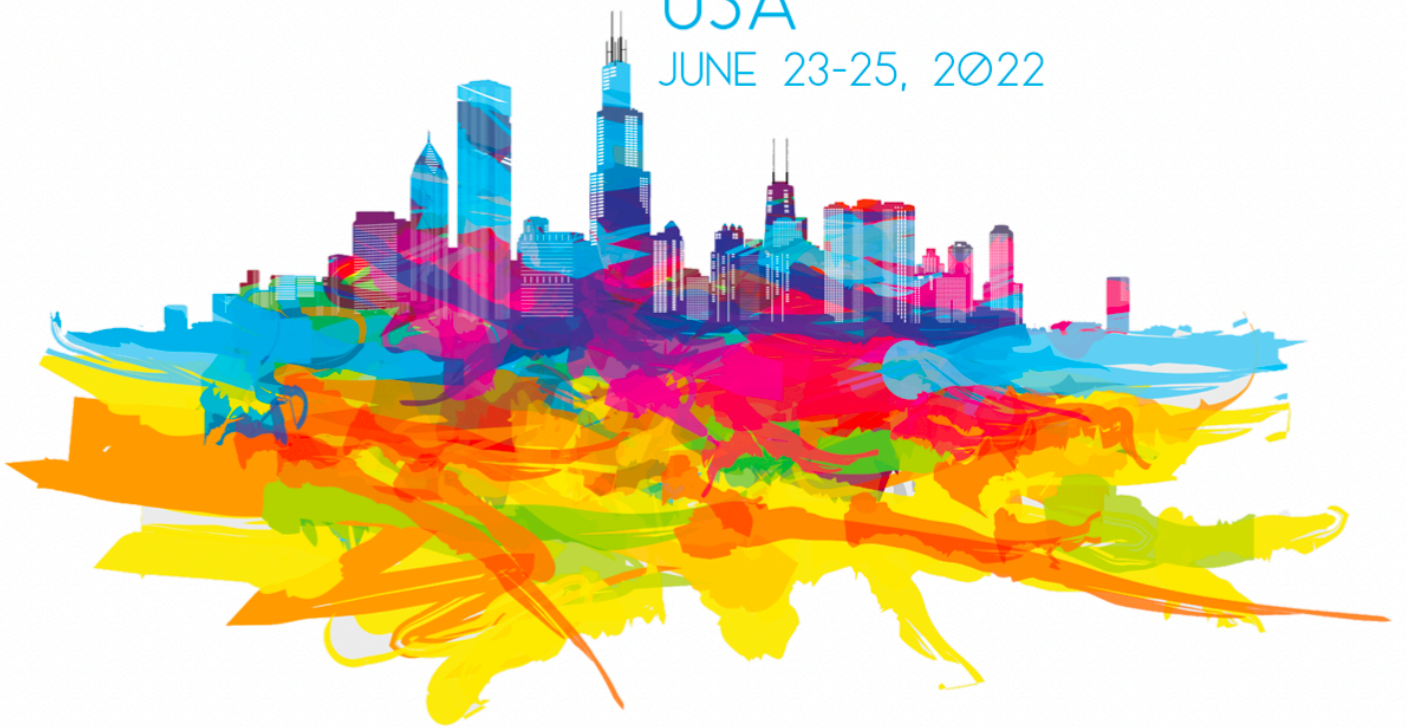


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PITT HOPKINS RESEARCH SYMPOSIUM & FAMILY CONFERENCE

CHICAGO, ILLINOIS
USA

JUNE 23-25, 2022



CONFERENCE PRESENTED BY:



PITT HOPKINS
RESEARCH FOUNDATION

Don't just hope for MIRACLES, FIGHT for THEM!



PHRF Mission Statement

The mission of the Pitt Hopkins Research Foundation (PHRF) is to support research dedicated to finding a treatment, and an eventual cure of Pitt Hopkins syndrome and other similar disorders. The PHRF is also dedicated to supporting the Pitt Hopkins community with resource recommendations, parental support and the latest medical information.

The Board of Directors of the Pitt Hopkins Research Foundation is comprised of individuals dedicated to advancing research and supporting families of children and adults with Pitt Hopkins Syndrome. The Board is actively engaged in fundraising efforts towards research, in providing parental support to families worldwide, and in offering communication and media representation for the Foundation.

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PITT HOPKINS

RESEARCH FOUNDATION

Don't just hope for MIRACLES, FIGHT for THEM!

Thursday, June 23rd, 2022

Conference site: **Embassy Suites by Hilton Chicago O'Hare Rosemont 5500 N River Rd, Rosemont, IL 60018**

9:00 – 9:05 a.m. **Welcome**
Audrey Davidow Lapidus
President, Pitt Hopkins Research Foundation

SESSION I

9:05 – 9:30 a.m. **Regulation of TCF4-dependent gene expression in the nervous system**
Tõnis Timmusk, PhD - *Virtual*
Department of Chemistry and Biotechnology
Tallinn University of Technology, Estonia

9:30 – 9:55 a.m. **A Brain-on-Chip (BoC) platform for preclinical assessment of CRISPRa as a potential treatment for PTHS**
Gad Vatine, PhD - *Virtual*
Physiology and Cell Biology Department
Regenerative Medicine and Stem Cell (RMSC) Research Center
Ben-Gurion University of the Negev

9:55 – 10:20 a.m. **Transcription Factor 4 regulates the density and connectivity of specific subclasses of inhibitory neurons**
Brady Maher, PhD
Lieber Institute for Brain Development
Johns Hopkins Medical School

10:20 – 10:35 a.m. **Questions for Session I Talks**

10:35 – 10:50 a.m. *Break*
Coffee and refreshments available

SESSION II

- 10:55 – 11:20 a.m. **Therapeutic strategies for Pitt-Hopkins syndrome**
Ben Philpot, PhD - *Virtual*
University of North Carolina at Chapel Hill
UNC Neuroscience Center
- 11:20 – 11:45 a.m. **Rescuing molecular, cellular and network alterations in Pitt Hopkins brain organoids**
Alysson Muotri, PhD - *Virtual*
Director of the UCSD Stem Cell Program
University of California San Diego, School of Medicine
- 11:45 – 12:10 p.m. **In vitro characterization and drug candidate testing on Pitt Hopkins Patient astrocytes and neurons**
Kathrin Meyer, PhD
Center for Gene Therapy, The Research Institute at Nationwide Children's Hospital
- 12:10 – 12:25 p.m. **Questions for Session II Talks**
- 12:30 – 1:30 p.m. *Lunch*

SESSION III

- 1:30 – 1:55 p.m. **ssRNA therapy for Pitt Hopkins syndrome**
Michelina Iacovino, PhD
Associate Professor of Pediatrics
The Lundquist Institute Harbor-UCLA Medical Center
- 1:55 – 2:20 p.m. **TET Enzyme Inhibition to Enhance Cognition**
Andrew Kennedy, PhD
Assistant Professor of Chemistry and Neuroscience
Bates College
- 2:20 – 2:45 p.m. **Preclinical assessment of CRISPR-mediated gene regulation to rescue haploinsufficiency in Pitt Hopkins syndrome**
Kyle Fink, PhD
Neurology Department & Stem Cell Program
University of California, Davis Medical Center
- 2:45 – 3:00 p.m. **Questions for Session III Talks**
- 3:00 – 3:15 p.m. *Break*

SESSION IV

- 3:20 – 3:45 p.m. **Rescue of behavioral phenotypes in Tcf4+/- mice by overexpression of MeCP2**
Colleen Niswender, PhD
Professor of Pharmacology and Warren Center for Neuroscience
Drug Discovery
Vanderbilt University School of Medicine
- 3:45 – 4:10 p.m. **Microbiota Transfer Therapy for Children with Pitt Hopkins Syndrome**
James B. Adams, PhD
Director of Autism Research Program, Arizona State University
Rosa Krajmalnik-Brown, PhD
Director, Biodesign Institute, Arizona State University
- 4:10 – 4:35 p.m. **A Portrait of Pitt Hopkins Patients from the PTHS Clinic at Colorado Children’s Hospital**
Jessica Duis, MD
Associate Professor of Pediatrics & Medical Genetics
Director of Pitt-Hopkins Center of Excellence at Children’s
Hospital Colorado
- 4:35 – 5:00 p.m. **Rescuing TCF4 behavioral and neuronal spine phenotypes in mice**
Patricia Cogram, PhD - *Virtual*
Associate Professor
Universidad de Chile, Chile
- 5:00 – 5:15 p.m. **Questions for Session IV Talks**

Adjourn
-
- 5:00 – 6:30 p.m. **Family Meet & Greet**
Join us in the atrium for complimentary appetizers and drinks until 6:30 p.m.



Friday, June 24th, 2022

8:00 – 8:10 a.m. **Welcome**
Theresa Pauca
Vice President, Pitt Hopkins Research Foundation

LAY TALKS & PARENT INFORMATION

8:10 – 8:30 a.m. **A Foundation's Journey through Drug Development: The Need to Move FAST**
Allyson Berent, DVM, Dipl. ACVIM - *Virtual*
CSO, Foundation for Angelman Syndrome Therapeutics
COO, GeneTx Biotherapeutics
Director Interventional Endoscopy Services, The Animal Medical Center
Mother to Quincy: A beautiful little girl living with Angelman syndrome

8:30 – 8:50 a.m. **Pitt Hopkins 101 - An overview of the Therapeutic Landscape for Pitt Hopkins Syndrome**
Audrey Davidow Lapidus
President, Pitt Hopkins Research Foundation

8:50 – 9:05 a.m. **Gene Therapy From Concept To Creation**

Step 1 – Proving gene therapy works in Brain Organoid Models
Alysson Muotri, PhD - *Virtual*
Director of the UCSD Stem Cell Program
University of California San Diego, School of Medicine

9:10 – 9:25 a.m. **Step 2 - Taking the findings into mouse models**
Patricia Cogram, PhD - *Virtual*
Associate Professor
Universidad de Chile, Chile

9:30 – 9:45 a.m. **Step 3 - Next Steps, From mouse model to human**
Yael Weiss, PhD
Chief Executive Officer
Mahzi Therapeutics

9:45 – 10:00 a.m. *Questions*

10:00 – 10:15 a.m. *Break*

- 10:15 – 10:30 a.m. **NNZ-2591 as a Treatment for Pitt Hopkins Syndrome**
Nancy Jones, PhD - *Virtual*
Vice President, Clinical Development
Neuren Pharmaceuticals
- 10:30 – 10:35 a.m. *Questions*
- 10:40 – 10:55 a.m. **Treatment options from the Colorado Clinic**
Jessica Duis, MD
Associate Professor of Pediatrics & Medical Genetics
Director of Pitt-Hopkins Center of Excellence at Children's
Hospital Colorado
- 10:55 – 11:00 a.m. *Questions*
- 11:05 – 11:25 a.m. **Microbiota Transfer Therapy for Children with Pitt Hopkins Syndrome**
James B. Adams, PhD
Director of Autism Research Program, Arizona State University
Rosa Krajmalnik-Brown, PhD
Director, Biodesign Institute, Arizona State University
- 11:25 – 11:35 a.m. *Questions*
- 11:35– 11:55 a.m. **Moving Forward – A Roadmap to Treatment**
Audrey Davidow Lapidus
President, Pitt Hopkins Research Foundation
- 12:00 – 1:00 p.m. *Lunch in the atrium*
- 1:00 – 2:45 p.m. **Mom Break-Out Group*** lead by Diane Krell
PTHS Mom and Director, Pitt Hopkins Research Foundation
- 3:00 – 4:30 p.m. **Dad Break-Out Group*** lead by Martin Cuevas
PTHS Dad and Clinical Coordinator
- *Please note: Dad & Mom Breakout Sessions are strictly for parents (no children), so please plan accordingly.
- 4:30 – 4:45 p.m. **From Overwhelmed to Empowered: A Journey Towards “Peace of Mind” and a 3-Month Solo Vacation**
Ingrid Harding, PTHS Mom and Co-founder of Rett syndrome Research Trust and Girl Power 2 Cure
- 4:45 – 5:30 p.m. **Transition to Adulthood Panel**
Lead by PTHS moms
Nicole Dyehouse
Ingrid Harding
Connie McNay

5:30 p.m. **Group Photo in the atrium of the hotel**

Adjourn

Guests of the Embassy Suites have a complimentary evening reception available from 5:00 - 6:30 p.m.

Dinner on your own.

8:00 p.m. Viewing of *Growing Together*, a disability documentary by Sofia Pauca, Pitt Hopkins Sibling



Saturday, June 25th, 2022

8:30 – 9:45 a.m. **Making Sense of Sensory Processing - It's all connected!**
Kelly Beins, OTR/L
Strategic Clinical Consultant

9:45 – 10:00 a.m. *Break*

10:00 – 10:30 a.m. **A Family's Journey with AAC**
Jessica Fletcher
PTHS mom, Vice President, Pitt Hopkins Research Foundation

10:30 – 12:00 p.m. **Fundamentals of Spelling to Communicate and Presuming Competence**
Kelly Berg
S2C Practitioner

12:00 – 1:00 p.m. *Lunch in the atrium*

1:00 – 1:30 p.m. **Parent Talks: Getting Started with AAC**
Nicole Anderson
Director, Pitt Hopkins Research Foundation

This session will focus on a range of tools and strategies for individuals who have Pitt Hopkins to use to increase their communication and participation skills. Concrete ideas for knowing where to start and how to start in supporting individuals with Pitt Hopkins will be discussed, and keys to successful implementation of tools and strategies will be shared. Participants will leave with practical ideas and suggestions that they can use with their family members.

15 min Q & A

1:45 – 2:30 p.m. **Parent Talks: Beyond Requesting, why literacy matters and how to get there?**
Audrey Davidow Lapidus
President, Pitt Hopkins Research Foundation

This talk is targeted to intermediate AAC users who want to take the next step or who feel a little stuck in the world of basic AAC. You've mastered "I want French Fries" and "I Feel Sad." But what about novel sentence generation? Or having your child share sentences about their hopes and fears? For that, what you need is something simpler than most grid AAC Devices: 26 Letters! This talk will focus on promoting literacy and how to best support various motoric levels to help aide your child in sharing their thoughts on a deeper level.

15 min Q & A

2:45 – 3:45 p.m. **IEPs and how to advocate for your child**
Traci Green, Director, Pitt Hopkins Research Foundation, M. Ed
Special Education
Jessica Fletcher, Vice President, Pitt Hopkins Research
Foundation, M. Ed

This talk will focus on preparing for IEP meetings and understanding your parental rights, from placement and educational program options to advocating for your child in the education setting. We will also work together on creating an "All About Me" sheet, to help ease into new schools, new teachers, new therapists, etc.

3:45 p.m. Conference Close

In a separate room:

• 9:45 a.m. - 10:30 a.m. Spanish Speaking Group Zoom Lead by Paul Pauca, PTHS dad and co-founder of the PHRF



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Abstracts & Participant Biosketches



Timmusk Lab

Tõnis Timmusk, Ph.D.

Professor

Chair of Molecular Biology

Department of Chemistry and Biotechnology

Tallinn University of Technology

E-mail: tonis.timmusk@ttu.ee

Tõnis Timmusk (TT) is a molecular neurobiologist with with more than 30-years research interests in gene regulation and signaling in the nervous system. After obtaining PhD in medical sciences from Karolinska Institutet, Sweden in 1994 he continued his scientific career in Karolinska Institute and Uppsala University in Sweden and in University of Helsinki in Finland.

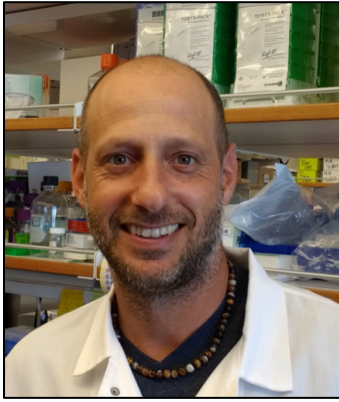
Since 2002 he is full professor of molecular biology in Tallinn University of Technology, Estonia. TT has published more than 100 publications, including in *Nature*, *Science*, *Nature Genetics*, *Neuron*, *Journal of Neuroscience*, *eLife* and others. His published work has contributed to the understanding of the function, signaling and gene regulation of neurotrophic factors of nerve growth factor (NGF) and glial-cell derived neurotrophic factor (GDNF) family ligands and their receptors and to activity-dependent transcriptional mechanisms that underlie nervous system function in health and disease. TT has also published studies of several nervous system diseases, including Parkinson and Huntington diseases, schizophrenia and cognitive disorders. His lab is also studying the function and mechanism of action of the bHLH transcription factor TCF4, the gene of which is mutated in Pitt-Hopkins syndrome (PTHS) and associated with schizophrenia. TT has been the principal investigator of many projects, including the highly prestigious Wellcome Trust International Senior Research Fellowship, and the inventor of several international patent and patent application families. He is the scientific co-founder of two biotech companies.

Title: *Regulation of TCF4-dependent gene expression in the nervous system*

Abstract: Pitt-Hopkins syndrome (PTHS) is a cognitive disorder caused by de novo genetic mutations of the transcription factor 4 (TCF4) gene that encodes a basic helix-loop-helix transcription factor. We have previously demonstrated that human TCF4 gene is transcribed using numerous 5' exons yielding in TCF4 protein isoforms with different N-termini that vary in their ability to regulate transcription. Considering the diversity of TCF4 isoforms, it is important to know which isoforms are expressed at the critical period of brain development when gene therapies for PTHS are planned to be developed. Additionally, we have found that PTHS-associated mutations impair the functions of TCF4 by diverse mechanisms. Furthermore, our previously published data show that neuronal activity leads to phosphorylation of TCF4 and activation of its transcriptional activity, indicating that synaptic activation of nerve cells regulates TCF4 function. In the present study, to further understand regulation of TCF4-dependent gene expression in the nervous system, we have aimed to (1) characterize developmental expression pattern of TCF4 mRNA and protein isoforms in different embryonic and postnatal stages of mouse, rat and human development; (2) analyze the functional impact of TCF4 mutations/variations associated with PTHS, mild to moderate intellectual disability and schizophrenia; (3) identify neuronal activity-regulated TCF4 target genes in cortical neurons using whole transcriptome analysis by RNA-seq and ChIP-seq; (4) identify TCF4 interaction partners in cortical neurons involved in TCF4-dependent transcription.

Gad Vatine, Ph.D.

Assistant Professor



Physiology and Cell Biology Department and Regenerative
Medicine and Stem Cell (RMSC) Research Center
Ben-Gurion University of the Negev
E-mail: vatineg@bgu.ac.il

Dr. Vatine received his Ph.D. in 2011 from Tel Aviv University where, under the guidance of Dr. Yoav Gothilf, he studied the mechanisms underlying light-entrainment of the circadian clock using zebrafish as a model. He followed this with a short postdoc at Bar-Ilan University in the laboratory of Dr. Lior Appelbaum where he established a zebrafish model for the rare psychomotor disability disorder MCT8-deficiency. Next, he joined the laboratory of Dr. Clive Svendsen at the Regenerative Medicine Institute at Cedars-Sinai where he pioneered the concept of disease modeling at blood-brain barrier (BBB) using patient-specific induced pluripotent stem cells (iPSCs). In 2017, he became an assistant professor at Ben-Gurion University. The Vatine lab generates personalized bioengineered platforms, such as Organs-on-Chips, to study mechanisms underlying various rare neurological disorders, and to test potential treatments.

Title: *A Brain-on-Chip (BoC) platform for preclinical assessment of CRISPRa as a potential treatment for PTHS*

Abstract: The development of technologies to reprogram cells into a pluripotent stage, and their subsequent ability to differentiate to any cell type opened opportunities for modeling the human brain in a personalized manner. However, in vitro cell culture systems often fall short in representing the complexity of the physiological environment. Recently, bioengineered platforms known as Organ-on-Chip (OoC) have emerged as a promising approach to improve the physiological relevance of cells grown in culture. By mimicking the multicellular architectures, tissue-tissue interfaces, physicochemical microenvironments and vascular perfusion of the body, OoCs produce levels of tissue and organ functionality not possible with conventional 2D or 3D culture systems. We have recently combined iPSCs with OoC technology to generate a personalized Brain-on-Chip (BoC) platform, which enables to study genetic neurological disorders in a more physiologically relevant manner and to test potential therapeutics.

We have generated and characterized iPSCs from four PTHS patients carrying a variety of heterozygous mutations in TCF4, and from their sex-matched healthy family relatives. iPSCs were differentiated into neural progenitor cells, which were seeded and differentiated within the OoC to form a PTHS-BoC-on-Chip. In the future we will use this platform for testing potential gene therapy approaches for PTHS.



Maher Lab

Brady J. Maher Ph.D.

Lead Investigator

Lieber Institute for Brain Development

Assistant Professor

Department of Psychiatry and Behavioral Sciences; Department
of Neuroscience Johns Hopkins School of Medicine

E-mail: brady.maher@libd.org

Brady J. Maher, Ph.D. is a Lead Investigator in the Developmental Neurobiology and Functional Genomics division of the Lieber Institute and an Associate Professor in the Department of Psychiatry and Behavioral Sciences and the Solomon H. Snyder Department of Neuroscience at the Johns Hopkins University School of Medicine.

Dr. Maher's research is focused on understanding fundamental mechanisms involved in brain development and brain function with an emphasis on how dysfunction in these mechanisms can result in neurodevelopmental and psychiatric disorders. By focusing on key developmental genes that are associated with psychiatric risk, his research group is both enhancing our primary understanding of brain development while also making significant inroads into identifying pathophysiological mechanisms underlying psychiatric disorders.

A major focus of his lab is to understand the function of Transcription Factor 4 (TCF4) gene. TCF4 is a clinically pleiotropic gene having association with schizophrenia and autism spectrum disorder (ASD). Autosomal dominant mutations in TCF4 result in Pitt Hopkins syndrome, a rare neurodevelopmental disorder with a variety of symptoms including developmental delays, intellectual disability, absent speech, and breathing abnormalities. His group has shown that TCF4 is an activity-dependent transcription factor that is a critical regulator of cortical development. They have shown TCF4 regulates several developmental steps including cell fate specification, neuronal migration, cortical column formation, and neuronal excitability.

Recently, Dr. Maher's group demonstrated that TCF4 directly regulates oligodendrocyte development and myelination. This work has led to the hypothesis that defects in myelination are a common pathophysiology across the autism spectrum. His research group is now working on genetic and pharmacological approaches to rescue myelination in PTHS models, with the ultimate goal of applying these therapeutic approaches to PTHS and more broadly to ASD.

Title: *Transcription Factor 4 regulates the density and connectivity of specific subclasses of inhibitory neurons*

Abstract: Transcription Factor 4 (TCF4) is a type I basic helix-loop-helix transcription factor and autosomal dominant mutations in TCF4 causes Pitt-Hopkins Syndrome (PTHS), a neurodevelopmental disorder with autistic features. Our understanding of the resulting pathophysiology from TCF4 haploinsufficiency or expression of a dominant negative TCF4 protein remains incomplete. During embryonic development, GABAergic interneurons originate from the proliferative zones of the ventral forebrain and in situ hybridization suggests Tcf4 transcript is enriched in this brain region. In addition, single cell RNA sequencing data indicates Tcf4 is highly expressed across many subclasses of GABAergic interneurons and TCF4 is a known dimerization partner of ASCL1 (or MASH1), a critical regulator of interneuron development. Therefore, we hypothesized that TCF4 may play a critical role in regulating interneuron development and function. To test this idea, we quantified GABAergic interneuron density in a PTHS mouse model that harbors a truncated Tcf4 gene (Tcf4+tr) and compared it to WT littermates. Immunohistochemistry (IHC) for the major cortical GABAergic markers showed a significant decrease in the PV+ population across several brain areas including medial prefrontal cortex, primary motor cortex, striatum, and basal lateral amygdala. Likewise, qPCR and cell-type specific expression analysis (CSEA) of bulk RNAseq data also predicted a significant decrease in the proportion of PV+ cells. Interestingly, this deficit was specific to PV+ cells, as IHC and CSEA analysis of SST+ interneurons was not different. Furthermore, we quantified interneuron density by crossing a tdTomato reporter line with a variety of GABAergic subclass-specific cre lines and observed that Tcf4 mutation resulted in a reduction in PV+ interneurons across all brain regions measured, while VIP+ cells showed a significant reduction only in the motor cortex and SST+ interneuron density was unchanged. Lastly, we characterized the impact Tcf4 mutation had on the physiology of PV+ interneurons by performing targeted whole-cell electrophysiology. We observed no differences in the intrinsic membrane properties, however the frequency of spontaneous inhibitory synaptic currents was significantly reduced. Together, these results suggest that the density and connectivity of inhibitory neurons in the PTHS mouse is perturbed, and these changes may in part underlie the cognitive deficits and autistic features present within this patient population.

Philpot Lab



Ben Philpot, Ph.D.

Kenan Distinguished Professor

Associate Director, UNC Neuroscience Center

Department of Cell Biology & Physiology

E-mail: bphilpot@med.unc.edu

Dr. Ben Philpot is a Kenan Distinguished Professor in the Neuroscience Center and Department of Cell Biology & Physiology at the University of North Carolina. He earned his Ph.D. in psychobiology from Dr. Peter Brunjes at the University of Virginia and performed a postdoctoral fellowship in the laboratory of Dr. Mark Bear at Brown University and M.I.T., where he made important contributions to our understanding of experience-dependent brain development. He is currently the Associate Director of the UNC Neuroscience Center and a member of the Carolina Institute for Developmental Disabilities, for which he helps direct a cross-disciplinary postdoctoral training grant for neurodevelopmental disorders. Dr. Philpot's current research seeks to understand the pathophysiology underlying monogenic neurodevelopmental disorders, and he uses this information to develop small molecule and gene therapies to treat these disorders. His research focuses on early-stage development of treatments for Pitt-Hopkins, Dup15q, and Angelman syndromes. Dr. Philpot has made key therapeutic discoveries, including developing an approach to unsilence the epigenetically-repressed paternal UBE3A allele as a novel treatment strategy for Angelman syndrome. Dr. Philpot has >90 peer-reviewed scientific publications. He has advised prominent biotech and pharmaceutical companies, and serves on the scientific advisory committee for the Angelman Syndrome Foundation. He has won multiple awards, including the NARSAD Young Investigator Award, a Whitehall Foundation fellowship, and the Dr. Claudia Benton Award for Scientific Research, and is currently a SFARI Investigator of the Simons Foundation.

Title: *Therapeutic strategies for Pitt-Hopkins syndrome*

Abstract: Pitt-Hopkins syndrome (PTHS) is caused by haploinsufficiency of TCF4. Accordingly, a promising therapeutic strategy is to normalize TCF4 levels. Such an approach first requires an in-depth understanding of the spatio-temporal and cell type-specific distribution of TCF4 across brain development. Moreover, genetic interventions also require having appropriate preclinical PTHS models and identifying developmental windows for successful interventions. Here I will provide an overview of my lab's efforts to provide foundational insights that will guide genetic interventions, as well as our lab's efforts to develop clinically-relevant genetic therapies.



Muotri Lab

Alysson Muotri, Ph.D.

University of California San Diego, School of Medicine

Professor of Pediatrics

Professor of Cellular & Molecular Medicine

Director of the UCSD Stem Cell Program

Director of the UCSD Archealization Center

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Dr. Muotri is a professor at the Departments of Pediatrics and Cellular & Molecular Medicine at UC San Diego. He is also the Director of the Stem Cell Program and Archealization Center. Dr. Muotri earned a BSc in Biological Sciences from the State University of Campinas in 1995 and a Ph.D. in Genetics in 2001 from University of Sao Paulo, in Brazil. He moved to the Salk Institute as Pew Latin America Fellow in 2002 for a postdoctoral training in the fields of neuroscience and stem cell biology. His research focuses on brain evolution and modeling neurological diseases using human induced pluripotent stem cells and brain organoids. He has received several awards, including

the prestigious NIH Director's New Innovator Award, NARSAD, Emerald Foundation Young Investigator Award, Surugadai Award, Rock Star of Innovation, NIH EUREKA Award, two Telly Awards for Excellence in Science Communication among several others.

Title: *Rescuing molecular, cellular and network alterations in Pitt-Hopkins brain organoids*

Abstract: Transcription Factor 4 (TCF4) has been associated with autism, schizophrenia, and other neuropsychiatric disorders. However, how pathological TCF4 mutations affect the human neural tissue is poorly understood. Here, we derive neural progenitor cells, neurons, and brain organoids from skin fibroblasts obtained from children with Pitt-Hopkins Syndrome carrying clinically relevant mutations in TCF4. We show that neural progenitors bearing these mutations have reduced proliferation and impaired capacity to differentiate into neurons. We identify a mechanism through which TCF4 loss-of-function leads to decreased Wnt signaling and then to diminished expression of SOX genes, culminating in reduced progenitor proliferation in vitro. Moreover, we show reduced cortical neuron content and impaired electrical activity in the patient-derived organoids, phenotypes that were rescued after correction of TCF4 expression or by pharmacological modulation of Wnt signaling. This work delineates pathological mechanisms in neural cells harboring TCF4 mutations and provides a potential target for therapeutic strategies for genetic disorders associated with this gene.



Kathrin Meyer, Ph.D.

Center for Gene Therapy, The Research Institute at Nationwide Children's Hospital
E-mail: Kathrin.Meyer@nationwidechildrens.org

Kathrin C. Meyer, PhD, studied Cellular and Molecular Biology at the Institute of Cell Biology in Berne, Switzerland. Her post-doctoral research was performed in Brian Kaspar's laboratory at the Center for Gene Therapy in Columbus Ohio. During that time, Dr. Meyer established a new and fast reprogramming method for in vitro modeling of neurodegenerative diseases using patient skin cells. Moreover, Dr. Meyer developed intrathecal gene therapy programs for several neurodegenerative diseases including Spinal Muscular

Atrophy and Batten Disease. Multiple clinical trials that are based on this work are currently ongoing at Nationwide Children's Hospital. In 2017, Dr. Meyer became a Principal Investigator at Nationwide Children's Hospital. She is also an Assistant Professor in the Department of Pediatrics at The Ohio State University, Columbus, Ohio.

Title: *In vitro characterization and drug candidate testing on Pitt Hopkins Patient astrocytes and neurons*



Michelina Iacovino, Ph.D.

Associate Professor of Pediatrics
The Lundquist Institute Harbor-UCLA Medical Center
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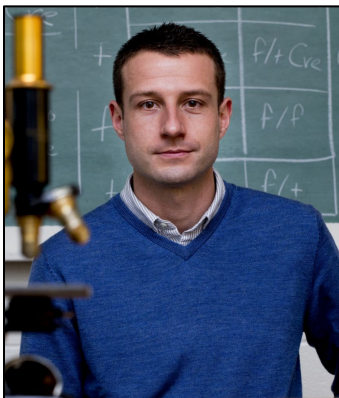
Dr. Iacovino is an Associate professor at the Lundquist Institute at Harbor-UCLA medical center. She received her Ph.D. in Biochemistry from the University of Molise, Italy. During her postdoctoral fellowship, she trained under the mentorship of Dr. Michael Kyba, studying the role of Hox genes in blood development. In 2012 with her expertise in stem cell biology, she was recruited by the Department of Pediatrics at the Harbor-UCLA Medical Center and the Lundquist Institute to join a team of

investigators working on rare disorders. She is currently working on the therapeutic development for Sanfilippo disorders and Pitt Hopkins syndromes.

Title: *ssRNA therapy for Pitt Hopkins syndrome*

Abstract: Pitt Hopkins is a rare genetic disorder that causes intellectual disability, speech and motor impairment, and breathing problems. The disorder is caused by hemizygote loss of TCF4 (haploinsufficiency). TCF4 is a transcription factor that controls other genes essential for brain development and cognition.

An ideal treatment for haploinsufficiency would double the functional gene expression to compensate for the missing copy. Controlling levels may be critical when haploinsufficiency involves a transcription factor like TCF4, whose levels may need to be tightly regulated. WE previously showed that small double-stranded RNA (ds-RNA) molecules can induce TCF4 expression through a naturally occurring mechanism as a way to fine-tune gene activity -- known as RNA interference. Here we have extended our screening to single-stranded RNA (ss-RNA) oligonucleotides capable of inducing TCF4 expression. ssRNA are easier to deliver into the cells and therefore have a simpler path into the clinic. Our screening shows that ssRNA molecules are capable to induce TCF4 expression both at the RNA and protein level. We are currently preparing experiments to test their efficacy in vitro.



Kennedy Lab

Andrew John Kennedy, Ph.D.

Associate Professor

Department of Chemistry and Biochemistry

Bates College

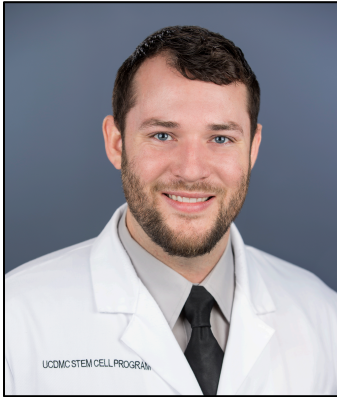
E-mail: akennedy@bates.edu

My research interest is to understand the chemical mechanisms that encode and maintain long-term memory. Currently, we investigate how learning imprints information onto the epigenome of participating neurons, and how these experientially-driven changes to the structure of the genome affect the expression of genes that facilitate plasticity. Using gene knockout models, antisense oligonucleotides, and novel small molecules, we probe the function of epigenetic modifying enzymes in and specific epigenetic markers of memory function. A major goal of our laboratory is to develop therapies that manipulate the epigenome to enhance cognition in cases of intellectual disability, such as Pitt-Hopkins Syndrome.

Title: *TET Enzyme Inhibition to Enhance Cognition*

Abstract: Ten-eleven translocation (TET) enzymes mediate the de-methylation of DNA, an epigenetic biochemical mechanism that regulates long-term gene expression patterns, cell identity, and cellular plasticity. TET enzymes catalyze the benzylic oxidation of 5-methylcytosine to 5-hydroxymethylcytosine, the rate-limiting enzymatic step in a process that yields an unsubstituted cytosine nucleotide in DNA. Increased expression of the TET enzymes and subsequent reductions in neural DNA methylation has been associated with Tcf4 haploinsufficiency, the genetic cause of Pitt-Hopkins Syndrome (PTHS). Here, we present data that TET enzyme inhibitors decrease the production of 5-hydroxymethylcytosine in mouse hippocampal neurons, causing the buildup of methylated DNA. In vivo administration of a TET inhibitor was also shown to rescue spatial learning and memory in a mouse model of PTHS, suggesting the TET enzymes and DNA methylation could be epigenetic therapeutic targets for PTHS.

Kyle Fink, Ph.D.



Assistant Professor

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University of California, Davis Medical Center
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Dr. Fink received a Ph.D. in Neuroscience from Central Michigan University in 2013 and a Ph.D. in Neuroimmunology from the University of Nantes in 2013. Dr. Fink then did his postdoctoral training at the UC Davis Institute for Regenerative Cures and Stem Cell Program. Dr. Fink joined the faculty at UC Davis in the Neurology Department in 2017. The Fink laboratory focuses on the therapeutic development of gene modifying modalities such as Zinc Fingers, Transcription Activator-like Effectors, and CRISPR/Cas9 to

treat genetically-linked neurological disorders.

Title: *Preclinical assessment of CRISPR-mediated gene regulation to rescue haploinsufficiency in Pitt Hopkins syndrome*

Abstract: CRISPR-mediated gene activation (CRISPRa) utilizes a single guide RNA (sgRNA) to recruit a nuclease-deficient Cas9 (dCas9) fused to transcriptional activator domains to promote targeted gene expression. It was recently demonstrated that CRISPR-mediated activation (CRISPRa), used to directly target the endogenous gene regulatory elements (promoter or enhancer) that regulate specific genes successfully rescued a haploinsufficient organoid models of PTHS. This strategy provides a potential therapeutic strategy for TCF4 haploinsufficiency. Importantly, a single CRISPRa could potentially be used for all PTHS patients regardless of their specific genetic abnormality leading to the haploinsufficiency. This work focuses on the development of AAV-ready CRISPRa constructs to accelerate the translational potential of a CRISPRa approach for Pitt Hopkins.



Colleen Niswender, Ph.D.

Professor of Pharmacology

Department of Pharmacology and Warren Center for Neuroscience
Drug Discovery
Vanderbilt University School of Medicine
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Colleen Niswender is an Associate Professor of Pharmacology and the Warren Director of Molecular Pharmacology for the Warren Center for Neuroscience Drug Discovery at Vanderbilt University. In her role as Director of Molecular Pharmacology for the WCNDD, she has been involved in multiple small molecule discovery projects to search for and characterize novel allosteric modulators of various metabotropic glutamate and muscarinic receptors. In July of 2017,

the WCNDD filed and was granted open IND status for clinical studies with an M1 positive allosteric modulator (PAM) for cognition; this program has now been licensed to Acadia Pharmaceuticals. Additionally, the WCNDD team advanced another compound, a PAM of metabotropic glutamate receptor 4 for Parkinson's disease, into the clinic via a WCNDD-partnered startup company based in Nashville, TN, Appello Pharmaceuticals. The Center, which operates as a small biotech within Vanderbilt, now has a pipeline of products as well as multiple collaborations with industrial partners. In recent years, Dr. Niswender has established a research program around the therapeutic potential of WCNDD targets in Rett syndrome and other neurodevelopmental disorders such as Pitt Hopkins syndrome and Neurofibromatosis Type I.

Title: *Rescue of behavioral phenotypes in Tcf4+/- mice by overexpression of MeCP2*

Abstract: Studies examining neurodevelopmental disorders (NDDs) have emphasized a relationship between Pitt-Hopkins syndrome (PTHS), which is caused by mutations in the Transcription Factor 4 (TCF4) gene, and myelination defects that suggest novel molecular phenotypes in NDDs (Phan et al., 2020). Known phenotypic overlap between PTHS and another NDD, Rett syndrome (RTT) suggests that there are possible common signaling pathways between the two NDDs. We have found that increasing expression of Methyl-CpG-Binding Protein 2 (MeCP2), the causative protein in most cases of RTT, decreases hyperactivity and corrects impairments in learning and memory in a mouse model of PTHS (Tcf4+/-). The current study sought to understand if the positive effects of MeCP2 overexpression on behavioral phenotypes in the Tcf4+/- model were due to a reversal of myelination deficits. We generated four distinct animal genotypes (WT, MECP2Tg1/o, Tcf4+/-, MECP2Tg1/o; Tcf4+/-) and performed RNA-sequencing from the hippocampus and striatum followed by qRT-PCR, which identified myelin-associated genes as differentially expressed in MECP2Tg1/o; Tcf4+/- mice in both brain regions. However, further examination showed that the direction of gene expression changes, when compared to wild-type mice, was the same for both the Tcf4+/- and MECP2Tg1/o; Tcf4+/- genotypes, and Western blotting revealed decreased expression for the myelinating oligodendrocyte (OL) protein Myelin Oligodendrocyte Glycoprotein (MOG) in mice of both the Tcf4+/- and MECP2Tg1/o; Tcf4+/- . We are currently evaluating potential changes in OL morphology and number (mature and immature cells), myelin synthesis, and neuronal spine density via immunohistochemistry of brain sections. Altogether, our results to date indicate that behavioral phenotypic rescue in MECP2Tg1/o; Tcf4+/- mice may not result from a correction of myelination deficits characteristic of Tcf4+/- animals.



James B. Adams, Ph.D.

President's Professor

Director of Autism Research Program

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James B. Adams, Ph.D., is the Director of the Autism/Asperger's Research Program at Arizona State University. His research focuses on the medical causes of autism and how to treat and prevent it including the areas of nutrition (vitamins/minerals, essential fatty acids, carnitine, digestive enzymes, special diets), oxidative stress, gut problems, gut bacteria, toxic metals, and seizures. He has published over 180 peer-reviewed scientific articles, including over 50 related to autism. He is also the Past

President of the Autism Society of Greater Phoenix, the President of the Autism Nutrition Research Center, President of Autism Diagnostics, and chair of the Scientific Advisory Board of the Neurological Health Foundation. He has an adult daughter with autism.



Rosa Krajmalnik-Brown, Ph.D.

Director, Biodesign Institute
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Dr. Rosa Krajmalnik-Brown is a Professor at the School of Sustainable Engineering and The Built Environment, the Biodesign Swette Center for Environmental Biotechnology, and the Biodesign Center for Applied Microbiomics at Arizona State University. She has a Ph.D. in Environmental Engineering from Georgia Tech. She was awarded an NSF CAREER award, was selected Fulton Engineering Exemplar Faculty, and as Distinguished Alumni from UAM-Mexico (Where she received her college degree). She has funding for her research from many federal agencies including NIH, DoE, DoD, and NSF. She is a pioneer in research on gut microbiome and autism. She is author of 4 patents and more than 100 peer-reviewed publications. She specializes on molecular microbial ecology for bioremediation, the use of microbial systems for bioenergy production, and the human intestinal microbial ecology and its relationship to obesity, metabolism, and autism.

Title: *Microbiota Transfer Therapy for Children with Pitt Hopkins Syndrome*

Abstract: Six children with Pitt Hopkins Syndrome and chronic constipation were enrolled in a Phase 2 randomized double-blind placebo-controlled clinical trial of microbiota transfer therapy (MTT). MTT involved treatment with vancomycin/placebo for 10 days, followed by a half-day of fasting and a bowel cleanse, followed by 4 days of high-dose microbiota/placebo capsules and 12 weeks of maintenance dose/placebo. In Part 1, half the group received MTT and half received only the bowel cleanse. In Part 2, the placebo group was switched to MTT, and the original treatment group was observed. In Part 3, the participants were observed for 3 months post-treatment. There were substantial improvements in gastrointestinal symptoms and pain, and some improvement in Pitt Hopkins-related symptoms. Treatment benefits generally continued after treatment stopped.



Jessica Duis, MD

Associate Professor of Pediatrics & Medical Genetics
Director of Pitt-Hopkins Center of Excellence at 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 and personalized therapeutic interventions for individuals with neurodevelopmental disorders to improve quality of life on a day-to-day basis. She has

designed and worked on many clinical trials and currently focuses on management guidelines and establishing outcome measures that quantitatively capture clinically significant features of neurodevelopmental disorders in the natural environment. She lives in Colorado with her husband and three kids, and enjoys spending time with her family outdoors.

Title: *Treatment Options from The Colorado Clinic*

Abstract: We at the PHRF hold Dr. Duis in high regard. She is a physician who takes her time and thinks out of the box when it comes to treating our kids. The Pitt Hopkins Center for Excellence at Colorado Children's Hospital has been open for more than a year now and Dr. Duis has seen about 10-12 patients. In this talk, she will share some of the beneficial meds she is using to treat behaviors, sleep and gut issues, in the hopes that you can share these ideas with your own doctors back home.

***she cannot prescribe and is not giving medical advice. These are merely suggestions to be discussed with your own doctors back home.*

Friday Talk from Dr. Duis: Dr. Jessica Duis from Children's Hospital Colorado will discuss the experience of the multidisciplinary team and clinical spectrum seen at their Center of Excellence including expansion of the phenotype of Pitt Hopkins Syndrome.



Patricia Cogram, Ph.D.

Universidad de Chile, Chile
IEB, Faculty of Science
Genetics and Evolutionary Genetics Department
Associate Professor
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Patricia Cogram, Ph.D., is an Associate Professor of Genetics at the IEB Genetics Department, University of Chile. She graduated at University College London, BSc in Evolutionary genetics and MSc in Neuroscience at Oxford University, UK. Dr Cogram received a Ph.D. in Neurodevelopment at University College London, UK and conducted postdoctoral studies at the Medical Research Council, UK and UCL. Patricia is a leading researcher in the field of neurodevelopmental rare diseases. Dr. Cogram has over 20 years of experience in drug discovery. As the Director of the Biomedicine Division at Fraunhofer-Gesellschaft Research Laboratories, she led several programs on neuroinflammation and rare diseases to the pipeline of different Pharma Corporations. She is also the CEO of GeN Ltd a biotechnology company dedicated to disease target identification and drug development in PTHS, FXS and other forms of Autism.

Title (Thursday): *Rescuing TCF4 behavioral and neuronal spine phenotypes in mice*

Abstract: Tcf4 haploinsufficient mice have deficits in spatial memory, anxiety and general activity. We treated new-born Tcf4^{+/-} mice (Postnatal day 1 (P1)) and prevented the emergence of disease signs including anxiety-like behavior, memory problems, and abnormal neuronal spines in affected mice brains suggesting that restoring normal levels of the Tcf4 gene is a potential therapy for Pitt-Hopkins syndrome, which otherwise has no specific treatment. We also observed that restoring activity of the gene from embryonic life fully prevented PTHS signs.

Title (Friday): *Cell Culture Bank: Pitt Hopkins Chile; Taking the findings into mouse models*

Abstract: Patricia Cogram¹, Marcelo Ezquer², Elisabeth Behrens^{2,3}
¹IEB, Faculty of Science, University of Chile, Chile
² Clinica Alemana, Chile
³ JJ Aguirre Hospital, Chile

In collaboration with Professor Alysson Muotri, the families in Chile and the PTHS Foundation support we are growing and expanding the Pitt Hopkins Cell Bank using skin fibroblasts from Pitt Hopkins and unaffected relatives so that researchers all over the world will have easier access to skin with the aim to test novel therapeutics for Pitt Hopkins Syndrome, and understand the underlying cause of Pitt Hopkins and have the greatest likelihood of having a profound impact on symptoms.



Allyson Berent, DVM, Dipl. ACVIM

CSO, Foundation for Angelman Syndrome Therapeutics

COO, GeneTx Biotherapeutics

Director Interventional Endoscopy Services, The Animal Medical Center

Mother to Quincy: A beautiful little girl living with Angelman syndrome

Dr. Allyson Berent is a veterinary internal medicine specialist who serves as the Director of Interventional Endoscopy Services at the largest animal hospital in the world, The Animal Medical Center, in New York City. After graduating from Cornell University College of Veterinary Medicine she completed an internship at the University of Minnesota and a residency in Small Animal Internal Medicine at the Veterinary Hospital of the University of Pennsylvania. After completing a fellowship in interventional radiology at the Veterinary Hospital of the University of Pennsylvania, a fellowship in Endourology at Thomas Jefferson University, and an Interventional radiology fellowship at the Hospital of the University of Pennsylvania, she served as an Adjunct Assistant Professor in Internal Medicine and Interventional Radiology/ Interventional Endoscopy at the Matthew J. Ryan Veterinary Hospital of the University of Pennsylvania. Dr. Berent has a particular focus on medical device development, stem cell therapy through regenerative medicine and selective arterial delivery, ureteral diseases, urinary incontinence and minimally invasive management of upper tract urinary obstructions and biliary obstructions. In 2014 Dr. Berent's daughter was diagnosed with a rare non-degenerative neurogenetic disorder called Angelman syndrome. In October of 2015 she joined to Board of Directors as a Scientific Director for the Foundation for Angelman Syndrome Therapeutics (FAST), and in March of 2016 became the Chief Science Officer for the Foundation. Dr. Berent helped to spearhead the development of a pre-competitive biomarker and outcome measure consortium in order to bring patient focused outcome measures forward for human clinical trials (Angelman Syndrome Biomarker and Outcome Measure Consortium-ABOM) and now serves as the Director of this consortium. Dr. Berent Co-Founded the International Angelman Syndrome Research Council (INSYNC-AS). Through FAST, Dr. Berent collaborated with a consortium of scientists to encourage translational research opportunities, in order to help bring novel genetic therapies forward toward human clinical trials. Through this work, with the foundation, Dr. Berent co-founded GeneTx Biotherapeutics, a company singularly focused to advance an antisense oligonucleotide (ASO) therapy through IND enabling studies and a phase 1/2 clinical trial. Dr. Berent currently serves as the Chief Operating Officer of GeneTx Biotherapeutics, who partnered with Ultragenyx Pharmaceuticals in August of 2019. The Phase 1/2 clinical trial started enrolling patients in February 2020 as the first intrathecally delivered ASO for Angelman syndrome, a study of safety and tolerability of GTX-102.

Title: *A foundation's journey through drug development: The need to move FAST*



Yael Weiss, Ph.D.

Chief Executive Officer
Mahzi Pharmaceuticals
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Yael Weiss completed her MD PhD at Hadassah Medical School at the Hebrew University in Jerusalem. She has over 20 years of industry experience in medical/clinical and business development roles at Genzyme, Merck and Ultragenyx. Yael founded and is the CEO of Mahzi Therapeutics in 2020 to bring therapies to patients with underdiagnosed ultra rare genetic neurodevelopmental disorders. Mahzi works closely with patient foundations to support their journey towards drug development, and bring programs into Mahzi once pre-clinical proof of concept is established. Throughout

her career Yael has mentored and guided bio entrepreneurs in their journey taking scientific inventions from bench to bedside.

Title: *Next Steps, From mouse model to human*

Abstract: What does it take to take a treatment from the lab to the clinic? Yael Weiss of Mahzi Therapeutics will share her company's plan to help us get there.



Nancy Jones, Ph.D.

Vice President, Clinical Development
Neuren Pharmaceuticals
E-mail: NJones@neurenpharma.com

Dr. Jones joined Neuren in January 2013. She leads the design and implementation of Neuren's clinical studies in neurodevelopmental disorders. Prior to joining Neuren, she held a senior position at Autism Speaks, the largest science and advocacy organization in the US focused on autism spectrum and related disorders. She was at Autism Speaks for 6 years, directing the overall operations of the Autism Treatment Network, a network of hospitals and medical centers dedicated to improving access to comprehensive, coordinated medical care for individuals with ASD.

She also oversaw the Autism Clinical Trials Network, a network developed to promote and expedite clinical trials in ASD, and played a lead role in an initiative to enhance the development of syndrome-specific outcome measures for treatment trials in ASD. Dr. Jones received her Ph.D. in Applied Linguistics from the University of California, Los Angeles where she focused on the neurobiology of language and neurodevelopmental disorders.

Title: *NNZ-2591 as a Treatment for Pitt Hopkins Syndrome*

Abstract: NNZ-2591 is a novel drug being developed by Neuren Pharmaceuticals for the treatment of Pitt Hopkins syndrome. This presentation will provide an overview of NNZ-2591, how we believe it works, and why it might be a potentially useful treatment for PTHS. We will also give an update on the Phase 2 clinical trial.



Ingrid Harding

PTHS Mom and Co-founder of Rett syndrome Research Trust and Girl Power 2 Cure

Ingrid Harding is mom to 3 including full-time caregiver to Sarah (PTHS / formerly RTT), age 21. She is the co-founder of the Rett Syndrome Research Trust and currently serves on its Board of Trustees. She also founded Girl Power 2 Cure and served as President until 2017 when Sarah was newly diagnosed with Pitt Hopkins syndrome.

Title: *From Overwhelmed to Empowered: A Journey Towards "Peace of Mind" and a 3-Month Solo Vacation*

Abstract: No matter the age or health of your child, you probably spend time worrying about what would happen if anything were to happen to you. Let's spend 30 minutes getting you ready to start on your own P.O.M. (Peace of Mind) Journey. I promise we'll laugh a lot and you'll leave with a starter list that will lead you to a new way of thinking about all the things you do (or wish you could do!) for your child.



Kelly Beins, OTR/L

Strategic Clinical Consultant
Unyte-iLs (www.integratedlistening.com)

Kelly Beins is a seasoned Occupational Therapist with over 27 years of experience, including 17 years with certification in sensory integration, and experience in pediatrics and rare genetic disorders. Kelly believes in evidence-informed interventions and is passionate about using an integrative model of practice. Kelly has recently joined Unyte-iLs as a strategic clinical consultant. She is also growing a global mentorship program to make best-practices more accessible for other sensory OT's in private practice. Kelly specializes in Polyvagal Theory, she is an international speaker, has published two children's books and was named, Autism

Parenting Magazine's "Top OT" for two consecutive years.

Kelly is a sensory parent herself, which has led to many insightful and unexpected learnings along her professional and personal path.

Title: *Making Sense of Sensory Processing - It's all connected!*

Abstract: Sensation is the foundation of our central nervous system, and everything we do! Understanding basic concepts of sensory processing and the role that sensory plays in childhood behavior and neurodevelopment is essential to supporting children and to making life EASIER for parents. Kelly will share her "Sensory Parenting an EASIER Way" framework and participants will learn key concepts about sensory processing, child development, polyvagal theory and practical strategies for supporting improvement in daily activities of children with Pitt Hopkins.



Kelly Berg

S2C Practitioner

Growing Kids Therapy Center (www.growingkidstherapy.com)

Kelly began working with individuals with complex communication and sensory-motor differences in 2019 as a Spelling to Communicate Practitioner at Growing Kids Therapy Center. She was guided to the field having a degree in Communication Sciences and Disorders from the University of Texas, as well as through her relationship with the local spelling community in the town of Herndon. As an S2C Practitioner, she works one on one with individual clients as well as in groups, and also teaches the ACTS Parent Cohort for families of individuals who spell to communicate. In addition to her work at Growing Kids, Kelly is also a member of the Leadership Cadre at the International Association for Spelling as Communication, whose mission is to advance communication access for nonspeaking individuals globally through training, education, advocacy, and research. Kelly has served as a mentor, and is currently a Cohort Leader for the S2C Professional training program.

Title: *Fundamentals of Spelling to Communicate and Presuming Competence*

Abstract: This presentation by S2C Practitioner and I-ASC Leadership Cadre member Kelly Berg will cover the basics of Spelling to Communicate (S2C) and Presuming Competence. Topics will include S2C as a means of AAC (Augmentative and Alternative Communication), the neurology of language and speech, why a lack of speech is not indicative of a lack of intelligence, and why we should be presuming competence in all nonspeakers.



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