January 12, 2024

FGFR Syndromes Collaborative Research Network Conference

Speaker and Moderator Biographies

Michael Bober, MD, PhD, *Vice President, Clinical Development and Medical Affairs at Tyra Biosciences. He is a pediatrician and medical geneticist. He was an expert in the diagnosis and management of skeletal dysplasias before transitioning into industry and drug development.*

Ashley Brock *is a 19 year old and has Apert Syndrome. Ashley has had 52 surgeries to date. Her craniofacial team is in Dallas and she lives near Houston, Texas. Her best friend Olivia has Pfeiffer syndrome. They try to meet up for doctor appointments and surgeries to make it more fun.*

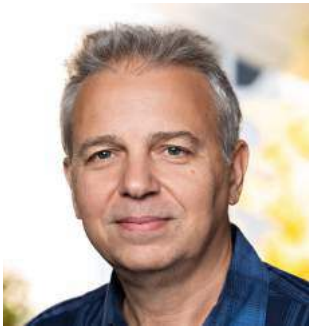
Lia Burton *is an award-winning set designer for film and TV, and has designed for shows like HBO's Insecure and Euphoria. She was born with Pfeiffer Syndrome. She lives in Los Angeles with her dog, Salami.*

Marcela Cabal Castro MD, MS,*. a Colombian plastic surgeon graduated in 2010 from Universidad el Bosque, did a subspecialty in maxillofacial and Craniofacial surgery in Fundación Universitaria de Ciencias de la Salud and has been working with both aesthetic and reconstructive surgery for more than 12 year in Cali Colombia and working in the formation of a Craniofacial clinic that can help people from the Colombian southwest region, including multidisciplinary attention and longtime follow up of the treatment processes. Also operating Smile voluntarily for many years and teaching in Universidad El Valle Cali Colombia in the Plastic Surgery section.*

Rosie Comprosky *was born in the fall of 2018 at home, Rosie's family was unprepared for her needs as she had no pre-diagnoses. Within 45 minutes of her birth, she was rushed to the hospital and diagnosed with several rare conditions rarely seen together. Despite stumping her doctors initially, Rosie continues to defy the odds. Facing severe complications after a tracheostomy placement, her tenacious spirit prevailed, and she is now a spunky, spirited, joyous 5-year-old. Rosie, known for saying 'bet' whenever someone doubts her abilities, has become a beloved friend to her community. Just say 'hi,' and you've got a new best friend.*

Jake Dahl. MD, PhD, MBA, *is a pediatric otolaryngologist at Seattle Children's Hospital and associate professor in the Department of Otolaryngology-Head and Neck Surgery at the University of Washington. Dr. Dahl has a significant interest in basic and clinical scientific research related to vascular anomalies, tracheal malformations, airway management in patients with complex craniofacial disorders, and patient quality and safety.*

Emily Durham, PhD, *joined The TBCK Foundation in July 2023 as the Research Engagement Director. She holds a PhD in biomedical sciences from the Medical University of South Carolina and an MA in research ethics from Duquesne University. Currently, Emily is a postdoctoral fellow at Children's Hospital of Philadelphia where she studies skull and brain development and changes caused by loss of TBCK and other neurodegenerative conditions.*



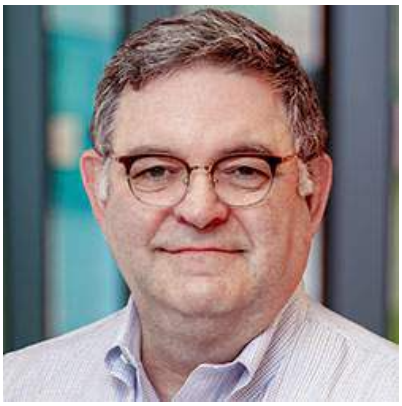
Aris N. Economides*,* ***PhD****, (VP, Regeneron) leads two groups: Connective Tissue Diseases, and Synthetic Biology. Research interests include drug development for skeletal diseases, and mechanisms of BMP/TGFß signaling.*

Kristy Lund, RN, *a mother of three and an 18-year Labor and Delivery Registered Nurse, initially experienced the joy of welcoming two healthy boys. However, the journey took an unexpected turn with the arrival of her baby girl, Hadley. Following numerous prenatal appointments and diagnostic procedures, an FGFR mutation was confirmed after 11 weeks of uncertainty. Introduced to Pfeiffer Syndrome, a supportive community on social media became a source of strength. Over the past 7 years, Kristy has navigated the challenges and joys of caring for Hadley and is now empowered to help others facing similar journeys.*

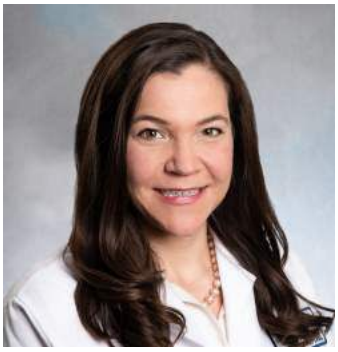
Ellen Morgan*, founder of the 501(c)(3) non-profit PROS Foundation, has >25 years of drug development experience in various sectors, including large pharma, small biotech, and CRO. Her expertise extends to leading successful Clinical Research Organizations, such as Synteract and Agility Clinical. Previously serving as President, Rare Diseases at Precision for Medicine, Ellen is dedicated to accelerating effective treatments for rare diseases through PROS. She holds a BS in Chemistry and an MS in Management and Industrial Engineering from Rensselaer Polytechnic Institute, with post-graduate studies in statistics and genetics.*

Susan Motch Perrine PhD, *received her B.S. and M.S. degrees in Animal Sciences from Penn State, then went on to complete her doctorate in the Intercollege Graduate Degree Program in Physiology between the Penn State Medical School and University Park campuses. She completed a post-doc in craniofacial research in the lab of Dr. Joan Richtsmeier and continued on with research in the Richtsmeier lab until July 2023 when Dr. Richtsmeier retired. She and colleague Dr. Kazuhiko Kawasaki now lead the lab and continue pursuit of developmental craniofacial research. She currently holds a position as Associate Research Professor in the Department of Anthropology at Penn State, and dedicates the rest of her time to family, the arts, and horse-inclusive activities.*

***Ken Nischal, MD, FAAP, FRCOphth**, is the Chief of the Division of Pediatric Ophthalmology and Strabismus at UPMC Children's Hospital of Pittsburgh, Director of Pediatric Program Development at the UPMC Eye Center, and Professor of Ophthalmology at the University of Pittsburgh School of Medicine. Dr. Nischal graduated from Kings College Hospital, London , did his Ophthalmology at Oxford and his fellowship in pediatric ophthalmology at SickKids, Toronto. He was appointed to Great Ormond St. Hospital for Children, London in 1999 and recruited by UPMC Children's Hospital in 2011.*



***Jonathan Perkins**, **DO,** Professor, Pediatric Otolaryngologist, is the Director of the Seattle Children's Hospital's Vascular Anomalies Program, for the past twenty-three years. In this role he has used his biorepository of surgically obtained vascular anomaly tissue samples to collaborate with developmental vascular biologist, Mark Majesky and geneticists William Dobyns and James Bennett to discover aspects of cellular biology and genetics for some vascular anomalies. This information has formed the basis for new vascular anomaly diagnostic tests, using cell-free DNA and targeted therapies, using kinase inhibitors.*



***Rebecca Reimers MD, M.P.H, F.A.C.O.G** Maternal-fetal medicine and medical geneticist at San Diego Perinatal and Rady's Children's Hospital. She is a perinatologist and clinical geneticist trained in the Harvard system at Brigham and Women's, Massachusetts General, and Boston Children's Hospital. She is currently the principal investigator for a study on whole genome sequencing in fetuses with congenital heart disease and is interested in prenatal diagnosis and therapy for health issues that affect the health of children and families.*

***Rosanna Samuel, DClinPsy**, is the Senior Clinical Psychologist in the Oxford Craniofacial Unit at Oxford University Hospitals NHS. Rosanna earned her doctorate in Clinical Psychology in 2019 and has worked in the craniofacial service since then. Working with children and their families and supporting them through surgeries and psychological distress associated with visible differences is her dream job.*

***Sabrina Robineau-Seitz** was born with Pfeiffer Syndrome, Type 1 and a congenital heart defect. She has had 24 surgeries. Sabrina was born and raised in Quebec, Canada and then she moved to the United States in 2019. She has a bachelor's degree in psychology and an associate's degree in early childhood education. She is fluent in French and American Sign Language. For the past 8 years, she worked with children of all ages (from 6 months to 12 years old). Sabrina believes it is important to spread awareness about craniofacial conditions and facial differences so that the world can be a more accepting place for everyone.*

***Atlas Shanklin** is 8 years old and lives in Bellevue, Washington with his parents, two sisters, a dog and cat, and his rabbit, "Thumper." He enjoys acting, dancing, playing baseball, and learning about science. Atlas has Crouzon syndrome and enjoys chatting it up with the doctors in the Craniofacial unit at Seattle Children's. His favorite thing to do after seeing the doctors in the clinic is to go get a donut in the cafeteria.*

***Savannah Smith**, a 13-year-old with a passion for modeling, dancing, music, singing, performing, and traveling. She attends weekly ballet classes and actively participates in her church's Stars Ministry, assisting special needs community members. An avid Jeep enthusiast, Savannah is involved in local Jeep and car clubs. As an advocate and model for the non-profit "I'm\|Possible," she promotes acceptance and inclusion. Savannah started modeling at 9, walking in four NYFW fashion shows in 2020 and has since been in over 25 shows and featured in six fashion magazines. She also enjoys attending CCA Retreats.*

***Carolina Sommer,** CEO/Founder of Born a Hero Research Foundation, Co-Founder of NW Rare Disease Coalition, and Founder of Seattle Rare Disease Fair, leads studies on 20 FGFR Syndromes. She spearheads various programs, including the Seattle Rare Disease Fair and NW Rare Disease Coalition initiatives. Additionally, Carolina co-founded the ABC Kind Program, a national curriculum promoting acceptance of differences, and authored the Lucy's Journey books. With a Bachelor's degree in Theoretical Math from the University of Washington, Carolina is actively involved in advocacy groups like Rare Disease Access Working Group, EveryLife Foundation, We Work for Health, Voters for Cures, and WA Health Access Network. Born in Medellin, Colombia, Carolina has resided in the Seattle area since childhood and was a NASA Jet Propulsion Laboratory intern.*

Helen Webb-Prosser was born in Vermont in 1988 and diagnosed with Pfeiffer Syndrome. Their Craniofacial team was in Boston. Helen works in Special Education. They live in Maine with their wife and 8-year-old son.

Patricia (Patti) Winters *began her career as a prenatal genetic counselor at various academic centers. Patti is now an Associate Director in Medical Affairs at Illumina, focusing on the responsible implementation of NIPT into clinical practice.*

For more information about the FGFR Syndromes Collaborative Research Network Conference, please visit fgfr.org.