

# Workshops Exploring Outcome Measures for the Most Severely Affected Populations

November 4 and 11, 2021

# Dr. Terry Jo Bichell, PhD, MPH

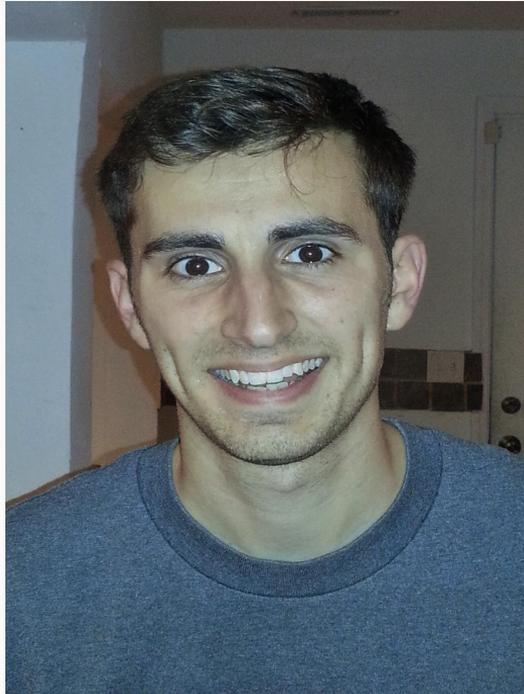
## Executive Director of COMBINEDBrain



Terry Jo Bichell worked as a documentary filmmaker in the early days of videotape, then became a public health nurse-midwife after filming a difficult birth in West Africa. When her youngest child, Lou, was diagnosed with Angelman syndrome, she switched from midwifery to clinical research on Angelman syndrome. Eventually, she went back to school to earn a PhD in neuroscience from Vanderbilt University in an effort to find treatments for her son. Along the way, she studied gene-environment interactions in Huntington disease as well as circadian aspects of Angelman syndrome and was a columnist for HDBuzz. After graduating, she was the Founding Director of the Angelman Biomarkers and Outcome Measures Alliance until 2018. Dr. Bichell founded a new non-profit in 2019, COMBINEDBrain (Consortium for Outcome Measures and Biomarkers for Neurodevelopmental Disorders), to assist other rare and ultra-rare neurogenetic disorders with clinical trial preparations.

# Dr. Kyle J. Horning, PhD

## COMBINEDBrain Post-Doctoral Fellow



Kyle J. Horning received his bachelor's degree in neuroscience in 2010 from Drew University. He continued his training in neuroscience as a research technician at the Maryland Psychiatric Research Center until 2013, when he began graduate school at Vanderbilt University. At Vanderbilt, he received additional training in toxicology and bioinformatics while studying gene and environmental interactions in Huntington's disease for his dissertation. He received his PhD in neuroscience from Vanderbilt in 2020. In January 2021, Dr. Horning joined COMBINEDBrain as a postdoctoral science officer.

# JayEtta Hecker

## Executive Director and Co-founder of the International SCN8A Alliance, Wishes for Elliott, and DEE-P Connections



JayEtta is an economist who specialized in independent public policy analyses over a 44 year career with the federal government. JayEtta served in multiple federal agencies, the Carter White House, and as a senior executive within the Congressional research arm, the Government Accountability Office. She was a frequent witness before Senate and House committees providing evidence from evaluations she directed of major public policy issues. She served on multiple committees of the Transportation Research Board within the National Academy of Sciences.

Elliott was her first grandchild and she was an active caregiver and partner in his diagnostic odyssey. She partnered with Gabi, her daughter and Elliott's mother, to form Wishes for Elliott to advance SCN8A research and create a lasting legacy for Elliott, who continues to serve as an inspiration for all who know him. She was a co-founder and serves as Executive Director of the International SCN8A Alliance and DEE-P Connections, helping build innovative collaborations to accelerate research and to improve the outcomes of all those struggling with DEEs.

# Gabi Conecker

## Co-founder and President of the International SCN8A Alliance and DEE-P Connections



Gabi is mom to Elliott, now 9 years old, who struggles with one of the most severe SCN8A mutations. At diagnosis, the family received the one published article on SCN8A in children offering little to no guidance on care, treatment, or prognosis. Gabi founded Wishes for Elliott to fight for better treatment and research for SCN8A families. Even though she was still working full time building maternal/child health programs in sub-Saharan Africa and caring for a severely medically fragile child, she was inspired to bring the passion and urgency of SCN8A families to the nascent but growing community of SCN8A researchers and clinicians – and do everything possible to improve understanding for better treatment and outcomes for all SCN8A children.

She co-founded DEE-P Connections, in 2019, out of desperation to see better tools, resources and research for the most severely affected families, like her own.

# Dr. Anne Berg, PhD



The focus of my research for over 30 years has been the "natural" history of pediatric seizures and epilepsy with a specific emphasis on seizure outcomes, developmental and cognitive consequences the impact on quality of life, impact on families, and the implications of all of these considerations for care and care-models. In 2007, I played a key role in focusing NINDS research priorities on the cognitive, developmental, and behavioral co-morbidities of epilepsy and organized sessions at the American Epilepsy Society meeting to investigate these areas and identify research opportunities for improving patients' outcomes. In 2013, I Co-Chaired the NINDS-sponsored Curing Epilepsies conference and also organized and led the workshop, "Priorities in Pediatric Epilepsy Research: Improving Children's Futures Today. I was one of the initial founders of the Pediatric Epilepsy Research Consortium (PERC) which has grown to over 40 centers in the US. My current work centers on developing resources and means to facilitate rapid, collection of high-value information about the abilities and impairments in a large range of domains affected by developmental brain disorders and early life epilepsies. The parent-caregiver voice in identifying research priorities and directing investigations is a central component to these efforts. A driving force behind my research endeavors is to develop and use evidence to transform attitudes and approaches toward early life epilepsy from one of futility to one in which precision diagnosis can lead to precision therapies and ultimately to improved outcome for the affected individual, the family, and society as a whole.

# Chere Chapman

## CEO Ardea Outcomes



With an Honours BSc in biology, Chère Chapman set out for a year in rural Japan as part of a global teaching and cross-cultural exchange program of the Japanese government. Following two years back in Canada for her Master's of Health Science in Community Health and Epidemiology from University of Toronto, Chère returned in Asia as an HIV/AIDS epidemiologist at the Communicable Disease Centre (CDC) in Singapore. Over the next decade, Chère did her MBA at London Business School before returning to Asia once again to establish a consultancy in Singapore and Vietnam developing multi-stakeholder partnerships with international NGOs and Fortune 100 companies. Chère has negotiated complex projects from Papua New Guinea to India to Bangladesh for organizations such as CARE International, Chevron, Visa, and The Clinton Foundation.

Chère's East Coast Canadian roots eventually pulled her back to Nova Scotia where she is now CEO of Ardea Outcomes, a specialist contract research organization focused on patient-centered outcomes. Chère is a past board member for the University of King's College and Touch Sala Bai, a charity fighting human trafficking in Cambodia. Chère is currently an associate at Creative Destruction Lab Atlantic, a founder of Sandpiper Ventures, and the chair of ONSIDE, a multi-stakeholder organization driving inclusive innovation driven entrepreneurship in Eastern Canada.

Chère lives in Halifax with her husband Gord, their four crazy kids, and adorable pandemic puppy.

# Dr. Jenny Downs, BApp1Sci (physio) MSc PhD



Dr Jenny Downs is Head of the Disability Research Program and Co-Head of the Child Disability Team at the Telethon Kids Institute in Perth. Her current programs include research on rare disorders including Rett syndrome, the CDKL5 Deficiency Disorder, MECP2 Duplication syndrome and Prader-Willi syndrome, as well as Down syndrome, autism and cerebral palsy which occur more frequently.

Dr Downs led the development of gross motor and hand function measures for Rett syndrome, and a quality of life measure for children with intellectual disability, the Quality of Life Inventory – Disability. (QI-Disability). She is lead PI for the Australian site of an NIH funded US-Australian study developing and validating outcome measures for the CDKL5 Deficiency Disorder.

Her vision is that all children with developmental vulnerability or disability will live with reduced impairments and optimal quality of life, and that research findings will be distributed equitably across the community.

# Natasha Ludwig, PhD - Neuropsychologist



Dr. Natasha Ludwig received her bachelor's degree in Neuroscience from Union College and her master's and doctoral degree in Clinical Psychology with emphasis on neuropsychology and cognitive neuroscience from Georgia State University. Dr. Ludwig completed an APA-accredited internship in neuropsychology rehabilitation/pediatric psychology and a postdoctoral fellowship in pediatric neuropsychology at Kennedy Krieger Institute/Johns Hopkins School of Medicine.

Dr. Ludwig joined the Neuropsychology Department at Kennedy Krieger Institute in 2019 and is an Assistant Professor of Psychiatry and Behavioral Sciences at the Johns Hopkins University School of Medicine. Clinically, Dr. Ludwig provides neuropsychological evaluations for children with a wide variety of medical and neurodevelopmental conditions from birth through adulthood, primarily within the Congenital/Genetic Conditions Clinic and the Epilepsy and Brain Injury Rehabilitation Clinic.