&

YELLOW BRICK ROAD

PROJECT

2024 HNRNP Conference & Family Meeting

July 27th thru July 30th, 2024

FAMILIES, CLINICIANS, & RESEARCHERS

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change lives

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Greater Baltimore Medical Center, Harvey Institute for Human Genetics

COMBINEDBrain

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Once Upon A Gene

Once Upon A Gene

Saturday, July 27th		
7:00-9:00 PM	**WELCOME SOCIAL** YBRP and HNRNPFF Directors welcome families and other attendees to kick off this year's joint conference! Let's get to know each other and enjoy some light snacks!	*LEGACY BALL-ROOM*
Sunday, July 28th		
8:00-5:00	***NHS/CLINICAL EVALUATIONS-SEE INDIVIDUAL EVALUATION SCHEDULES***	***Eval Rooms***
8:00-9:30	***YBRP/HNRNPFF provided BREAKFAST***	***Legacy Ballroom***
9:45-10:10	**Welcome:** YBRP and HNRNP Family Foundation Representatives kick off the 2024 joint conference.	*LEGACY BALL-ROOM*
10:15-10:45	**Patient Registry:** Simons Searchlight shares the importance and benefits of participation in the patient registry.	*LEGACY BALL-ROOM*
10:45-10:55	*BREAK*	*LEGACY*
11:00-11:30	**Genetics 101:** Dr. Maddie Gillentine explains the basic genetics behind *HNRNP* variants.	*LEGACY BALL-ROOM*
11:35-12:00	**The Clinical Spectra of HNRNP-RNDDs:** Dr. Tonie Kline presents on the features seen among and within HNRNPs.	*LEGACY BALL-ROOM*
12:00-1:25	***YBRP/HNRNPFF provided LUNCH***	*Legacy Ballroom*
1:30-1:55	**Neurodevelopmental PAG Consortium:** COMBINEDBrain's Dr. Terry Jo Bichell discusses how they are partnering with YBRP & other rare disease organizations.	*LEGACY BALL-ROOM*
2:00-2:25	**Pediatric Translational Neuroscience Initiative:** Rachael Mercer presents on the structure and mission of St. Jude Children's Research Hospital's PTNI program which is engaged in rare pediatric neurological disorders research.	*LEGACY BALL-ROOM*
2:25-2:35	*BREAK*	*LEGACY*
2:40-3:10	**The importance of evaluations and bio samples:** HNRNPFF representatives share what happens with data and biosamples.	*LEGACY BALL-ROOM*
3:15-3:30	**Closing Remarks:** YBRP and HNRNPFF representatives close out day one of the 2024 conference.	*LEGACY BALL-ROOM*
4:00-5:30	**Lightning Talks:** *(Researchers Only "Think Tank" Session)*	*Ridlon*
7:00-9:00	*Researcher Science Mixer – Private Event for Research Team and Guest Speakers*	*Formula Brewing*

Monday, July 29th				
8:00-5:00	***NHS/CLINICAL EVALUATIONS-SEE INDIVIDUAL EVALUATION SCHEDULES***			***Eval Rooms***
8:00-9:30	***YBRP/HNRNPFF provided BREAKFAST***			***Legacy Ballroom***
HNRNPH2* Track** ***LEGACY: Catterall	***HNRNPU* Track** ***LEGACY: Barlow***	***HNRNPK* Track** ***LEGACY: Bergsma***	**Growing Cohort *HNRNP*s Track** ***Denton***	
YBRP: YBRP explains our mission & what's been going on behind the scenes to drive the mission forward. ***9:45-10:15***	**HNRNPU NHS:** Dr. Meena Balasubramanian presents on HNRNPU Natural History data. ***9:45-10:15***	**HNRNPK NHS:** Dr. Billie Au and Dr. Tonie Kline present on HNRNPK Natural History data. ***9:45-10:15***	**HNRNP-RNDD NHS:** Dr. Maddie Gillentine presents on HNRNP-RNDD Natural History data. ***9:45-10:15***	
HNRNPH2 & Seizures: YBRP Vice President Haim Farkash & Dr. Jennifer Bain discuss recent and other seizure- related deaths among H2 patients and the impact on the community. ***10:20-10:45***	**HNRNPU NHS Extended** ***10:20-10:45***	**HNRNPK Research:** Dr. Sean Post discusses his research with an HNRNPK mouse model. ***10:20-10:45***	**How to Grow Ultra Rare Patient Population** ***10:20-10:45***	
HNRNPH2 GROUP PHOTO	*HNRNPU* GROUP PHOTO	*HNRNPK* GROUP PHOTO	*HNRNP* GROUP PHOTO	
10:55-11:05	*BREAK*			*LEGACY*
HNRNPH2* Track** ***LEGACY: Catterall		**All *HNRNP*s Track** ***LEGACY: Barlow/Bergsma***		
HNRNPH2 NHS & Clinical Trial: Dr. Jennifer Bain presents the most recent data from the HNRNPH2 Natural History Study ***11:05-11:35***		**Expert Panel:** Getting the most out of SLP, OT and ABA. ***11:05-11:35***		
HNRNPH2 ASO CLINICAL TRIAL: Dr. Laurence Mignon of n-Lorem presents on their ASO drug development and the upcoming N of 1 clinical trial, including remarks from the Glenn Family on their child as the pilot patient. ***11:40-12:10***		**HNRNP-RNDD Disease Models:** A session discussing current disease models in HNRNP-RNDDs (fish, iPSC, mouse, organoids, etc.). ***11:40-12:10***		
12:15-1:30	***YBRP/HNRNPFF provided LUNCH***			***Legacy Ballroom***
1:30-1:45	*GROUP PHOTO – ALL ATTENDEES*			*LEGACY*
HNRNPH2 RESEARCH: Dr. Ané Korff and Rachael Mercer present on the most recent pre-clinical research at St. Jude Children's Research Hospital as part of the Pediatric Translational Neuroscience Initiative (PTNI). ***1:50-2:25***		**Therapeutics Panel:** (ASOs, drug repurposing, gene therapy) with Dr. Jennifer Bain, Dr. Meena Balasubramanian, and Dr. Chris Ricupero. ***1:50-2:25***		
HNRNPH2 RESEARCH: Prof. Rotem Karni /Dr. Manar Abu Diab present on the research through Andlit into the downstream effects of HNRNPH2 mutations and other treatment pathways for HNRNPH2-RNDD. ***2:30-3:00***		**"My doctor has never heard of the HNRNP-RNDDs" Panel:** Clinicians and researchers discuss what parents and doctors providing care for a patient should know about HNRNP-RNDDs such as therapies to consider and other comorbidities. ***2:30-3:00***		
3:00-3:15	*BREAK*			*LEGACY*
Round Table 1: Sleep Disturbance ***LEGACY: Catterall 3:15-3:45***	**Round Table 2: Orthopedics** ***LEGACY: Catterall 3:15-3:45***	**Round Table 3: CVI** ***LEGACY: Barlow 3:15-3:45***	**Round Table 4: Seizures** ***LEGACY: Bergsma 3:15-3:45***	
Round Table 5: Planning for the Future ***LEGACY: Catterall 3:15-3:45***		**Round Table 6: GI Issues** ***LEGACY: Barlow 3:15-3:45***	**Round Table 7: Communication** ***LEGACY: Bergsma 3:15-3:45***	
Round Table 8: Sleep Disturbance ***LEGACY: Catterall 3:45-4:15***	**Round Table 9: Orthopedics** ***LEGACY: Catterall 3:45-4:15***	**Round Table 10: CVI** ***LEGACY: Barlow 3:45-4:15***	**Round Table 11: Seizures** ***LEGACY: Bergsma 3:45-4:15***	
Round Table 12: Planning for the Future ***LEGACY: Catterall 3:45-4:15***		**Round Table 13: GI Issues** ***LEGACY: Barlow 3:45-4:15***	**Round Table 14: Communication** ***LEGACY: Bergsma 3:45-4:15***	
HNRNPH2 SOCIAL ***LEGACY: Catterall 7:00-9:00***	*HNRNPU* SOCIAL ***LEGACY: Barlow 7:00-9:00***	*HNRNPK* SOCIAL ***LEGACY: Bergsma 7:00-9:00***	Growing Cohort *HNRNP*s SOCIAL ***Denton 7:00-9:00***	

Tuesday, July 30th		
8:00-5:00	***NHS/CLINICAL EVALUATIONS-SEE INDIVIDUAL EVALUATION SCHEDULES***	***Eval Rooms***
8:00-9:30	***YBRP/HNRNPFF provided BREAKFAST***	***Legacy Ballroom***
HNRNPH2* Track** ***LEGACY: Catterall	**All *HNRNPs* Track** ***LEGACY: Barlow/Bergsma***	
YBRP: YBRP discusses our strategic plan and what's next to move our mission forward as we embark on the pilot clinical trial and move closer to broader clinical trials. ***9:45-10:15***	**HNRNPFF:** HNRNPFF Representatives explain their mission and how to move the mission forward. ***9:45-10:15***	
HNRNPH2 RESEARCH: Dr. Christopher Ricupero presents on the HNRNPH2 patient biorepository and biomarkers, as well as research updates into potential treatment pathways for HNRNPH2-RNDD. ***10:20-10:45***	**Expanding our NHS:** HNRNPFF Representatives will discuss how we plan to update our Natural History Study with launch of the Genial platform. ***10:20-10:45***	
10:45-10:55	*BREAK*	*LEGACY*
YBRP HNRNPH2 TOWN HALL: YBRP Directors & HNRNPH2 Researchers hold a Q&A session with a panel of our guest speakers and the leading researchers in HNRNPH2 Neurodevelopmental Disorder answering questions direct from families. ***11:00-12:00***	**HNRNPFF TOWN HALL:** HNRNPFF Directors & Researchers hold a Q&A session with a panel of leading researchers in HNRNP-RNDDs answering questions. ***11:00-12:00***	
12:00-1:25	***YBRP/HNRNPFF provided LUNCH***	***Legacy Ballroom***
1:30-1:55	**RESEARCH:** Methylation studies w/ Dr. Rosanna Weksberg.	*LEGACY BALLROOM*
1:30-1:55	**Rare Life:** Once Upon a Gene - Effie Parks.	*LEGACY BALLROOM*
2:00-2:25	**What's Next:** Clinicians/Researchers - What's Next in HNRNP-RNDD Research?	*LEGACY BALLROOM*
2:30-2:45	**Closing Remarks:** YBRP and HNRNPFF Representatives close out the 2024 conference.	*LEGACY BALLROOM*
3:30-4:30	**YBRP HNRNPH2 AWARENESS WALK**	*Courtyard*

YBRP Board of Directors

Trish Flanagan, President & Board Director is a co-founding member of The Yellow Brick Road Project and mom to Morgan, touched by an HNRNPH2 mutation. A seasoned Early Childhood Educator, she earned her Bachelors Degree of Science from Penn State University majoring in Human Development and Family Studies with an Early Childhood focus. Her first role working with children was within the Child Life Program at Westchester Medical Center in NY providing play experiences to children as well as offering psychosocial support to families. She went on to earn her Masters Degree in Early Childhood Education from the College of New Rochelle and resides and teaches in the suburbs of New York.

Haim Farkash, Vice President & Board Director is a co-founding member of the Project and champions the memory of his daughter Yaeli, who recently passed from Sudden Unexpected Death in Epilepsy (SUDEP). He is the Chief Operating Officer in the MT Trading Group, a global consortium specializing in international procurement, marketing, and distribution of merchandise in developing countries in Africa and Asia. Haim has extensive experience in the structuring of cross-border transactions and supply chain of goods with a focus on maritime law and international direct and indirect tax considerations. Prior to joining MT Trading Group, Haim was an associate in the commercial litigation group of one of the leading law firms in Israel. Haim was admitted to the Israeli Bar and a graduate of Bar-Ilan University, earning his LLB with concentrations in Law and Economics.

Paula Tyson, Secretary, Treasurer & Board Director is a co-founding member of the Project and mother to Lillie, impacted by an HNRNPH2 genetic variation. She is currently a Compensation Consultant and Analyst at ChaseCompGroup, and before joining the firm in 2011, worked independently for consulting and other service firms, in client relations, account management, and accounting functions. Prior to her independent work, she worked for a private company for six years in personnel management and client relations and began her career as an analyst and consultant for Watson Wyatt (now Towers Watson) in the areas of human resources and organization effectiveness. She obtained her Bachelor's degree in Romance Languages from the University of Georgia, graduating Magna Cum Laude.

Robert Glenn, Director of Technology & Board Director "Bobby" is father to Rose and member of the board of directors. He studied Molecular and Cellular Biology at the University of Colorado, Boulder, but ultimately found his passion working in various roles in business development and marketing in the tech industry. As the Director of Marketing and Communications, Robert is committed to leveraging his experience for raising awareness for HNRNPH2, along with other orphan diseases. His goal is to raise funds in order to further develop research into this new genetic mutation.

Stacy Paddon, International Outreach Coordinator & Board Director Stacy Paddon is mom to daughter Cady and a member of the board of directors. She currently works for the Business Intelligence team at Christie's auction house in London, having previously spent time in the company's Human Resources office where she managed European recruitment. She has previously held management positions at the National Trust, the Vancouver 2010 Olympic and Paralympic Winter Games Organizing Committee and ING Insurance. She holds a Bachelor of Journalism degree from Carleton University with Combined Honors in Political Science and is a Chartered Insurance Professional.

YBRP Committees & Members

International Delegate Committee

TRISH FLANAGAN – International Delegate, Eastern US (NY)
PAULA TYSON – International Delegate, Eastern USA (GA)
NICOLE GLENN – International Delegate, Western US(CA)
TANYA DeKONINCK – International Delegate, Canada
STACY PADDON – International Delegate, UK & Europe (UK)
ANDREIA RAMOS – International Delegate, Europe (Portugal)
BEATRIZ CORTES – International Delegate, Europe (Spain)
ANDREIA ZANELATO—International Delegate, Europe (France)
ELISA REVIGLIO – International Delegate, Europe (Italy)
LIV-STEPHANIE BANTLE – International Delegate, Baltics (Norway)
VERA MAKAROVA – International Delegate, Russia/Ukraine (Russia)
ESTHER OLLECH – International Delegate, Israel
ALANA FAHEY – International Delegate, Australia
KAORI SUZUKI – International Delegate, Japan

Outreach & Action Committee

ANGELA LINDIG – Western & Midwestern US: AK, CA, HI, ID, IL, IN, KY, MI, OH, OR, WA, WI
TRISH FLANAGAN – Northeast US: CT, MA, ME, NH, NJ, NY, RI, VT
JOLEEN GREENWOOD – MidAtlantic US: DE, DC, MD, PA, VA, WV
PAULA TYSON – Southeast US: AL, FL, GA, LA, MS, NC, SC, TN
LOURDES RUBIO – Mountain US: AZ, NV, NM, TX
GABRIEL TORRES – Mountain US: AZ, NV, NM, TX
BREANNE MAIN – Mountain US: CO, MT, UT, WY

Marketing Committee

PAULA TYSON – Eastern USA (Georgia)

STACY PADDON – UK

ALANA FAHEY – Australia

VERA MAKAROVA – Russia

BEN SHAW – UK

ANNA AFZAL-GOULD – UK

ANGELA TRIPP LINDIG – Western USA (Idaho)

HNRNPFF Board of Directors

Leila Margolis, Founder/President is one of our founders. Her son, Sidney, is diagnosed with SYNCRIP/HNRNPQ -Related Neurodevelopmental Disorder. In another life, Leila earned her bachelor of science in sociology at Mt. Saint Mary's University in Los Angeles, CA. She has a background in women's studies, community engagement, and is a trained postpartum doula. Leila and her family felt lost after Sidney was diagnosed. Her hope in starting this organization is not only to further research and treatment, but to make sure no other HNRNP-RNDD family feels alone ever again.

Susan Altschuller, Founder/Treasurer is one of our founders and our treasurer, and mom to Gus, who has HNRNPU-Related Neurodevelopmental Disorder. Susan has two decades of financial management, investor relations, and business planning experience with leading pharmaceutical and biotechnology companies. Most recently, she serves as the Chief Financial Officer of Cerevel Therapeutics. Susan has earned a BSE in Biomedical Engineering with Honors from Tulane University, a Ph.D. in Biomedical Engineering from the Illinois Institute of Technology, and an MBA from the MIT Sloan School of Management.

Amanda Reuther is our **Community Relations Director**. In 2016, after a four-year search for answers, Amanda's daughter Paeyton was diagnosed with HNRNPU-Related Neurodevelopmental Disorder, propelling Amanda into the realm of rare disease advocacy. Amanda has been a successful business owner in San Diego, CA since 2005. Her passion for advocacy led her to complete the Special Education Law and Advocacy Series, earning certification from the University of San Diego School of Law in 2021. Amanda serves as board President for the Special Education Parent Council for Encinitas Union School District.

Dr. Maddie Gillentine, PhD, Research Director, HNRNP Family Foundation, Variant Scientist, Seattle Children's Hospital. Maddie is a genetics researcher and Variant Scientist at Seattle Children's Hospital and a founding member. She currently works with genetic data diagnosing pediatric patients. Maddie did her postdoctoral work in the lab of Dr. Evan Eichler at University of Washington, where she analyzed large data sets of genomic data to identify novel genetic disorders, particularly those impacting gene families. This work was how she identified the larger group of HNRNP-Related Neurodevelopmental Disorders. She currently serves as the Research Director of the HNRNP Family Foundation. Dr. Gillentine's PhD work at Baylor College of Medicine in Houston, Texas was with induced pluripotent stem cell models of neurodevelopmental disorders. She also has two autistic brothers, which fueled her interest in neurodevelopmental disorder genetics.

International Delegates:

KAORI SUZUKI – International Delegate, Japan

About Conference Speakers

Dr. Jennifer Bain, MD, PhD
Assistant Professor of Neurology and Pediatrics
Columbia University Irving Medical Center

Dr. Jennifer Bain, MD, PhD is an assistant professor of neurology and pediatrics at Columbia University Medical Center and the affiliated New York-Presbyterian Hospital System. She is also on the Scientific/Medical Advisory Boards of both YBRP and HNRNPFF. Dr. Bain completed both an MD and PhD, as well as general pediatrics residency at Rutgers – New Jersey Medical School in New Jersey. She then trained in Child Neurology at New York-Presbyterian/Columbia University Irving Medical Center in New York City and is a board-certified neurologist with special certification in Child Neurology seeing both inpatient and outpatient pediatric neurology patients. She works as a physician scientist with expertise in development, behavioral neurology and autism. Her clinical research has focused on studying the genetics of neurodevelopmental disorders including autism and cerebral palsy. Dr. Bain authored the first manuscript describing HNRNPH2 related disorder and is running the HNRNPH2 Natural History Study. She has been working closely with several patient advocacy groups, researchers, and Simons Searchlight to continuously move forward in the understanding of the developing brain.

Dr. Ané Korff, PhD
Senior Scientist
St. Jude Children's Research Hospital

Dr. Ané Korff is a Senior Scientist in the Department of Cell and Molecular Biology at St. Jude Children's Research Hospital (SJCRH). SJCRH is leading cutting-edge research to understand the molecular basis of neurological disorders, elucidate normal cellular functions of disease-associated proteins, and pursue a unique opportunity to accelerate the development of novel therapies for children living with these neurological disorders through the Pediatric Translational Neuroscience Initiative (PTNI). Dr. Korff is currently involved in mouse model studies of HNRNPH2 under the guidance of the Dr. Paul Taylor, MD, PhD., the Director of PTNI.

Rachael Mercer
Program Manager- Alliances, Pediatric Translational Neuroscience Initiative (PTNI) Office of Strategy & Alliances
St. Jude Children's Research Hospital

Rachael joined the Pediatric Translational Neuroscience Initiative (PTNI) at St. Jude Children's Research Hospital in February 2024. As Program Manager-Alliances within the Office of Strategy and Alliances, Rachael builds and manages relationships with external stakeholders, such as patient organizations, and facilitates internal coordination to advance the mission of PTNI. Rachael's background is in patient advocacy. Before joining St. Jude, Rachael worked as a consultant bringing the voice of disease communities to the drug development process. Rachael also has experience providing direct support to patients and families through her role at a national patient organization and as a Child Life Specialist at Le Bonheur Children's Hospital in Memphis, TN.

Dr. Christopher Ricupero, PhD
Assistant Professor & Associate Research Scientist
Columbia University Irving Medical Center

Dr. Christopher Ricupero, PhD is an Assistant Professor at Columbia University Irving Medical Center in New York City. Dr. Ricupero received his PhD in Neuroscience and investigates rare neurological diseases and disorders within the HNRNP gene family. He is currently building a human HNRNPH2-RNDD biorepository for the patient and research communities. This biobank will facilitate HNRNPH2 modeling in a dish using patient stem cells and neurons. His lab is active in precision-based therapeutics development using gene targeting approaches. Dr. Ricupero is a Scientific Advisory Board member of the HNRNP Family Foundation, Better Future 4 U patient advocacy organization (HNRNPU) and member of the Consortium on Neurodevelopmental Studies of Autism Spectrum and Related Disorders at Columbia University.

Dr. Meena Balasubramanian, MBBS, DCH, FRCPCH, MD, FHEA,
Director of Research and Honorary Consultant Clinical Geneticist
Sheffield Children's NHS Foundation Trust
Senior Clinical Lecturer, Division of Clinical Medicine, University of Sheffield

Dr. Meena Balasubramanian was among the first to identify HNRNPU -Related Neurodevelopmental Disorder. She has several studies focused on genotype-phenotype correlation in newly identified genes. She has established her own lab on zebrafish disease models for genetic disorders. Meena is a leading researcher into several rare neurodevelopmental disorders (RNDD) genes and runs natural history studies, including the study for HNRNPU. Meena's group have published the largest clinical cohorts of individuals with variation in HNRNPU and have written the expert literature reviews (GeneReview/Orphanet/NORD guides) for HNRNPU.

Dr. Billie (Ping-Yee) Au, MD, PhD
Clinical Assistant Professor,
Department of Medical Genetics
Department of Pediatrics,
University of Calgary

Dr. Billie Au is a clinical assistant professor and clinical geneticist in Calgary. She did her medical training in the MD/PhD program at the University of Toronto and residency training in University of Calgary. She sees a wide variety of patients but has a particular interest in genetic neurodevelopmental and neurodegenerative disorders. Her research focuses on gene discovery, novel syndrome delineation, and understanding genotype-phenotype relationships. She is also interested in the use of molecular and cellular phenotyping like DNA methylation to clarify and improve diagnosis and to understand outcomes. She has been involved in the initial gene discovery for Au-Kline syndrome and has been working on HNRNPK related conditions ever since. She currently leads the natural history study for HNRNPK related disorders with Dr. Antonie Kline and collaborates with other researchers working on this gene.

About Conference Speakers

Dr. Laurence Mignon, PhD
Executive Director of
Clinical Development
n-Lorem Foundation

Laurence “Laury” Mignon is the executive director of clinical development at n-Lorem Foundation. Laurence has more than 20 years of experience as a seasoned academic researcher and clinical developer with a passion for rare diseases. Most recently, Laurence worked at Ionis Pharmaceuticals both in the Translational Medicine group and the Neurology Franchise Clinical Development group. In those roles, she implemented a novel genomic study to better understand genotype-phenotype associations and disease pathways, initiated the first clinical study in myotonic dystrophy type 1 and was a key member of the clinical team that brought SPINRAZA® (nusinersen), a disease-modifying therapy for patients with spinal muscular atrophy, to the market. Prior to Ionis, Laurence worked at Orexigen Therapeutics on the development of an obesity medication and at the Neuroscience Education Institute developing Continuing Medical Education material aimed at deconstructing the mechanism of action of psychiatric medications for psychiatrists, primary care physicians, and registered nurses. Laurence’s broad and diverse experience allows her to be strategically creative in her approach to deal with uncharted challenges. Laurence received her PhD in Pharmacology and Experimental Therapeutics from Loyola University Chicago and was a researcher for 6 years in the Department of Neurology at UCLA in the laboratory of Marie-Françoise Chesselet, studying properties of muscle-derived stem cells and the role of the serotonin pathway in Parkinson’s disease.

Prof. Rotem Karni
Chief Technology Officer
Andlit Therapeutics

Prof. Rotem Karni is the CTO of Andlit and serves as Chair of the Department of Biochemistry and Molecular Biology at the Institute for Medical Research, Hebrew University - Hadassah Medical School. He holds a Ph.D. in Biochemistry from the Hebrew University and was a postdoctoral fellow in the laboratory of Prof. Adrian Krainer at Cold Spring Harbor Laboratory, NY, applying his knowledge of RNA stability and splicing to search for new therapeutic approaches for rare genetic diseases. Dr. Karni has published his research in top-tier journals and holds several patents related to the technologies he is developing.

Dr. Manar Abu Diab, PhD
Postdoctoral Fellow
Institute for Medical Research
Israel-Canada

Manar Abu Diab, PhD holds a Ph.D. in Human Genetics from the Hebrew University of Jerusalem. She is presently a postdoctoral fellow in the laboratory of Prof. Rotem Karni in the Institute for Medical Research Israel-Canada (IMRIC), at the Faculty of Medicine, Hebrew University of Jerusalem. Manar’s research is an integral part of the HNRNPH2 project at Andlit Therapeutics. Manar is applying her knowledge of RNA stability and splicing to search for new therapeutic approaches for rare genetic diseases.

Dr. Antonie D Kline, MD
Director of Clinical Genetics,
Medical Director of the Randolphs B
Capone Cleft Lip and Palate Program,
Greater Baltimore Medical Center

Dr. Antonie Kline, with Dr. Billie Au, identified and continues to study Au-Kline syndrome and HNRNPK-Related Neurodevelopmental Disorders. She received her medical degree from Jefferson Medical College, Philadelphia, PA, her postdoctoral training in medical genetics at Jefferson Medical College, Philadelphia, PA, and her clinical cytogenetics training at the Kennedy Krieger Institute of the Johns Hopkins University School of Medicine, Baltimore, MD. She is board-certified in clinical genetics, clinical cytogenetics, and clinical molecular genetics. She is also a Fellow of the American Academy of Pediatrics and a Founding Fellow of the American College of Medical Genetics. Her areas of expertise include evaluation of multiple birth defects and/or developmental issues, as well as correlation and interpretation of cytogenetic changes in a clinical setting.

Dr. Terry Jo Bichell, PhD, MPH
Chief Executive Officer
COMBINEDBrain

Dr. Terry Jo Bichell, PhD, MPH 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, to assist other rare and ultra-rare neurogenetic disorders with clinical trial preparations.

Dr. Rosanna Weksberg, M.D., Ph.D.
Clinical Geneticist
The Hospital for Sick Children
Professor of Pediatrics and
Molecular Genetics
University of Toronto

Rosanna Weksberg is a clinical geneticist at the Hospital for Sick Children and professor of pediatrics and molecular genetics at the University of Toronto. Her research focuses on the identification of epigenetic alterations associated with human disease, specifically in the areas of growth and neurodevelopment, including autism. In addition, her research addresses the effects of environmental exposures, including therapeutic agents, on the epigenotype.

About Conference Speakers

Dr. Sean Post, PhD
Associate Professor, Dept. of Leukemia,
Division of Cancer Medicine, MD
Anderson Cancer Center, Houston, TX;
Associate Professor, Dept. of Genetics,
Division of Basic Science Research, MD
Anderson Cancer Center, Houston, TX

Dr. Sean Post is an Associate Professor in the Department of Leukemia at MD Anderson Cancer Center in Houston, Texas. Dr. Post received his Ph.D. in Molecular Medicine from the University of Texas Health Science Center at San Antonio in 2003. Dr. Post trained as postdoctoral fellow in Department of Genetics at the MD Anderson Cancer Center under the tutelage of Dr. Gigi Lozano. Dr. Post's research is focused on identifying and understanding how novel genetic alterations impact cancer progression. He also focuses on mouse models of HNRNPK-Related Neurodevelopmental Disorders, as many of the HNRNP genes are upregulated in cancer. His laboratory's long-term objectives are developing personalized treatment strategies for patients with hematologic malignancies.

Kalayla Hough, MS, CCC-SLP
Speech Language Pathologist

Kalayla is a licensed and ASHA certified speech language pathologist. She holds a Master's degree in Speech Language Pathology from the University of Nebraska at Omaha. Kalayla has experience in pediatric outpatient and in-home settings serving families from a variety of cultural backgrounds. She is a certified Infant Family Specialist who believes that supporting the child-caregiver relationship is essential to making meaningful progress. Kalayla has experience working with children of all ages with language delays, motor speech disorders (e.g., Apraxia), autism, ADHD, and rare genetic disorders. For the past seven years, Kalayla has worked closely with occupational therapists, physical therapists, and special educators. This gives her a unique ability to make appropriate referrals and hold in mind whole child development as it relates to communication.

Laura Barringer
Head of School
Academy for Precision Learning

From her early days as a Special Education teacher, mentor, and instructional coach in NYC public schools, Laura has dedicated her career to advocating for diverse needs. Her transition to consulting with Microsoft's Neurodiversity Hiring Program allowed her to champion inclusive hiring practices, ensuring that opportunities are accessible to all. Currently, as the Head of School at the Academy for Precision Learning, she is privileged to lead an institution that embodies these values and strives to foster an environment where every student feels seen, valued, and empowered.

Katie Schripsema, MOTR/L, MT-BC
Occupational and Music Therapist

Katie Schripsema is a licensed occupational therapist and board-certified music therapist. She holds a Master's degree in Occupational Therapy from the University of New Mexico and Bachelor's degrees in Music Therapy and Flute Performance from Michigan State University in East Lansing, Michigan. Katie's education and experience as both an occupational therapist and music therapist give her a unique skill set that she brings to each therapy session. She is passionate about exploring how music and movement support improved sensory processing and sensory integration.

Dr. Barbara Corneo, PhD
Associate Professor of Rehabilitation and Regenerative Medicine
Director, Stem Cell Core Facility
Columbia University Irving Medical Center

Dr. Barbara Corneo began her academic path in Italy and pursued her PhD in Paris, where she made significant discoveries related to mutations in the Rags genes, particularly in patients with severe combined immune deficiency. Her move to the United States for a post-doc allowed her to further delve into this area, continuing her research on these genes and proteins. Her interests then shifted towards stem cells, leading her to undertake a second post-doc in the lab of Dr. Gordon Keller, a prominent figure in stem cell research. Here, she focused on developing protocols to differentiate human embryonic stem cells into liver and pancreas cells. As a junior investigator, Barbara collaborated with Dr. Sally Temple, a renowned expert in neural stem cells and stem cell therapeutic applications in eye diseases. This is when Barbara first derived induced pluripotent stem cells (iPSCs) from eye tissues, an achievement that deeply captivated her and solidified her commitment to stem cell research. Currently, Barbara serves as an Associate Professor at Columbia University, where she directs the Stem Cell Core. In this role, she oversees a team of four highly skilled scientists who assist researchers by deriving iPSCs from various tissues, differentiating them into different progenitor cells, and utilizing CRISPR technology for gene editing purposes.

Effie Parks
Once Upon A Gene
Podcaster

Effie Parks, originally from beautiful Montana, has become a guiding light in the rare disease community following her son Ford's diagnosis with CTNNB1 syndrome. Settling in Washington, she transformed her family's journey into a crusade for advocacy, support, and empowerment for families navigating similar challenges. As the host of the "Once Upon a Gene" podcast, Effie has been recognized for several awards including WEGO Health and Podcast Magazine for her impactful storytelling and resource-sharing in the realm of rare genetic disorders. Effie extends her advocacy through speaking engagements at medical and patient advocacy conferences, sharing her experiences and insights from her work to bridge the gap between all rare disease stakeholders. Her skill in community engagement, developed through her advocacy, empowers her efforts in building a supportive network and raising awareness. With a mission to leave the world better than she found it, Effie is dedicated to fostering a more informed and empathetic environment for those impacted by rare diseases. Her work embodies resilience and compassion, inspiring and uniting the rare disease community. Effie's journey is not just about sharing stories, it's about driving change and creating a lasting impact in the world of rare genetic conditions.

SPECIAL THANKS TO

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All of our amazing speakers!

Yellow Brick Road Project and HNRNP Family Foundation volunteers who generously devote their time and efforts to further the cause for HNRNP-RNDD patients.

And, of course, to ***all of our families***, the reason we do what we do!

Join us on our road to a cure!

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