UAB HEERSINK SCHOOL OF MEDICINE
The University of Alabama at Birmingham

Pediatric Neurology

2021 Publications

Title

AJOB Empir Bioeth. 2021 Apr 12;1-17. *The Therapeutic Odyssey: Positioning Genomic Sequencing in the Search for a Child's Best Possible Life*. Janet Elizabeth Childerhose, Carla Rich, Kelly M East, Whitley V Kelley, Shirley Simmons, Candice R Finnila, Kevin Bowling, Michelle Amaral, Susan M Hiatt, Michelle Thompson, David E Gray, James M J Lawlor, Richard M Myers, Gregory S Barsh, Edward J Lose, **Martina E Bebin**, Greg M Cooper, Kyle Bertram Brothers .

Am J Hum Genet. 2021 Apr 21;S0002-9297(21)00138-5. *Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism.* Maya Chopra, Meriel McEntagart, Jill Clayton-Smith, Konrad Platzer, Anju Shukla, Katta M Girisha , Anupriya Kaur, Parneet Kaur, Rolph Pfundt, Hermine Veenstra-Knol, Grazia M S Mancini, Gerarda Cappuccio, Nicola Brunetti-Pierri, Fanny Kortüm, Maja Hempel, Jonas Denecke, Anna Lehman, CAUSES Study; Tjitske Kleefstra, Kyra E Stuurman, Martina Wilke, Michelle L Thompson, **E Martina Bebin**, Emilia K Bijlsma, Mariette J V Hoffer, Cacha Peeters-Scholte, Anne Slavotinek, William A Weiss, Tiffany Yip , Ugur Hodoglugil, Amy Whittle, Janette diMonda, Juanita Neira, Sandra Yang, Amelia Kirby, Hailey Pinz, Rosan Lechner, Frank Sleutels, Ingo Helbig, Sarah McKeown, Katherine Helbig, Rebecca Willaert, Jane Juusola, Jennifer Semotok, Medard Hadonou, John Short, Genomics England Research Consortium; Naomi Yachelevich, Sajel Lala, Alberto Fernández-Jaen, Janvier Porta Pelayo, Chiara Klöckner, Susanne B Kamphausen, Rami Abou Jamra, Maria Arelin, A Micheil Innes, Anni Niskakoski, Sam Amin, Maggie Williams, Julie Evans, Sarah Smithson, Damian Smedley, Anna de Burca, Usha Kini, Martin B Delatycki, Lyndon Gallacher, Alison Yeung, Lynn Pais, Michael Field, Ellenore Martin, Perrine Charles, Thomas Courtin, Boris Keren, Maria Iascone, Anna Cereda, Gemma Poke, Véronique Abadie, Christel Chalouhi, Padmini Parthasarathy, Benjamin J Halliday, Stephen P Robertson, Stanislas Lyonnet, Jeanne Amiel, Christopher T Gordon.

Am J Physiol Cell Physiol. 2021 Aug 1;321(2):C230-C246. *Mechanics of dystrophin deficient skeletal muscles in very young mice and effects of age.* **Michael A Lopez**, Sherina Bontiff, Mary Adeyeye, Aziz I Shaibani, **Matthew S Alexander**, Shari Wynd, Aladin M Boriek.

Ann Clin Transl Neurol. 2021 Aug 19;10.1002/acn3.51441 *Gut microbiome is associated with multiple sclerosis activity in children.* Mary K Horton, Kathryn McCauley, Douglas Fadrosh, Kei Fujimura, Jennifer Graves, **Jayne Ness**, Yolanda Wheeler , Mark P Gorman, Leslie A Benson, Bianca Weinstock-Guttman, Amy Waldman, Moses Rodriguez, Jan-Mendelt Tillema, Lauren Krupp, Anita Belman, Soe Mar, Mary Rensel, Tanuja Chitnis, Theron Charles Casper, John Rose, Janace Hart, Xiaorong Shao, Helen Tremlett, Susan V Lynch, Lisa F Barcellos, Emmanuelle Waubant, U.S. Network of Pediatric MS Centers.

Ann Neurol. 2021 Jan 22. *Multisite Study of Evoked Potentials in Rett Syndrome*. Joni N Saby, Timothy A Benke, Sarika U Peters, Shannon M Standridge, Junko Matsuzaki, Clare Cutri-French, Lindsay C Swanson, David N Lieberman, Alexandra P Key, **Alan K Percy**, Jeffrey L Neul, Charles A Nelson, Timothy P L Roberts, Eric D Marsh.

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Ann Neurol. 2021 Jan 7. *Tuber locations associated with infantile spasms map to a common brain network*. Alexander L Cohen, Brechtje Pf Mulder, Anna K Prohl, Louis Soussand, Peter Davis, Mallory R Kroeck, Peter McManus, Ali Gholipour, Benoit Scherrer, **E Martina Bebin**, Joyce Y Wu, Hope Northrup, Darcy A Krueger, Mustafa Sahin, Simon K Warfield, Michael Fox, Jurriaan M Peters, TACERN study group.

Clin Pharmacol Ther. 2021 Aug 31; 10.1002/cpt.2408. *Pharmacogenetic predictors of cannabidiol (CBD) response and tolerability in treatment-resistant epilepsy.* Brittney H Davis, T Mark Beasley, Michelle Amaral, Jerzy P Szaflarski, Tyler Gaston, Leslie Perry Grayson, David G Standaert, **E Martina Bebin**, Nita A Limdi, UAB CBD Study Group (includes all the investigators involved in the UAB EAP CBD program).

Epilepsy Behav. 2021 Jul 14;122:108190. *Longitudinal impact of cannabidiol on EEG measures in subjects with treatment-resistant epilepsy*. Leslie Grayson, Steve Ampah, Kathleen Hernando, **Pongkiat Kankirawatana**, Tyler Gaston, Gary Cutter, Jerzy P Szaflarski, **Elizabeth Martina Bebin,** UAB CBD Program.

Epilepsy Behav. 2021 Mar 2;117:107862. *Long-term safety and efficacy of highly purified cannabidiol for treatment refractory epilepsy*. Tyler E Gaston, Steve B Ampah, **E Martina Bebin**, Leslie P Grayson, Gary R Cutter, Kathleen Hernando, Jerzy P Szaflarski, UAB CBD Program.

Epilepsy Curr. 2021 Mar 10;1535759721999670. *Treatment of Super-Refractory Status Epilepticus: A Review*. Juan G Ochoa, Michelle Dougherty, Alex Papanastassiou, Barry Gidal, **Ismail Mohamed**, David G Vossler.

Eur J Hum Genet. 2020 Oct;28(10):1422-1431. *A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome.* Theodore G Drivas, Dong Li, Divya Nair, Joseph T Alaimo, Mariëlle Alders, Janine Altmüller, Tahsin Stefan Barakat, **E Martina Bebin**, Nicole L Bertsch, Patrick R Blackburn, Alyssa Blesson, Arjan M Bouman, Knut Brockmann, Perrine Brunelle, Margit Burmeister, Gregory M Cooper, Jonas Denecke, Anne Dieux-Coëslier, Holly Dubbs, Alejandro Ferrer, Danna Gal, Lauren E Bartik, Lauren B Gunderson, Linda Hasadsri, Mahim Jain, Catherine Karimov, Beth Keena, Eric W Klee, Katja Kloth, Baiba Lace, Marina Macchiaiolo, Julien L Marcadier, Jeff M Milunsky, Melanie P Napier, Xilma R Ortiz-Gonzalez, Pavel N Pichurin, Jason Pinner, Zoe Powis, Chitra Prasad, Francesca Clementina Radio, Kristen J Rasmussen, Deborah L Renaud, Eric T Rush, Carol Saunders, Duygu Selcen, Ann R Seman, Deepali N Shinde, Erica D Smith, Thomas Smol, Lot Snijders Blok, Joan M Stoler, Sha Tang, Marco Tartaglia, Michelle L Thompson, Jiddeke M van de Kamp, Jingmin Wang, Dagmar Weise, Karin Weiss, Rixa Woitschach, Bernd Wollnik, Huifang Yan, Elaine H Zackai, Giuseppe Zampino, Philippe Campeau, Elizabeth Bhoj.

J Child Neurol. 2021 Apr 5;8830738211002679. *Pediatric N-Methyl-d-Aspartate (NMDA) Receptor Encephalitis, With and Without Herpes Encephalitis*. **Lydia Marcus, Jayne M Ness.**

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J Child Neurol. 2021 Dec 7;8830738211027972. *Brain Magnetic Resonance Imaging (MRI) White Matter Hyperintensities in Cyclic Vomiting Syndrome With or Without Migraine*. **Salman Rashid,** Samantha Weaver, Khaled Al-Robaidi, **Leon Dure,** Sumit Singh.

J Neurodev Disord. 2021 Sep 16;13(1):40. *Current neurologic treatment and emerging therapies in CDKL5 deficiency disorder.* Heather E Olson, Carolyn I Daniels, Isabel Haviland, Lindsay C Swanson, Caitlin A Greene, Anne Marie M Denny, Scott T Demarest , Elia Pestana-Knight, Xiaoming Zhang, Ahsan N Moosa, Andrea Fidell, Judith L Weisenberg, Bernhard Suter, Cary Fu, Jeffrey L Neul, **Alan K Percy**, Eric D Marsh, Timothy A Benke , Annapurna Poduri.

JAMA Pediatr. 2021 Feb 1. *Association of Headache With School Functioning Among Children and Adolescents in the United States*. **Scott B Turner**, Christina L Szperka, Andrew D Hershey, Emily F Law, Tonya M Palermo, Cornelius B Groenewald.

Life (Basel). 2021 Jul 4;11(7):648. *The Interplay of Mitophagy and Inflammation in Duchenne Muscular Dystrophy*. Andrea L Reid, **Matthew S Alexander**.

Muscle Nerve. 2021 Mar 2. *hnRNP L is essential for myogenic differentiation and modulates myotonic dystrophy pathologies*. **Matthew S Alexander,** Rylie M Hightower, Andrea L Reid, Alexis H Bennett, Lakshmanan Iyer, Donna K Slonim , Madhurima Saha, Genri Kawahara, Louis M Kunkel, Alan S Kopin, Vandana A Gupta, Peter B Kang, Isabelle Draper.

Neurol Neuroimmunol Neuroinflamm. 2021 Aug 5;8(5):e1049. *Familial History of Autoimmune Disorders Among Patients With Pediatric Multiple Sclerosis*. Benjamin M Greenberg, Theron Charles Casper, Soe S Mar, **Jayne M Ness**, Patricia Plumb, Shannon Liang, Manu Goyal , Bianca Weinstock-Guttman, Moses Rodriguez, Gregory S Aaen, Anita Belman, Lisa F Barcellos, John W Rose, Mark P Gorman, Leslie A Benson, Meghan Candee, Tanuja Chitnis, Yolanda C Harris, Ilana L Kahn, Shelly Roalstad, Janace Hart, Timothy E Lotze, Mary Rensel, Jennifer P Rubin, Teri L Schreiner, Jan-Mendelt Tillema, Amy Tara Waldman, Lauren Krupp, Jennifer Graves, Kaylea Drake, Emmanuelle Waubant.

Neurology. 2021 Feb 10;10.1212/WNL.0000000000011653. *Association of de novo RNF213 Variants with Childhood Onset Moyamoya Disease and Diffuse Occlusive Vasculopathy.* Amélie Pinard, Maximillian D J Fiander, Alana C Cecchi, Andrea L Rideout, Mohamed Azouz, Stuart M Fraser, P Daniel McNeely, Simon Walling, **Sarah C Novara**, Anna C E Hurst, Dongchuan Guo, Sandhya Parkash, Michael J Bamshad, Deborah A Nickerson, Anthony M Vandersteen, Dianna M Milewicz.

Neurology. 2021 Jun 9;10.1212/WNL.0000000000012261. *Factors Associated With, and Mitigation Strategies for, Healthcare Disparities Faced by Patients With Headache Disorders*. Jessica Kiarashi, Juliana VanderPluym, Christina L Szperka, **Scott Turner**, Mia T Minen, Susan Broner, Alexandra C Ross, Amanda E Wagstaff, Marissa Anto, Maya Marzouk, Teshamae S Monteith, Noah Rosen, Salvador L Manrriquez, Elizabeth Seng, Alan Finkel, Larry Charleston 4 th.

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Pediatr Neurol. 2021 Jul 24;123:50-66. *Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations.* Hope Northrup, Mary E Aronow, **E Martina Bebin**, John Bissler, Thomas N Darling, Petrus J de Vries, Michael D Frost, Zoë Fuchs, Elizabeth S Gosnell, Nishant Gupta, Anna C Jansen, Sergiusz Jóźwiak, J Chris Kingswood, Timothy K Knilans, Francis X McCormack, Ashley Pounders, Steven L Roberds, David F Rodriguez-Buritica, Jonathan Roth, Julian R Sampson, Steven Sparagana, Elizabeth Anne Thiele, Howard L Weiner, James W Wheless, Alexander J Towbin, Darcy A Krueger, International Tuberous Sclerosis Complex Consensus Group.

Prog Neuropsychopharmacol Biol Psychiatry. 2021 Feb 4;109:110259. *Microbial community changes in a female rat model of Rett syndrome*. A Gallucci, K C Patterson, A R Weit, W J Van Der Pol, L G Dubois, **A K Percy**, C D Morrow , S L Campbell, M L Olsen.

Seizure. 2021 Aug 2;91:503-506. *What is the optimal duration for vigabatrin monotherapy in patients with infantile spasms: 6 months or longer?* Béatrice Desnous, Marien Lenoir, Jonathan Y Bitton, Mélina Arbour, Nathalie Villeneuve, Sharon Whiting, **Ismail Mohammed** , Elaine C Wirrell, Luis Bello-Espinosa, Gabriel M Ronen, Anne Lortie, Ala Birca.

Seizure. 2021, 89, 73-80 3. *Cognitive outcome in children with infantile spasms using a standardized treatment protocol. A five-year longitudinal study.* Bitton JY, Desnous B, Sauerwein H, Connolly M, Weiss K, Donner E, Whiting S, **Mohamed IS**, Wirrell E, Ronen G, Lortie A.