fast Dream Big.

2022 FAST Translational Research Symposium and ABOM Scientific Meeting
Speaker Bios

Allyson Berent, DVM, DACVIM

FAST

Dr. Berent is a veterinary internal medicine specialist/interventionalist who graduated from Cornell University and completed her residency at the University of Pennsylvania, where she served as an Adjunct Assistant Professor before joining the Animal Medical Center in NYC. She is the Director of Interventional Endoscopy, focusing on clinical trials researching medical devices particularly for ureteral and biliary obstructions. In 2014 Dr. Berent's daughter, Quincy, was diagnosed with Angelman syndrome. In 2015 she joined the Board of Directors for the Foundation for Angelman Syndrome Therapeutics (FAST), becoming the Chief Science Officer. Dr. Berent serves as the co-director of the Angelman Syndrome Biomarker and Outcome Measure Consortium, to co-director for the International Angelman Syndrome Research Council (INSYNC-AS), and is an advisor to numerous pharmaceutical companies working on therapeutic candidates for rare neurodevelopmental disorders. Dr. Berent co-founded GeneTx Biotherapeutics, a company focused on advancing an antisense oligonucleotide therapy for AS, where she was the Chief Operating Officer. GeneTx was acquired in 2022 by Ultragenyx Pharmaceuticals, after launching the Phase1/2 clinical trial, and she now serves as a consultant for Ultragenyx.

Jennifer Panagoulias, RAC

FAST, Angelman Syndrome Biomarker and Outcome Measure Consortium

Jennifer has worked in drug development for over 20 years, primarily focused on advancing global development programs for the treatment of rare neurological diseases. She spent 16 years in Regulatory Affairs at Genzyme where she held various roles including Global Therapeutic Head, Regulatory Affairs Neurology. At Genzyme, she supported global registration efforts for Myozyme® (alglucosidase alfa), an enzyme replacement therapy for children and adults with Pompe disease; a rare, genetic, neuromuscular disorder. Jennifer has experience in working with global regulatory agencies including the US FDA, the European Medicines Agency, and the Pharmaceutical and Medical Device Agency in Japan to support diverse drug and biological therapy development programs in both the development and post-marketing stages. Most recently, she has worked on the development of oligonucleotide drugs as Senior Vice President of Regulatory, Compliance, and Policy at Wave Life Sciences. Jennifer is Co-Director of the Angelman Syndrome Biomarker and Outcome Measure Consortium (ABOM).

Dora Markati, MD, MPhil

University of Oxford

Dora completed her studies in Medicine in 2016. In 2020 she was awarded her MPhil degree in Biological Science from the University of Cambridge (UK), during which she worked on the molecular mechanisms of nervous system development. She has received clinical training in General Paediatrics at Chelsea and Westminster Hospital (London, UK), Addenbrooke's Hospital (Cambridge, UK), and in Neonatology at St Mary's Hospital (London, UK). She is currently a DPhil student in the Department of Paediatrics. Her studies are funded by the Department of Paediatrics and the Alexander S. Onassis Foundation.

She is working in the team of Professor Servais at the University of Oxford. She has co-designed the Natural History Study for Angelman syndrome sponsored by the University of Oxford and funded by the Foundation for Angelman Syndrome Therapeutics UK. Overall, her research interest lies in drug development for rare neurogenetic conditions. Aspects of her research include: (1) the translation of preclinical discoveries into clinical applications and novel therapies, (2) the design and conduct of natural history studies and clinical trials, and (3) the discovery and validation of biomarkers using different approaches and techniques of molecular biology.

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Bridgette Kelleher, PhD

Purdue University

Dr. Bridgette Kelleher is an associate professor of psychological sciences and serves as Director of the Purdue Autism Research Center. She obtained her Ph.D. from the University of South Carolina (2015) after completing her APA-certified clinical residency at the Medical University of South Carolina/Charleston Consortium. Kelleher's internationally-recognized research program focuses on optimizing clinical outcomes in "high risk" populations, with particular focus on using technology-driven solutions to assess and treat families affected by rare disorders. Her current research addresses three primary questions:

1. How can remote technology be used to improve clinical monitoring of children at "high risk" for atypical development?
2. How can wearable technologies be optimized for clinical purposes and populations?
3. Can remotely collected "real world" data improve mental health treatment, including for caregivers affected by the COVID-19 pandemic?

Kelleher is addressing these questions through a variety of active research grants funded by the National Institutes of Health and patient-focused foundations.

Jessica Duis, MD, MSc

Children's Hospital Colorado

Dr. Jessica Duis is an Associate Professor of Pediatrics and Genetics at Children's Hospital Colorado, University of Colorado. She did her medical training at Johns Hopkins School of Medicine in Baltimore, MD. She completed a post-doctoral fellowship in the Johns Hopkins Department of Psychiatry and Behavioral Sciences. She is a board-certified pediatrician and medical geneticist who practices in the area of genetics and complex/special care pediatrics and primarily performs diagnostic work up and management for individuals with rare disorders focused on neurogenetic conditions and rare genetic causes of obesity and metabolic conditions. She has focused her career on chromosome 15 disorders including Angelman Syndrome, Duplication 15q, and Prader-Willi syndrome. She has founded and built Centers of Excellence for Angelman, Prader-Willi, duplication 15q, and Pitt Hopkins Syndromes. Dr. Duis' career has spanned translational, clinical and bench research. She is passionate about establishing standards of care, developing objective and sensitive outcome measures and improving clinical trial design for individuals with neurodevelopmental disorders to improve quality of life and equity of care.

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Laurent Servais, MD, PhD

University of Oxford

I am Professor of Pediatric Neuromuscular Diseases at the Muscular Dystrophy United Kingdom (MDUK) Oxford Neuromuscular Centre and Invited Professor of Child Neurology at Liège University. I lead the Specialised Translational Research Oxford Neuromuscular Group (STRONG).

After graduating from Louvain Medical School, Brussels, Belgium in 1999, I completed a PhD in Neuroscience (cerebellar electrophysiology in alert living mice) from the Free University of Brussels, Belgium, followed by residencies in child neurology at the Free University of Brussels and Robert Debré Hospital, Paris. In 2008, I took a position in neuromuscular disease and clinical research at the Institute of Myology in Paris, where my interest and expertise in neuromuscular diseases flourished. I was subsequently appointed Head of Clinical Trials and Database Services. Most recently, I served as Head of the Institute of Myology's I-Motion (Institute Of Muscle-Oriented Translational Innovation), and Head of the Neuromuscular Centre in Liège, Belgium. I have joined MDUK Oxford Neuromuscular Center and the University of Oxford in September 2019.

I have been involved as principal investigator in numerous clinical trials to test treatments for Duchenne muscular dystrophy, spinal muscular atrophy (SMA), X-linked Myotubular Myopathy, Angelman syndrome and other neuromuscular conditions.

I am the leader of the newborn screening program for SMA in southern Belgium where we have also conducted a medico-economic analysis of newborn screening. This program has now grown into "Babydetect", an innovative universal newborn screening program that aims to identifiy at birth any early onset treatable severe condition. My main research expertise covers the development of innovative outcome measures, including connected devices for real-life patients' evaluation. I have led the qualification of the first digital outcome by a regulatory agency, the SV95C in Duchenne Muscular Dystrophy- and I hope to reach a similar achievement in Angelman

Sarah Nelson Potter, PhD, CCC-SLP

RTI International

Sarah Nelson Potter is a research public health analyst at RTI International. She received her Ph.D. in the Human Development Graduate Group at the University of California, Davis in the fall of 2021. During her training at UC Davis, she worked with Dr. Leonard Abbeduto in the Laboratory on Language Development in Neurodevelopmental Disabilities at the UC Davis MIND Institute. Prior to joining Dr. Abbeduto's lab, Sarah received a master's degree in speech-language pathology from the University of Wisconsin-Madison. At RTI, Sarah supports projects focused on rare neurogenetic conditions, with the majority of her work focused on Angelman syndrome. Her projects are primarily related to the development and validation of outcome measures, and the dissemination of findings from the Angelman Natural History Study.

Angela You Gwaltney, PhD

RTI International

Angela Gwaltney, PhD, a research public health analyst in the GenOmics, Bioinformatics, and Translational Research Center at RTI International. Her area of expertise is in the use of statistical techniques grounded in social sciences with particular focus on improving health and well-being outcomes of individuals and families with rare hereditary conditions.

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Anjali Sadhwani, PhD

Boston Children's Hospital

Dr. Anjali Sadhwani is a pediatric psychologist working at Boston Children's Hospital (BCH) and Assistant Professor of Psychiatry at Harvard Medical School. She is passionate about working with children with complex medical and developmental challenges. She has been conducting clinical research in Angelman syndrome (AS) since November 2010. Currently she is the lead psychologist of the multi-center Angelman syndrome (AS) Natural History and has extensive experience administering neurodevelopmental and behavioral measures to this population.

Christina K. Zigler, PhD, MSEd

Duke University School of Medicine

Dr. Zigler is an Assistant Professor in the Department of Population Health Sciences at Duke University School of Medicine. A psychometrician and statistician by training, she uses rigorous, patient-centered methods to develop and evaluate clinical outcome measures. Specifically, her primary interest is in designing tools for children with rare diseases so that their voices and the voices of their families can be prioritized in research.

Dr. Zigler was part of the team that developed the Observer-Reported Communication Ability (ORCA) measure through a partnership with the Foundation for Angelman Syndrome Therapeutics (FAST). The ORCA measure was designed for use in clinical trials to capture caregiver perceptions of communication ability for individuals with Angelman syndrome, a rare neurodevelopmental disorder. Dr. Zigler continues to support the use of the measure in Angelman syndrome, and is currently PI of the FDA-funded study to expand the ORCA measurement model and gather sufficient validity evidence for its use in other neurodevelopmental disorders with similar communication impacts.

Michelle Campbell, PhD

U.S. Food and Drug Administration

Dr. Michelle Campbell is the Senior Clinical Analyst for Stakeholder Engagement and Clinical Outcomes in the Office of Neuroscience, Office of New Drugs (OND) in FDA's Center for Drug Evaluation and Research. Previously, Dr. Campbell was a reviewer on the Clinical Outcome Assessments (COA) Staff and Scientific Coordinator of the COA Qualification Program in OND. Dr. Campbell's focus is in patient-focused drug development and the use of patient experience data in the regulatory setting. Prior to joining FDA, Dr. Campbell spent more than 10 years conducting research in the academic-clinical setting, including five years in a neurology and developmental medicine department. Dr. Campbell earned her BA in Biology from the College of Notre Dame, her MS in Health Science) from Towson University and her PhD in Pharmaceutical Health Services Research from the University of Maryland School of Pharmacy.

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Mike Sidorov, PhD

Children's National Hospital

Mike Sidorov is an Assistant Professor in the Center for Neuroscience Research at Children's National Hospital in Washington, DC. Mike received his Ph.D. in neuroscience at MIT, where he studied mechanisms of plasticity in a mouse model of Fragile X syndrome, and did postdoctoral training in Dr. Ben Philpot's lab at UNC. Mike's lab at Children's National works at the circuit and behavioral levels in rodent models of Angelman syndrome and also works to develop biomarkers using human EEG data. Outside of the lab, Mike is a competitive fast-pitch wiffleball player.

Abigail Dickinson, PhD

UCLA

Dr. Dickinson is an Assistant Research Scientist neurologist specializing in autism and related neurodevelopmental disorders. She is currently junior faculty within the Department of Psychiatry and Biobehavioral Sciences at UCLA and leads the electroencephalography (EEG) lab at the UCLA Center for Autism Research and Treatment. Dr. Dickinson has specialized expertise in high-density EEG and signal processing, and is focused on characterizing neurophysiological biomarkers that can help us to better understand neurodevelopmental disorders. After earning her Ph.D. in the Sheffield Autism Research lab (UK), Dr. Dickinson has sought to develop biomarkers of autism that can act as early indicators in infants at risk for autism, and help us understand individual differences in cognitive trajectories. More recently, Dr. Dickinson has extended the biomarkers she has developed to understand other neurodevelopmental disorders, including Tuberous Sclerosis Complex and Angelman Syndrome. Over the last two years, Dr. Dickinson has collaborated closely with Dr. Mike Sidorov on characterizing specific neural markers in Angelman syndrome. Dr. Dickinson and Dr. Sidorov were delighted to receive a new investigator award from FAST earlier this year, which will support an exciting new project to evaluate EEG biomarkers in a large sample of children with Angelman Syndrome.

Dan Foti, PhD

Purdue University

Dr. Dan Foti is an Associate Professor of Psychological Sciences at Purdue University. He also serves as the Director of T32 Predoctoral Training Program through the Indiana Clinical and Translational Sciences Institute. He is a clinical psychologist by training and is an expert in the clinical translation of EEG methods. For more than 15 years, Dr. Foti has used laboratory-based EEG methods to characterize cognitive and affective functioning in psychiatric disorders, with a focus on understanding processes involved in illness risk and progression. More recently, Dr. Foti has collaborated with Dr. Bridgette Kelleher to adapt gold-standard, laboratory-based EEG methods for home-based assessment.

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Robert Komorowski, PhD

Ionis Pharmaceuticals

Rob received in BA in Psychology from Cornell University and his Ph.D. in neuroscience from Boston University. He did his postdoc at MIT with Dr. Mark Bear focusing on Fragile-X syndrome, Angelman syndrome, and Rett Syndrome. Since then he has worked at Vertex Pharmaceuticals and, most recently, at Biogen where he led projects designed to translate from the mouse to the clinic using EEG. He then decided to make the shift from research to clinical development at Ionis where he works primarily on the Ionis HALOS clinical trial in Angelman syndrome.

Alexandra Key, PhD

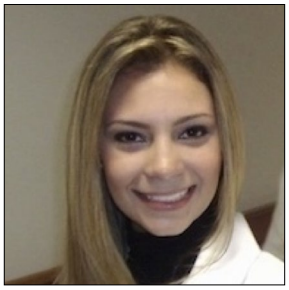
Vanderbilt University Medical Center

Dr. Alexandra P. Key is the Director of Human Psychophysiology lab at the Vanderbilt Kennedy Center (https://vkc.vumc.org/vkc/eeg/). Her research focuses on the development of noninvasive brain-based measures of cognitive, communicative, and social-emotional processes optimized for use in phenotyping and treatment studies in infants, children, and adults with developmental disabilities.

Erick Sell, MD

Children's Hospital of Eastern Ontario

He did a fellowship in Pediatric Neurology and a Fellowship in pediatric Epilepsy at the Hospital for Sick Children, Toronto. Together with Dr Jane Summers started the first Canadian Angelman Syndrome in Ottawa, Canada in 2016. The vision of the Angelman Clinic in Ottawa is to provide clinical expert opinion and access to new clinical therapies for patients and families.



Adriana Gomes, MD

UCSD/Rady Children's Hospital-San Diego

Adriana finished medical school in 2011 in Petrópolis- Rio de Janeiro- Brazil. In 2014 she completed her Residency in Pediatrics, followed by a Master's in Child and Maternal Health at the Universidade Federal Fluminense in Niterói- Rio de Janeiro- Brazil. In 2015 became a faculty member at the Faculdade de Medicina de Petrópolis.

Adriana moved to San Diego in 2021; she is ECFMG certified and is currently a visiting scholar at UCSD's Genetics and Dysmorphology department. Among her research interests is Angelman syndrome. Along with Dr. Lynne Bird, Adriana is a co-investigator of the Natural History Study of Angelman syndrome and the Tangelo and Ionis Clinical Trials.

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Kelly David

FAST

Kelly David, M.A., CCC-SLP, CBIS received her BS in Biology and Master of Arts degree in Communication Sciences and Disorders from the University of Central Florida. She has served populations from pediatric to geriatric, with developmental and/or acquired disorders. She currently works as a clinical instructor at the University of Central Florida's Communication Sciences and Disorders Clinic with individuals across the lifespan. She volunteers as co-Vice Chairperson for the Foundation for Angelman Syndrome Therapeutics (FAST). Her most important role, however, is as mother to Colin and Madeline. Her son, Colin, lives with Angelman syndrome.

Amanda Moore

Angelman Syndrome Foundation

Amanda Moore has served as the CEO of the Angelman Syndrome Foundation since 2019. She spent most of her career working in nonprofit leadership and development for the YMCA. In 2015, Amanda and her husband Adam were blessed with adopting twin boys Jackson and Baden. In 2016, Jackson was diagnosed with Angelman Syndrome and at that time Amanda vowed to take action to do what she could for Jackson and individuals like him. She served on the Angelman Syndrome Board for one year and now is serving in the role as CEO of the Angelman Syndrome Foundation.

Rob Carson, MD, PhD

Vanderbilt University Medical Center

Dr. Robert Carson is an Assistant Professor of Pediatrics at Vanderbilt University Medical Center with subspecialty training in pediatric neurology and epilepsy. He currently directs the Angelman Syndrome/ Dup15q clinic at Vanderbilt. His work in Angelman Syndrome first began in collaboration with Dr. Mark Grier in the lab of Dr. Andre Lagrange investigating myelination and mTOR signaling in Angelman Syndrome Model mice. Since that initial basic science exposure, his clinical focus has transitioned to the study of Angelman Syndrome and dup15q syndrome. He has been involved in both investigator-initiated and pharma-supported clinical trials in Angelman Syndrome. His current focus is on generating a better understanding of epilepsy and nonepileptic myoclonus in Angelman Syndrome. As part of this work, he plans to focus on the identification of existing FDA-approved medications which can be rapidly repurposed for use in the treatment of nonepileptic myoclonus.

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David J. Segal, PhD

University of California, Davis

David Segal received his Ph.D. from the University of Utah performed a post-doc at The Scripps Research Institute. Now at UC Davis, Dr. Segal's research focuses on gene and epigenome editing to treat Angelman syndrome and related neurologic disorders. He is a founding member of the Interventional Genetics Program at the UC Davis MIND Institute, an investigator in the NIH Somatic Cell Genome Editing Consortium, and Editor-in-Chief of Frontiers in Genome Editing.

Henriette O'Geen, PhD

University of California, Davis Genome Center

Henriette O'Geen is a Senior Project Scientist at the UC Davis Genome Center and a member of the Interventional Genetics Team. As part of nationwide collaborative efforts (ENCODE and REMC) she has used genomics approaches to study transcriptional networks and their relevance in development, differentiation and cell identity. Exploration of the role of epigenetics has proven to be informative for better understanding of disease and epigenetic mechanisms. These collaborative epigenomic efforts laid the foundation for the epigenome editing research program that she has established in the laboratory of Dr. David Segal at UC Davis. Epigenetic changes are the underlying cause for many human diseases, including Angelman syndrome, Fragile X syndrome, Prader-Willi syndrome, and various cancers. Dr. O'Geen's current research is focused on understanding and predicting the rules of heritable epigenetic reprogramming of transcription. This offers the unique opportunity to understand basic biological processes and to provide new insights for the treatment of human disease.

Maria Clarke, BS

University of California San Francisco

I am a Staff Research Associate in Tippi Mackeznie's Lab at the University of California San Francisco. I received my Bachelor's of Science in Neurobiology from UC Davis and have been working in the Mackenzie Lab for 3 years. Our lab is a part of UCSF's Center for Maternal Fetal Precision Medicine and The Eli and Edythe Broad Center of Regeneration Medicine and Stem Cell Research. We focus on developing prenatal therapies for rare genetic disorders.

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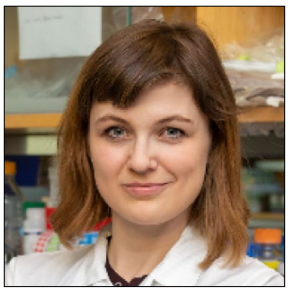

Ulrika Beitnere, PhD

University of California, Davis Genome Center

Ulrika is an experienced translational researcher with extensive experience in small molecule drug, zinc finger and CRISPR development for treating brain disorders. Since joining Segal's lab, she has been working on various approaches for treating Angelman Syndrome and delivering drugs to the brain safely.

She received her Ph.D. in Pharmacology from the University of Latvia. She is a co-author of 18 publications and is also the recipient of the Career Development Award in 2019 from the American Society of Gene and Cell Therapy (ASGCT). It is Ulrika's passion to explore the best long-term treatment tools for rare disorders.

Joerg Hipp, PhD

Roche

Dr. Joerg Hipp is a biomarker and experimental medicine leader at Roche. He has a background in Physics and Neuroscience. Before joining Roche, his main research focus was to understand the role of neuronal oscillations in normal and pathological brain function using different techniques including EEG, MEG and MRI. At Roche, he is responsible for the biomarker strategy and implementation in several neurodevelopmental disorder programs and heads a group that bundles clinical electrophysiology and sleep expertise to support the neuroscience portfolio.

Maria-Clemencia Hernandez, PhD

Hoffman-La Roche Ltd

Maria-Clemencia Hernandez, PhD, is a Distinguished Scientist in the Neuroscience & Rare Diseases department at Roche Pharma Research and Early Development.

Maria-Clemencia has more than 20 years' experience in drug discovery at Roche. Her research focus has been on inhibitory neurotransmission in neurological and psychiatric disorders. In particular, her work in the GABAA receptor field has contributed to the identification of novel GABAA subtype selective modulators including basmisanil and alogabat, two assets currently in phase 2 clinical studies.

Before joining Roche in November 1998, Maria-Clemencia was a Postdoctoral fellow in Molecular Neuro-Oncology at the University of California, San Francisco.

Maria-Clemencia obtained a BS degree at the Javeriana University in Bogota, Colombia, an MSc in Neurobiology from the Weizmann Institute of Science in Israel, and a PhD in Biochemistry from the University of Geneva, Switzerland.

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Wayne Chadwick, PhD

HEALX

Dr. Wayne Chadwick is a Principal Pharmacologist at Healx Ltd, an AI driven drug discovery company focused on identifying new treatments for rare diseases. Wayne received a doctorate in Biochemistry from the University of Port Elizabeth, South Africa, before moving to the United States to complete a postdoctorate in neuroscience at the NIH. Wayne has authored more than 30 scientific publications and book chapters and has over 10 years' experience in the pharmaceutical industry where he has led a number of drug discovery projects in neurodegenerative diseases and neurodevelopmental disorders.

John Marshall, PhD

Brown University

Dr. Marshall holds the position of full Professor of Medical Science at Brown University in the Department of Molecular Biology, Cell Biology & Biochemistry. He has a broad background in pharmacology, with specific training in molecular biology and electrophysiology techniques studying signal transduction pathways using electrophysiological (slice and patch clamping) and genetic approaches with a focus in developmental disorders, such as autism and intellectual disabilities. He studies BDNF signaling, LTP and behavior in rodents. The long- range goal for his research is to contribute to the development of therapeutic interventions that will enhance cognition in childhood neurodevelopmental disorders.

Timothy Fenton

University of California, Davis

Tim Fenton is a third year Pharmacology and Toxicology PhD Student in the Silverman Laboratory at the University of California, Davis. He received his BS in Cell Biology from UC Davis in 2012 and his MS in Applied Anatomy from Case Western Reserve University in 2016. Mr. Fenton's doctoral research aims to establish outcome measures for identifying and testing therapeutics in vitro using primary neuronal cultures and in vivo using rodent behavioral testing for the treatment of Angelman Syndrome. He is interested in finding structural and electrophysiological biomarkers in vitro and testing novel and repurposed therapeutics in vivo to accelerate preclinical drug development for AS.

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Scott Dindot, PhD

Texas A&M College of Medicine

Scott Dindot is an Associate Professor of Genomics and EDGES Fellow at Texas A&M University and the Executive Director of Molecular Genetics at Ultragenyx Pharmaceutical. He holds appointments in the Texas A&M College of Veterinary Medicine & Biomedical Sciences and the College of Medicine and is a member of the graduate faculty in Genetics, Biomedical Science, and Neuroscience. He performed his undergraduate studies at Howard Payne University and then at Texas A&M University, where he received a Bachelor of Science degree in Molecular and Cell Biology. He received a Doctor of Philosophy in Genetics from Texas A&M University under the guidance of Jorge Piedrahita and then performed his postdoctoral training as an NIH Fellow at Baylor College of Medicine under the direction of Dr. Arthur Beaudet.

Dr. Dindot's research focuses on the genetic and epigenetic basis of disease, with an emphasis on the study of genomic imprinting disorders. His lab currently focuses on understanding the imprinted regulation of the Angelman syndrome gene, developing novel therapies to treat Angelman syndrome patients, and understanding the pathophysiology of Angelman syndrome using animal models.

Z. Begum Yagci

North Carolina State University

Z. Begum Yagci is a BS and MS Chemical Engineering graduate of Bogazici University in Istanbul, Turkey. She is currently pursuing a PhD in Chemical and Biomolecular Engineering in the Keung Lab at North Carolina State University in Raleigh, NC, USA. There she is working on engineering isogenic models for Angelman Syndrome Class I/Class II deletion. She is also part of a collaborative project within Keung Lab in which they investigate how the absence of UBE3A affecting the cell-type composition in human cerebral organoids. Ms. Yagci was awarded the NC State University's Provost Doctoral Fellowship.

Dr. M. Rance MBBS BSc(Hons), MRCS(Eng)

PTC Therapeutics

Dr. Rance is Senior Director Clinical Development and Clinical Lead for Angelman Syndrome Gene Therapy Program at PTC Therapeutics.

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Luis Martinez, PhD

Baylor College of Medicine/Texas Children's Hospital

Luis Martinez, postdoc in the lab of Dr. Anne Anderson at the Neurological Research Institute at Texas Children's Hospital and Baylor College of Medicine, characterizing seizures and EEG abnormalities in Angelman syndrome with focus on febrile seizures and neuroimmune response.

Anne C. Wheeler, PhD

RTI International

Dr. Anne Wheeler is a neurodevelopmental psychologist and senior research analyst at RTI International, where she conducts multiple research projects focused on the development of tools and strategies to improve outcomes for individuals with intellectual/developmental disabilities, especially those with rare monogenetic/neurogenetic conditions. She has been involved in research on focused on individuals with neurogenetic conditions and their families for over 20 years. She is the director of the LADDER database, an effort to curate and harmonize data collected on individuals with Angelman or Dup15q syndromes around the world; is working on efforts to identify and improve outcome measures used to determine change in clinical trials for rare conditions and leads activities to reduce the age of diagnosis and provide targeted early intervention and support services for children diagnosed in infancy.

Dr. Wheeler is also an Adjunct Associate Professor of Psychiatry and School Psychology at the University of North Carolina at Chapel Hill and a practicing licensed psychologist at the Carolina Institute for Developmental Disabilities where she provides developmental and behavioral consultation for individuals with Angelman and Dup15q syndromes. She has been involved in clinical service provision and research focused on developmental and familial outcomes for children with genetic diagnoses for over 15 years.

Mindy Leffler, MEd

Casimir/Emmes

Mindy Leffler, M.Ed., Is the President of Casimir and plays a central role in the operations of Casimir, including writing of grants and business relations with pharmaceutical companies and research institutions. She spearheaded the Duchenne Muscular Dystrophy Video Assessment tool to assess quality of movement in Duchenne muscular dystrophy. Ms. Leffler has been fundamental in establishing the use of videos to assess neurodevelopment as a concept of interest. Along with Drs. Tan and Sadhwani, Ms. Leffler developed the NVA clinician reported outcome measure for Angelman syndrome in terms of identifying the tasks as well as the development of Manual of Instructions. Ms. Leffler has an active role in the development of the Angelman Video Assessment scorecards, led the Delphi panels, and integrated feedback from the Delphi panels in the development of the final scorecards. She will contribute to the data analysis and manuscript preparation.

Jake Donaghue, MD, PhD

Beacon Biosignals

Jake translates scientific innovation into clinical interventions. He recently completed his MD from Harvard Medical School and PhD in neuroscience from MIT, leading research into the effects of neuroactive compounds on brain network activity. His published works span epilepsy, cognition and machine learning methods for quantifying pharmacological effects on neural activity.