**, Hahn SH, **Gospe SM Jr**. Dramatic Response After Lamotrigine in a Patient With Epileptic Encephalopathy and a De Novo CACNA1A Variant. *Pediatr Neurol*. 2016 Jul;60:79-82. doi: 10.1016/j.pediatrneurol.2016.03.012. Epub 2016 Apr 1.

Twigg SRF, Hufnagel RB, Miller KA, Zhou Y, McGowan SJ, Taylor J, Craft J, Taylor JC, Santoro SL, Huang T, Hopkin RJ, Brady AF, Clayton-Smith J, Clericuzio CL, Grange DK, Groesser L, Hafner C, Horn D, Temple IK, **Dobyns WB**, Curry CJ, Jones MC, Wilkie AOM. A Recurrent Mosaic Mutation in SMO, Encoding the Hedgehog Signal Transducer Smoothed, Is the Major Cause of Curry-Jones Syndrome. *Am J Hum Genet*. 2016 Jun 2;98(6):1256-1265. doi: 10.1016/j.ajhg.2016.04.007. Epub 2016 May 26.

Mirzaa G, Timms AE, Conti V, Boyle EA, Girisha KM, Martin B, Kircher M, Olds C, Juusola J, Collins S, Park K, Carter M, Glass I, Krägeloh-Mann I, Chitayat D, Parikh AS, Bradshaw R, Torti E, Braddock S, Burke L, Ghedia S, Stephan M, Stewart F, Prasad C, Napier M, Saitta S, Straussberg R, Gabbett M, O'Connor BC, Keegan CE, Yin LJ, Lai AH, Martin N, McKinnon M, Addor MC, Boccuto L, Schwartz CE, Lanoel A, Conway RL, Devriendt K, Tatton-Brown K, Pierpont ME, Painter M, Worgan L, Reggin J, Hennekam R, Tsuchiya K, Pritchard CC, Aracena M, Gripp KW, Cordisco M, Van Esch H,

Pediatric Neurology Faculty Publications 2014-2025

Garavelli L, Curry C, Gorieli A, Kayserilli H, Shendure J, Graham J Jr, Guerrini R, **Dobyns WB**. PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight*. 2016 Jun 16;1(9). pii: e87623.

Di Donato N, Neuhaus T, Kahlert AK, Klink B, Hackmann K, Neuhaus I, Novotna B, Schallner J, Krause C, Glass IA, Parnell SE, Benet-Pages A, Nissen AM, Berger W, Altmüller J, Thiele H, Weber BH, Schrock E, **Dobyns WB**, Bier A, Rump A. Mutations in EXOSC2 are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. *J Med Genet*. 2016 Jun;53(6):419-25. doi: 10.1136/jmedgenet-2015-103511.

Aldinger KA, Mendelsohn NJ, Chung BH, Zhang W, Cohn DH, Fernandez B, Alkuraya FS, **Dobyns WB**, Curry CJ. Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive WNT1 mutations. *J Med Genet*. 2016 Jun;53(6):427-30. doi:10.1136/jmedgenet-2015-103476

Tully HM, Kukull WA, Mueller BA. Clinical and Surgical Factors Associated With Increased Epilepsy Risk in Children With Hydrocephalus. *Pediatr Neurol*. 2016 Jun;59:18-22. doi: 10.1016/j.pediatrneurol.2016.02.011

Chi DL, Momany ET, Mancl LA, Lindgren SD, Zinner SH, **Steinman KJ**. Dental Homes for Children With Autism: A Longitudinal Analysis of Iowa Medicaid's I-Smile Program. *Am J Prev Med*. 2016 May;50(5):609-615. doi:10.1016/j.amepre.2015.08.022. Epub 2015 Oct 26.

Hong CS, Wang AC, Bonow RH, Abecassis IJ, **Amlie-Lefond C**, Ellenbogen RG. Moyamoya Disease in a Patient with VACTERL Association. *World Neurosurg*. 2016 May;89:729.e7-729.e10. doi: 10.1016/j.wneu.2016.01.059. Epub 2016 Feb 2.

Bernier R, **Steinman KJ**, Reilly B, Wallace AS, Sherr EH, Pojman N, Mefford HC, Gerds J, Earl R, Hanson E, Goin-Kochel RP, Berry L, Kanne S, Snyder LG, Spence S, Ramocki MB, Evans DW, Spiro JE, Martin CL, Ledbetter DH, Chung WK; Simons VIP consortium. Clinical phenotype of the recurrent 1q21.1 copy-number variant. *Genet Med*. 2016 Apr;18(4):341-9. doi: 10.1038/gim.2015.78.

Alqahtani MF, Smith CM, Weiss SL, Dawson S, Ralay Ranaivo H, **Wainwright MS**. Evaluation of New Diagnostic Biomarkers in Pediatric Sepsis: Matrix Metalloproteinase-9, Tissue Inhibitor of Metalloproteinase-1, Mid-Regional Pro-Atrial Natriuretic Peptide, and Adipocyte Fatty-Acid Binding Protein. *PLoS One*. 2016 Apr 18;11(4):e0153645. doi: 10.1371/journal.pone.0153645. eCollection 2016..

Lockrow J, Longstreth W, Davis AP. Intracranial Aneurysms From Presumed Infective Endocarditis: The Dilemma of Persistently Negative Cultures. *Neurohospitalist*. 2016 Apr;6(2):80-6.. Epub 2015 Sep 21

Bernard TJ, Friedman NR, Stence NV, Jones W, Ichord R, **Amlie-Lefond C**, Dowling MM, Rivkin MJ. Preparing for a "Pediatric Stroke Alert". *Pediatr Neurol*. 2016 Mar;56:18-24. doi: 10.1016/j.pediatrneurol.2015.10.012. Epub 2015 Dec 11. Review.

Bennett JT, Tan TY, Alcantara D, Tétrault M, Timms AE, Jensen D, Collins S, Nowaczyk MJM, Lindhurst MJ, Christensen KM, Braddock SR, Brandling-Bennett H, Hennekam RCM, Chung B, Lehman A, Su J, Ng S, Amor DJ; University of Washington Center for Mendelian Genomics; Care4Rare Canada Consortium, Majewski J, Biesecker LG, Boycott KM, **Dobyns WB**, O'Driscoll M, Moog U, McDonnell LM. Mosaic Activating Mutations in FGFR1 Cause cephalocraniocutaneous Lipomatosis. *Am J Hum Genet*. 2016 Mar 3;98(3):579-587. doi: 10.1016/j.ajhg.2016.02.006

Rivkin MJ, Bernard TJ, Dowling MM, **Amlie-Lefond C**. Guidelines for Urgent Management of Stroke in Children. *Pediatr Neurol*. 2016 Mar;56:8-17. doi:10.1016/j.pediatrneurol.2016.01.016. Epub 2016 Jan 21. Review. Erratum in: *Pediatr Neurol*. 2016 Nov;64:105.

Tully HM, Ishak GE, Rue TC, Dempsey JC, Browd SR, Millen KJ, Doherty D, **Dobyns WB**. Two Hundred Thirty-Six Children With Developmental Hydrocephalus: Causes and Clinical Consequences. *J Child Neurol*. 2016 Mar;31(3):309-20. doi:10.1177/0883073815592222

Cohen AS, Yap DB, Lewis ME, Chijiwa C, Ramos-Arroyo MA, Tkachenko N, Milano V, Fradin M, McKinnon ML, Townsend KN, Xu J, Van Allen MI, Ross CJ, **Dobyns WB**, Weaver DD, Gibson WT. Weaver Syndrome-Associated EZH2 Protein Variants Show Impaired Histone Methyltransferase Function In Vitro. *Hum Mutat*. 2016 Mar;37(3):301-7. doi: 10.1002/humu.22946. Epub 2016 Jan 12.

Pediatric Neurology Faculty Publications 2014-2025

- Di Donato N, Rump A, Mirzaa GM, Alcantara D, Oliver A, Schrock E, **Dobyns WB**, O'Driscoll M. Identification and Characterization of a Novel Constitutional PIK3CA Mutation in a Child Lacking the Typical Segmental Overgrowth of "PIK3CA-Related Overgrowth Spectrum". *Hum Mutat.* 2016 Mar;37(3):242-5. doi:10.1002/humu.22933. Epub 2015 Dec 15.
- Elkind MS, Hills NK, Glaser CA, Lo WD, **Amlie-Lefond C**, Dlamini N, Kneen R, Hod EA, Wintermark M, deVeber GA, Fullerton HJ; VIPS Investigators*. Herpesvirus Infections and Childhood Arterial Ischemic Stroke: Results of the VIPS Study. *Circulation.* 2016 Feb 23;133(8):732-41. doi: 10.1161/CIRCULATIONAHA.115.018595. Epub 2016 Jan 26.
- Marin SE, **Saneto RP**. Neuropsychiatric Features in Primary Mitochondrial Disease. *Neurol Clin.* 2016 Feb;34(1):247-94. doi: 10.1016/j.ncl.2015.08.011. Review. PubMed PMID: 26614002.
- Storey GP, Gonzalez-Fernandez G, Bamford IJ, Hur M, McKinley JW, Heimbigner L, Minasyan A, Walwyn WM, **Bamford NS**. Nicotine Modifies Corticostriatal Plasticity and Amphetamine Rewarding Behaviors in Mice(1,2,3). *eNeuro.* 2016 Feb 2;3(1). pii:ENEURO.0095-15.2015. doi: 10.1523/ENEURO.0095-15.2015. eCollection 2016 Jan-Feb
- Burns TM, Smith GA, Allen JA, Amato AA, Arnold WD, Barohn R, Benatar M, Bird SJ, Bromberg M, Chahin N, Cialfoni E, Cohen JA, Corse A, Crum BA, David WS, Dimberg E, Sousa EA, Donofrio PD, Dyck PJ, Engel AG, Ensrud ER, Ferrante M, Freimer M, Gable KL, Gibson S, Gilchrist JM, Goldstein JM, Gooch CL, Goodman BP, Gorelov D, **Gospe SM Jr**, Goyal NA, Guidon AC, Guptill JT, Gutmann L, Gutmann L, Gwathmey K, Harati Y, Harper CM Jr, Hehir MK, Hobson-Webb LD, Howard JF Jr, Jackson CE, Johnson N, Jones SM, Juel VC, Kaminski HJ, Karam C, Kennelly KD, Khella S, Khoury J, Kincaid JC, Kissel JT, Kolb N, Lacomis D, Ladha S, Larriviere D, Lewis RA, Li Y, Litchy WJ, Logigian E, Lou JS, MacGowen DJ, Maselli R, Massey JM, Mauermann ML, Mathews KD, Meriggioli MN, Miller RG, Moon JS, Mozaffar T, Nations SP, Nowak RJ, Ostrow LW, Pascuzzi RM, Peltier A, Ruzhansky K, Richman DP, Ross MA, Rubin DI, Russell JA, Sachs GM, Salajegheh MK, Saperstein DS, Scelsa S, Selcen D, Shaibani A, Shieh PB, Silvestri NJ, Singleton JR, Smith BE, So YT, Solorzano G, Sorenson EJ, Srinivasen J, Tavee J, Tawil R, Thaisetthawatkul P, Thornton C, Trivedi J, Vernino S, Wang AK, Webb TA, Weiss MD, Windebank AJ, Wolfe GI. Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. *Muscle Nerve.* 2016 Feb;53(2):165-8. doi: 10.1002/mus.25009..
- Berg AT, **Dobyns WB**. Progress in autism research and postgenomic studies -Authors' reply. *Lancet Neurol.* 2016 Feb;15(2):136-137. doi:10.1016/S1474-4422(15)00403-2. Epub 2016 Jan 12
- Huang L, Vanstone MR, Hartley T, Osmond M, Barrowman N, Allanson J, Baker L, Dabir TA, Dipple KM, **Dobyns WB**, Estrella J, Faghfoury H, Favaro FP, Goel H, Gregersen PA, Gripp KW, Grix A, Guion-Almeida ML, Harr MH, Hudson C, Hunter AG, Johnson J, Joss SK, Kimball A, Kini U, Kline AD, Lauzon J, Lildballe DL, López-González V, Martinezmoles J, Meldrum C, Mirzaa GM, Morel CF, Morton JE, Pyle LC, Quintero-Rivera F, Richer J, Scheuerle AE, Schönewolf-Greulich B, Shears DJ, Silver J, Smith AC, Temple IK; UCLA Clinical Genomics Center, van de Kamp JM, van Dijk FS, Vandersteen AM, White SM, Zackai EH, Zou R; Care4Rare Canada Consortium, Bulman DE, Boycott KM, Lines MA. Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. *Hum Mutat.* 2016 Feb;37(2):148-54. doi: 10.1002/humu.22924. Epub 2015 Nov 19. Review
- Amlie-Lefond C**, Shaw D. Vascular disease. *Handb Clin Neurol.* 2016;136:1159-71. doi: 10.1016/B978-0-444-53486-6.00060-0. Review.
- Amlie-Lefond C**. Dissecting etiologies of posterior circulation stroke. *Dev Med Child Neurol.* 2016 Jan;58(1):10-1. doi: 10.1111/dmcn.12969
- Olson JD, Wander JD, Johnson L, Sarma D, Weaver K, **Novotny EJ**, Ojemann JG, Darvas F. Comparison of subdural and subgaleal recordings of cortical high-gamma activity in humans. *Clin Neurophysiol.* 2016 Jan;127(1):277-284. doi:10.1016/j.clinph.2015.03.014.
- D'Angelo D, Lebon S, Chen Q, Martin-Brevet S, Snyder LG, Hippolyte L, Hanson E, Maillard AM, Faucett WA, Macé A, Pain A, Bernier R, Chawner SJ, David A, Andrieux J, Aylward E, Baujat G, Caldeira I, Conus P, Ferrari C, Forzano F, Gérard M, Goin-Kochel RP, Grant E, Hunter JV, Isidor B, Jacqueline A, Jønhc AE, Keren B, Lacombe D, Le Caignec C, Martin CL, Männik K, Metspalu A, Mignot C, Mukherjee P, Owen MJ, Passeggeri M, Rooryck-Thambo C, Rosenfeld JA, Spence SJ, **Steinman KJ**, Tjernagel J, Van Haelst M, Shen Y, Draganski B, Sherr EH, Ledbetter DH, van den Bree MB, Beckmann JS, Spiro JE, Raymond A, Jacquemont S, Chung WK; Cardiff University Experiences of Children With Copy Number Variants (ECHO) Study; 16p11.2 European Consortium; Simons Variation in Individuals Project (VIP) Consortium. Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry.* 2016 Jan;73(1):20-30. doi: 10.1001/jamapsychiatry.2015.2123

Pediatric Neurology Faculty Publications 2014-2025

Poliachik SL, Friedman SD, Poliakov AV, Budech CB, Ishak GE, Shaw DW, **Gospe SM Jr.** Corpus Callosum Diffusion and Connectivity Features in High Functioning Subjects With Pyridoxine-Dependent Epilepsy. *Pediatr Neurol.* 2016 Jan;54:43-8. doi: 10.1016/j.pediatrneurol.2015.09.012.

Tsuchida TN, Acharya JN, Halford JJ, **Kuratani JD**, Sinha SR, Stecker MM, Tatum WO, Drislane FW. American Clinical Neurophysiology Society: EEG Guidelines Introduction. *Neurodiagn J.* 2016;56(4):231-234. doi:10.1080/21646821.2016.1245513.

2015

McCormick MA, Delaney JR, Tsuchiya M, Tsuchiyama S, Shemorry A, Sim S, Chou AC, Ahmed U, Carr D, Murakami CJ, Schleit J, Sutphin GL, Wasko BM, Bennett CF, Wang AM, Olsen B, Beyer RP, Bammler TK, Prunkard D, **Johnson SC**, Pennypacker JK, An E, Anies A, Castanza AS, Choi E, Dang N, Enerio S, Fletcher M, Fox L, Goswami S, Higgins SA, Holmberg MA, Hu D, Hui J, Jelic M, Jeong KS, Johnston E, Kerr EO, Kim J, Kim D, Kirkland K, Klum S, Kotireddy S, Liao E, Lim M, Lin MS, Lo WC, Lockshon D, Miller HA, Moller RM, Muller B, Oakes J, Pak DN, Peng ZJ, Pham KM, Pollard TG, Pradeep P, Pruett D, Rai D, Robison B, Rodriguez AA, Ros B, Sage M, Singh MK, Smith ED, Snead K, Solanky A, Spector BL, Steffen KK, Tchao BN, Ting MK, Vander Wende H, Wang D, Welton KL, Westman EA, Brem RB, Liu XG, Suh Y, Zhou Z, Kaeberlein M, Kennedy BK: A Comprehensive Analysis of Replicative Lifespan in 4,698 Single-Gene Deletion Strains Uncovers Conserved Mechanisms of Aging. *Cell Metab.* 2015. 22(5):p895-906. PMID: 26456335.

Johnson SC, Yanos ME, Bitto A, Castanza A, Gagnidze A, Gonzalez B, Gupta K, Hui J, Jarvie C, Johnson BM, Letexier N, McCanta L, Sangesland M, Tamis O, Uhde L, Van Den Ende A, Rabinovitch PS, Suh Y, Kaeberlein M: Dose-dependent Effects of mTOR Inhibition on Weight and Mitochondrial Disease in Mice. *Front. Genet.* 2015. 6:p247. PMID: 26257774.

Amlic-Lefond C, Rivkin MJ, Friedman NR, Bernard TJ, Dowling MM, deVeber G. The Way Forward: Challenges and Opportunities in Pediatric Stroke. *Pediatr Neurol.* 2016 Mar;56:3-7. doi: 10.1016/j.pediatrneurol.2015.10.021. Epub 2015 Dec 10. PubMed PMID: 26803334.

Schwarz AC, **Amlic-Lefond C**. A 15-Year-Old Boy With Trisomy 21 and Postoperative Weakness. *JAMA Pediatr.* 2016 Jan;170(1):85-6. doi:10.1001/jamapediatrics.2015.1677.

Crealey M, Allen NM, Webb D, **Bouldin A**, Mc Sweeney N, Peake D, Tirupathi S, Butler K, King MD. Sydenham's chorea: not gone but perhaps forgotten. *Arch Dis Child.* 2015 Dec;100(12):1160-2. doi: 10.1136/archdischild-2015-308693. Epub 2015 Sep 15

Roy A, Skibo J, Kalume F, Ni J, Rankin S, Lu Y, **Dobyns WB**, Mills GB, Zhao JJ, Baker SJ, Millen KJ. Mouse models of human PIK3CA-related brain overgrowth have acutely treatable epilepsy. *Elife.* 2015 Dec 3;4. pii: e12703. doi:10.7554/eLife.12703..

Mirzaa GM, Conti V, Timms AE, Smyser CD, Ahmed S, Carter M, Barnett S, Hufnagel RB, Goldstein A, Narumi-Kishimoto Y, Olds C, Collins S, Johnston K, Deleuze JF, Nitschké P, Friend K, Harris C, Goetsch A, Martin B, Boyle EA, Parrini E, Mei D, Tattini L, Slavotinek A, Blair E, Barnett C, Shendure J, Chelly J, **Dobyns WB**, Guerrini R. Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. *Lancet Neurol.* 2015 Dec;14(12):1182-95. doi: 10.1016/S1474-4422(15)00278-1. Epub 2015 Oct 29.

McMahon KQ, Papandreou A, Ma M, Barry BJ, Mirzaa GM, **Dobyns WB**, Scott RH, Trump N, Kurian MA, Paciorkowski AR. Familial recurrences of FOXP1-related disorder: Evidence for mosaicism. *Am J Med Genet A.* 2015 Dec;167A(12):3096-102. doi: 10.1002/ajmg.a.37353. Epub 2015 Sep 14.

Miller DT, Chung W, Nasir R, Shen Y, **Steinman KJ**, Wu BL, Hanson E. 16p11.2 Recurrent Microdeletion. 2009 Sep 22 [updated 2015 Dec 10]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018.

Pediatric Neurology Faculty Publications 2014-2025

Mirzaa GM, Collins S, **Dobyns WB**. Corrigendum to "Congenital microcephaly and chorioretinopathy due to de novo heterozygous KIF11 mutations: Five novel mutations and review of the literature. Am J Med Genet Part A 2014 164A:2879-86". Am J Med Genet A. 2015 Nov 14. doi: 10.1002/ajmg.a.37449. [

Shurtleff HA, Barry D, Firman T, **Warner MH**, Aguilar-Estrada RL, **Saneto RP**, **Kuratani JD**, Ellenbogen RG, **Novotny EJ**, Ojemann JG. Impact of epilepsy surgery on development of preschool children: identification of a cohort likely to benefit from early intervention. J Neurosurg Pediatr. 2015 Oct;16(4):383-92. doi:10.3171/2015.3.PEDS14359

Wrede JE, Mengel-From J, Buchwald D, Vitiello MV, Bamshad M, Noonan C, Christiansen L, Christensen K, Watson NF. Mitochondrial DNA Copy Number in Sleep Duration Discordant Monozygotic Twins. Sleep. 2015 Oct 1;38(10):1655-8. doi:10.5665/sleep.5068

Feyissa EA, Cornell E, Chandhok L, Wang D, **Ionita C**, Schwab J, Kostyun R, Wilton F, Rubin K. Impact of Co-management at the Primary-Subspecialty Care Interface on Follow-up and Referral Patterns for Patients With Concussion. Clin Pediatr (Phila). 2015 Sep;54(10):969-75. doi: 0.1177/0009922814566929

Mefford HC, Zemel M, Geraghty E, Cook J, Clayton PT, Paul K, Plecko B, Mills PB, Nordli DR Jr, **Gospe SM Jr**. Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu-Alu recombination. Neurology. 2015 Sep 1;85(9):756-62.

Arredondo J, Lara M, **Gospe SM Jr**, Mazia CG, Vaccarezza M, Garcia-Erro M, Bowe CM, Chang CH, Mezei MM, Maselli RA. Choline Acetyltransferase Mutations Causing Congenital Myasthenic Syndrome: Molecular Findings and Genotype-Phenotype Correlations. Hum Mutat. 2015 Sep;36(9):881-93. doi: 10.1002/humu.22823

Monrad P, Sannagowdara K, Bozarth X, Bhosrekar S, Hecox K, Nwosu M, Schwabe M, Meyer M, Szabo A, Prigge J, Lemke R, Horn B, Whelan HT. Haemodynamic response associated with both ictal and interictal epileptiform activity using simultaneous video electroencephalography/near infrared spectroscopy in a within-subject study. J Near Infrared Spectrosc. 2015;23(4):209-218. doi: 10.1255/jnirs.1170. Epub 2015 Sep 24. PMID: 26538840; PMCID: PMC4629858

Oegema R, Cushion TD, Phelps IG, Chung SK, Dempsey JC, Collins S, Mullins JG, Dudding T, Gill H, Green AJ, **Dobyns WB**, Ishak GE, Rees MI, Doherty D. Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Hum Mol Genet. 2015 Sep 15;24(18):5313-25. doi: 10.1093/hmg/ddv250. Epub 2015 Jun 30.

Blume HK. Headaches after Concussion in Pediatrics: a Review. Curr Pain Headache Rep. 2015 Sep;19(9):42. doi: 10.1007/s11916-015-0516-x. Review.

Ng YT, **Gospe SM Jr**, Sahin M. Pediatric Neurology 2014 Trainee Publication Award Winner: Dr. Mitchel T. Williams. Pediatr Neurol. 2015 Aug;53(2):103-4. doi:10.1016/j.pediatrneurol.2015.04.010.

Morgan LA, Buchhalter J. Psychogenic Paroxysmal Nonepileptic Events in Children: A Review. Pediatr Neurol. 2015 Jul;53(1):13-22.

Elbers J, **Wainwright MS**, **Amlie-Lefond C**. The Pediatric Stroke Code: Early Management of the Child with Stroke. J Pediatr. 2015 Jul;167(1):19-24.e1-4. doi:10.1016/j.jpeds.2015.03.051. Epub 2015 May 1. PubMed PMID: 25937428. 21: Amlie-Lefond C, Ellenbogen RG. Factors associated with the presentation of moyamoya in childhood. J Stroke Cerebrovasc Dis. 2015 Jun;24(6):1204-10. doi:10.1016/j.jstrokecerebrovasdis.2015.01.018. Epub 2015 Apr 10.

Roth CL, Eslamy H, Werny D, Elfers C, Shaffer ML, Pihoker C, Ojemann J, **Dobyns WB**. Semiquantitative analysis of hypothalamic damage on MRI predicts risk for hypothalamic obesity. Obesity (Silver Spring). 2015 Jun;23(6):1226-33. doi:10.1002/oby.21067. Epub 2015 Apr 17

Jansen LA, Mirzaa GM, Ishak GE, O'Roak BJ, Hiatt JB, Roden WH, Gunter SA, Christian SL, Collins S, Adams C, Rivière JB, St-Onge J, Ojemann JG, Shendure J, Hevner RF, **Dobyns WB**. PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. Brain. 2015 Jun;138(Pt 6):1613-28. doi: 10.1093/brain/awv045. Epub 2015 Feb 25.

Roosing S, Hofree M, Kim S, Scott E, Copeland B, Romani M, Silhavy JL, Rosti RO, Schroth J, Mazza T, Miccinilli E, Zaki MS, Swoboda KJ, Milisa-Drautz J, **Dobyns WB**, Mikati MA, Incecik F, Azam M, Borgatti R, Romaniello R, Boustany RM, Clericuzio CL, D'Arrigo S, Strømme P, Boltshauser E, Stanzial F, Mirabelli-Badenier M, Moroni I, Bertini E, Emma F,

Pediatric Neurology Faculty Publications 2014-2025

Steinlin M, Hildebrandt F, Johnson CA, Freilinger M, Vaux KK, Gabriel SB, Aza-Blanc P, Heynen-Genel S, Ideker T, Dynlacht BD, Lee JE, Valente EM, Kim J, Gleeson JG. Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. *Elife*. 2015 May 30;4:e06602. doi: 10.7554/eLife.06602.

Barkovich AJ, **Dobyns WB**, Guerrini R. Malformations of cortical development and epilepsy. *Cold Spring Harb Perspect Med*. 2015 May 1;5(5):a022392. doi:10.1101/cshperspect.a022392. Review.

Berg AT, **Dobyns WB**. Progress in autism and related disorders of brain development. *Lancet Neurol*. 2015 Nov;14(11):1069-70. doi:10.1016/S1474-4422(15)00048-4. Epub 2015 Apr 16

Saini A, Emke AR, Silva MC, **Perlman SJ**. Response to Eculizumab in Escherichia coli O157: H7-induced hemolytic uremic syndrome with severe neurological manifestations. *24817079 Clinical pediatrics*, 2015 April : 54(4)387-9

Hansen J, Snow C, Tuttle E, Ghoneim DH, Yang CS, Spencer A, Gunter SA, Smyser CD, Gurnett CA, Shinawi M, **Dobyns WB**, Wheless J, Halterman MW, Jansen LA, Paschal BM, Paciorkowski AR. De novo mutations in SIK1 cause a spectrum of developmental epilepsies. *Am J Hum Genet*. 2015 Apr 2;96(4):682-90. doi:10.1016/j.ajhg.2015.02.013. Erratum in: *Am J Hum Genet*. 2015 Jun 4;96(6):1009.

Luks VL, Kamitaki N, Vivero MP, Uller W, Rab R, Bovée JV, Rialon KL, Guevara CJ, Alomari AI, Greene AK, Fishman SJ, Kozakewich HP, Maclellan RA, Mulliken JB, Rahbar R, Spencer SA, Trenor CC 3rd, Upton J, Zurakowski D, Perkins JA, Kirsh A, Bennett JT, **Dobyns WB**, Kurek KC, Warman ML, McCarroll SA, Murillo R. Lymphatic and other vascular malformative/overgrowth disorders are caused by somatic mutations in PIK3CA. *J Pediatr*. 2015 Apr;166(4):1048-54.e1-5. doi:10.1016/j.jpeds.2014.12.069. Epub 2015 Feb 11

Lerche H, **Novotny EJ Jr**. Microscopic brain structure revisited in genetic epilepsy. *Neurology*. 2015 Mar 31;84(13):1290-1.

Szabo A, Hill MD, Scholz K, **Amlie-Lefond C**. Thrombolysis in pediatric stroke study. *Stroke*. 2015 Mar;46(3):880-5. doi: 10.1161/STROKEAHA.114.008210. Epub 2015 Jan 22.

Tully HM, Capote RT, Saltzman BS. Maternal and infant factors associated with infancy-onset hydrocephalus in Washington State. *Pediatr Neurol*. 2015 Mar;52(3):320-5. doi: 10.1016/j.pediatrneurol.2014.10.030.

Paciorkowski AR, McDaniel SS, Jansen LA, **Tully H**, Tuttle E, Ghoneim DH, Tupal S, Gunter SA, Vasta V, Zhang Q, Tran T, Liu YB, Ozelius LJ, Brashear A, Sweadner KJ, **Dobyns WB**, Hahn S. Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. *Epilepsia*. 2015 Mar;56(3):422-30. doi: 10.1111/epi.12914. Epub 2015 Feb 5..

Choe MC, **Blume HK**. Pediatric Posttraumatic Headache: A Review. *J Child Neurol*. 2016 Jan;31(1):76-85.

Simard-Tremblay E, Berry P, Owens A, Cook WB, **Sittner HR**, **Mazzanti M**, Huber J, **Warner M**, **Shurtleff H**, **Saneto RP**. High-fat diets and seizure control in myoclonic-astatic epilepsy: a single center's experience. *Seizure*. 2015 Feb;25:184-6. doi: 10.1016/j.seizure.2014.10.009

Mishra-Gorur K, Çağlayan AO, Schaffer AE, Chabu C, Henegariu O, Vonhoff F, Akgümüş GT, Nishimura S, Han W, Tu S, Baran B, Gümüş H, Dilber C, Zaki MS, HossniHAA, Rivière JB, Kayserili H, Spencer EG, Rosti RÖ, Schroth J, Per H, Çağlar C, Çağlar Ç, Dölen D, Baranoski JF, Kumandaş S, Minja FJ, Erson-Omay EZ, Mane SM, Lifton RP, Xu T, Keshishian H, **Dobyns WB**, Chi NC, Šestan N, Louvi A, Bilgüvar K, Yasuno K, Gleeson JG, Günel M. Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. *Neuron*. 2015 Jan 7;85(1):228. doi: 10.1016/j.neuron.2014.12.046.

Carapetian S (Randle), Hageman J, Lyons E, Leonard D, Janies K, Kelley K, Fuchs S. Emergency Department Evaluation and Management of Children With Simple Febrile Seizures. *Clin Pediatr (Phila)*. 2015;54:992-8.

Pediatric Neurology Faculty Publications 2014-2025

Carbonell A, Takeda A, Fahlgren N, **Johnson SC**, Cuperus JT, Carrington JC: New Generation of Artificial MicroRNA and Synthetic Trans-Acting Small Interfering RNA Vectors for Efficient Gene Silencing in Arabidopsis. *Plant Physiology*. 165(1):p15-29. 2014. PMID: 24647477.

Mishra-Gorur K, Çağlayan AO, Schaffer AE, Chabu C, Henegariu O, Vonhoff F, Akgümüş GT, Nishimura S, Han W, Tu S, Baran B, Gümüş H, Dilber C, Zaki MS, Hossni HA, Rivière JB, Kayserili H, Spencer EG, Rosti RÖ, Schroth J, Per H, Çağlar C, Çağlar Ç, Dölen D, Baranoski JF, Kumandaş S, Minja FJ, Erson-Omay EZ, Mane SM, Lifton RP, Xu T, Keshishian H, **Dobyns WB**, Chi NC, Šestan N, Louvi A, Bilgüvar K, Yasuno K, Gleeson JG, Günel M. Mutations in KATNB1 cause complex cerebral malformations by disrupting asymmetrically dividing neural progenitors. *Neuron*. 2014 Dec 17;84(6):1226-39. doi: 10.1016/j.neuron.2014.12.014. Erratum in: *Neuron*. 2015 Jan 7;85(1):228. *Neuron*. 2015 Jan 7;85(1):228

Johnson SC: Translational Medicine. A target for pharmacological intervention in an untreatable human disease. *Science*. 2014 Dec 5;346(6214). PMID: 25477449.

Mirzaa GM, Enyedi L, Parsons G, Collins S, Medne L, Adams C, Ward T, Davitt B, Bicknese A, Zackai E, Toriello H, **Dobyns WB**, Christian S. Congenital microcephaly and chorioretinopathy due to de novo heterozygous KIF11 mutations: five novel mutations and review of the literature. *Am J Med Genet A*. 2014 Nov;164A(11):2879-86. doi: 10.1002/ajmg.a.36707. Epub 2014 Aug 12. Review. PubMed

Wainwright MS, Grimason M, Goldstein J, Smith CM, **Amlie-Lefond C**, Revivo G, Noah ZL, Harris ZL, Epstein LG. Building a pediatric neurocritical care program: a multidisciplinary approach to clinical practice and education from the intensive care unit to the outpatient clinic. *Semin Pediatr Neurol*. 2014 Dec;21(4):248-54. doi: 10.1016/j.spen.2014.10.006. Epub 2014 Nov 1.

Ruhoy IS, Saneto RP. The genetics of Leigh syndrome and its implications for clinical practice and risk management. *Appl Clin Genet*. 2014 Nov 13;7:221-34. doi: 10.2147/TACG.S46176. eCollection 2014. Review..

Friedman SD, Ishak GE, Poliachik SL, Poliakov AV, Otto RK, Shaw DW, Willemsen MA, Bok LA, **Gospe SM Jr**. Callosal alterations in pyridoxine-dependent epilepsy. *Dev Med Child Neurol*. 2014 Nov;56(11):1106-10. doi: 10.1111/dmcn.12511. Epub

Johnson SC, Dong X, Vijg J, Suh Y: Genetic evidence for common pathways in human age-related diseases. *Aging Cell*. 2014 Oct;14(5):p809-17. PMID: 26077337.

Yamamoto S, Jaiswal M, Charng WL, Gambin T, Karaca E, Mirzaa G, Wiszniewski W, Sandoval H, Haelterman NA, Xiong B, Zhang K, Bayat V, David G, Li T, Chen K, Gala U, Harel T, Pehlivan D, Penney S, Vissers LELM, de Ligt J, Jhangiani SN, Xie Y, Tsang SH, Parman Y, Sivaci M, Battaloglu E, Muzny D, Wan YW, Liu Z, Lin-Moore AT, Clark RD, Curry CJ, Link N, Schulze KL, Boerwinkle E, **Dobyns WB**, Allikmets R, Gibbs RA, Chen R, Lupski JR, Wangler MF, Bellen HJ. A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*. 2014 Sep 25;159(1):200-214. doi: 10.1016/j.cell.2014.09.002

French CR, Seshadri S, Destefano AL, Fornage M, Arnold CR, Gage PJ, Skarie JM, **Dobyns WB**, Millen KJ, Liu T, Dietz W, Kume T, Hofker M, Emery DJ, Childs SJ, Waskiewicz AJ, Lehmann OJ. Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. *J Clin Invest*. 2014 Nov;124(11):4877-81. doi:10.1172/JCI75109. Epub 2014 Sep 24.

Kakar N, Ahmad J, Morris-Rosendahl DJ, Altmüller J, Friedrich K, Barbi G, Nürnberg P, Kubisch C, **Dobyns WB**, Borck G. STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. *Hum Genet*. 2015 Jan;134(1):45-51. doi: 10.1007/s00439-014-1487-4. Epub 2014 Sep 14.

Tully HM, Dobyns WB. Infantile hydrocephalus: a review of epidemiology, classification and causes. *Eur J Med Genet*. 2014 Aug;5(8):359-68. doi:10.1016/j.ejmg.2014.06.002.

Apkon SD, Grady R, Hart S, Lee A, McNalley T, Niswander L, Petersen J, Remley S, Rotenstein D, **Shurtleff H, Warner M**, Walker WO Jr. Advances in the care of children with spina bifida. *Adv Pediatr*. 2014 Aug;61(1):33-74. doi:10.1016/j.yapd.2014.03.007

Dobyns WB, Das S. LIS1-Associated Lissencephaly/Subcortical Band Heterotopia. 2009 Mar 3 [updated 2014 Aug 14]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews*®

Pediatric Neurology Faculty Publications 2014-2025

[Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from <http://www.ncbi.nlm.nih.gov/books/NBK5189/>.

Seltzer LE, Ma M, Ahmed S, Bertrand M, **Dobyns WB**, Wheless J, Paciorkowski AR Epilepsy and outcome in FOXP1-related disorders. *Epilepsia*. 2014 Aug;55(8):1292-300. doi: 10.1111/epi.12648. Epub WB. De novo mutations in the beta-tubulin gene TUBB2A cause simplified gyral patterning and infantile-onset epilepsy. *Am J Hum Genet*. 2014 Apr 3;94(4):634-41. doi: 10.1016/j.ajhg.2014.03.009.:

Mirzaa GM, Millen KJ, Barkovich AJ, **Dobyns WB**, Paciorkowski AR. The Developmental Brain Disorders Database (DBDB): a curated neurogenetics knowledge base with clinical and research applications. *Am J Med Genet A*. 2014 Jun;164A(6):1503-11. doi: 10.1002/ajmg.a.36517. Epub 2014 Apr 3. PubMed PMID:

Otten CE, Creutzfeldt CJ. Fulminant acute disseminated encephalomyelitis presenting in an adult. *JAMA Neurol*. 2014 71:648-9.

Nerloes A, Di Donato N, Masliah-Planchon J, Jongmans M, Abdul-Raman OA, Albrecht B, Allanson J, Brunner H, Bertola D, Chassaing N, David A, Devriendt K, Eftekhari P, Drouin-Garraud V, Faravelli F, Faivre L, Giuliano F, Guion Almeida L, Juncos J, Kempers M, Eker HK, Lacombe D, Lin A, Mancini G, Melis D, Lourenço, CM, Siu VM, Morin G, Nezarati M, Nowaczyk MJ, Ramer JC, Osimani S, Philip N, Pierpont ME, Procaccio V, Roseli ZS, Rossi M, Rusu C, Sznajer Y, Templin L, Uliana V, Klaus M, Van Bon B, Van Ravenswaaij C, Wainer B, Fry AE, Rump A, Hoischen A, Drunat S, Rivière JB, **Dobyns WB**, Pilz DT. Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. *Eur J Hum Genet*. 2015 Mar;23(3):292-301. doi: 10.1038/ejhg.2014.95. Epub 2014 Jul 23.

Rivkin MJ, deVeber G, Ichord RN, Kirton A, Chan AK, Hovinga CA, Gill JC, Bernard TJ, Rivkin MJ, Scholz K, deVeber G, Kirton A, Gill JC, Chan AK, Hovinga CA, Ichord RN, Grotta JC, Jordan LC, Benedict S, Friedman NR, Dowling MM, Elbers J, Torres M, Sultan S, Cummings DD, Grabowski EF, McMillan HJ, Beslow LA, **Amie-Lefond C**; Thrombolysis in Pediatric Stroke Study. Emergence of the primary pediatric stroke center: impact of the thrombolysis in pediatric stroke trial. *Stroke*. 2014 Jul;45(7):2018-23. doi: 10.1161/STROKEAHA.114.004919. Epub 2014 Jun 10

Beatty CW, Ko PR, Nixon J, **Gospe SM Jr**. Delayed-onset movement disorder and encephalopathy after oxycodone ingestion. *Semin Pediatr Neurol*. 2014 Jun;21(2):160-5. doi: 10.1016/j.spen.2014.06.009. Epub 2014 Jun 18. PubMed

Fernández-López D, **Natarajan N**, Ashwal S, Vexler ZS. Mechanisms of perinatal arterial ischemic stroke. *J Cereb Blood Flow Metab*. 2014 Jun;34(6):921-32. doi:10.1038/jcbfm.2014.41. Epub 2014 Mar 26. Review.

Tully HM, **Dobyns WB**. Infantile hydrocephalus: a review of epidemiology, classification and causes. *Eur J Med Genet*. 2014 Aug;57(8):359-68. doi:10.1016/j.ejmg.2014.06.002. Epub 2014 Jun 13. Review. PubMed PMID: 24932902;

Guerrini R, **Dobyns WB**. Malformations of cortical development: clinical features and genetic causes. *Lancet Neurol*. 2014 Jul;13(7):710-26. doi:10.1016/S1474-4422(14)70040-7. Epub 2014 Jun 2. Review.

Paciorkowski AR, Weisenberg J, Kelley JB, Spencer A, Tuttle E, Ghoneim D, Thio LL, Christian SL, **Dobyns WB**, Paschal BM. Autosomal recessive mutations in nuclear transport factor KPNA7 are associated with infantile spasms and cerebellar malformation. *Eur J Hum Genet*. 2014 May;22(5):587-93. doi:10.1038/ejhg.2013.196. Epub 2013 Sep 18.

Segal MM, Williams MS, Gropman AL, Torres AR, Forsyth R, Connolly AM, El-Hattab AW, **Perlman SJ**, Samanta D, Parikh S, Pavlakakis SG, Feldman LK, Betensky RA, **Gospe SM Jr** Evidence-based decision support for neurological diagnosis reduces errors and unnecessary workup. 23576414 *Journal of child neurology*, 2014 April : 29(4):487-92

Pearl PL, **Gospe SM Jr**. Pyridoxine or pyridoxal-5'-phosphate for neonatal epilepsy: the distinction just got murkier. *Neurology*. 2014 Apr 22;82(16):1392-4. doi: 10.1212/WNL.0000000000000351

Wallace SE, Conta JH, Winder TL, Willer T, Eskuri JM, Haas R, Patterson K, Campbell KP, Moore SA, **Gospe SM Jr**. A novel missense mutation in POMT1 modulates the severe congenital muscular dystrophy phenotype associated with POMT1 nonsense mutations. *Neuromuscul Disord*. 2014 Apr;24(4):312-20. doi:10.1016/j.nmd.2014.01.001. Epub 2014 Jan 11.

Cacciagli P, Desvignes JP, Girard N, Delepine M, Zelenika D, Lathrop M, Lévy N, Ledbetter DH, **Dobyns WB**, Villard L. AP1S2 is mutated in X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). *Eur J Hum Genet*. 2014 Mar;22(3):363-8. doi:10.1038/ejhg.2013.135. Epub 2013 Jun 12.

Pediatric Neurology Faculty Publications 2014-2025

Ruhoy IS, Merritt JL 2nd, **Amlie-Lefond C**. Cystathionine beta-synthase deficiency heralded by cerebral sinus venous thrombosis and stroke. *Pediatr Neurol*. 2014 Jan;50(1):108-11. doi: d10.1016/j.pediatrneurol.2013.08.021. Epub 2013 Oct 15.

Amlie-Lefond C, Gill JC. Approach to acute ischemic stroke in childhood. *Curr Treat Options Cardiovasc Med*. 2014 Jan;16(1):276. doi: 10.1007/s11936-013-0276-z.

van Karnebeek CD, Stockler-Ipsiroglu S, Jaggumantri S, Assmann B, Baxter P, Buhas D, Bok LA, Cheng B, Coughlin CR 2nd, Das AM, Giezen A, Al-Hertani W, Ho G, Meyer U, Mills P, Plecko B, Struys E, Ueda K, Albersen M, Verhoeven N, **Gospe SM Jr**, Gallagher RC, Van Hove JK, Hartmann H. Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations. *JIMD Rep*. 2014;15:1-11. doi: 10.1007/8904_2014_296.

Johnston JJ, Sapp JC, Curry C, Horton M, Leon E, Cusmano-Ozog K, **Dobyns WB**, Hudgins L, Zackai E, Biesecker LG. Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism. *Am J Med Genet A*. 2014 Jan;164A(1):120-8. doi: 10.1002/ajmg.a.36212.

Beatty CW, Creutzfeldt CJ, Davis AP, Hoffer Z, Khot SP. The diagnostic conundrum and treatment dilemma of a patient with a rapidly progressive encephalopathy. *Neurohospitalist*. 2014 Jan;4(1):34-41. doi:10.1177/1941874413496792.

Carapetian S (Randle), Hesselink J, Nass R, Trauner D, Stiles J. Face and location processing in children with early unilateral brain injury. *Brain Cogn*. 2014;88:6-13.

Wei Z, Chigurupati S, Bagsiyao P, **Henriquez A**, Chan SL. The brain uncoupling protein UCP4 attenuates mitochondrial toxin-induced cell death: role of extracellular signal-regulated kinases in bioenergetics adaptation and cell survival. *Neurotox Res*. 2009 Jul;16(1):14-29. doi: 10.1007/s12640-009-9039-8. Epub 2009 Mar 25.