# Annique HOGAN

## topic

**THE INS AND OUTS: MANAGING GI ISSUES IN GM1**

International GM1 Virtual
CURE GM1 COMMUNITY CONFERENCE

*Friday September 20,2024*

Annique K. Hogan, MD is a complex care general pediatrician at The Children's Hospital of Philadelphia (CHOP) and an Associate Professor of Clinical Pediatrics at the Perelman School of Medicine at the University of Pennsylvania. She has spent her career at CHOP, dedicated to the care of medically complex patients both in the inpatient and the ambulatory setting. She is the Medical Director of the inpatient Complex Care Service, the founder and Medical Director of the CHOP Compass Care program, the Medical Director of Care Management, and the Medical Director for CHOP Home Care. She loves creating and improving systems of care for children with medical complexity, teaching about complex care, and caring for these incredible children and their families.

# Cara WEISMANN

## topic

**ACADEMIA TO CLINICAL TRIALS**

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Dr. Weismann is the Director for the Program of Excellence (POE) in Mucopolysaccharidosis (MPS) and Lysosomal Storage Disease Advanced Therapies at the non-profit organization the Orphan Disease Center. Within the POE, she leads a grant program whose goal is to accelerate the development of therapeutics and provide scientific resources to the MPS and lysosomal storage disease communities. She has extensive experience in therapeutic research, drug development, business development, and data sharing policy. She is passionate about using these skills to bring treatments to rare disease patients throughout the world.

# Christian FREITAG

## topic

**AZAFAROS CLINICAL PROGRAM: UPDATE ON PRONTO AND RAINBOW STUDY DATA AND PLANNING FOR PHASE 3 PIVOTAL STUDY**

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Dr. Freitag obtained his medical degree from Kiel University, Germany. After several years in different hospital posts, he started his career in the pharmaceutical industry at Roche. After A few different roles in different clinical development, he started working fully in the rare disease area in 2018. He joined Azafaros in 2022 and has responsibility for the clinical development and medical management of the clinical programme.

# Christine WAGGONER

## topic

**CURE GM1 COMMUNITY CONFERENCE – CURE GM1 OVERVIEW, INITIATIVES, AND RESEARCH**

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Christine Waggoner and her husband Douglas Dooley founded the Cure GM1 Foundation in April 2015 in honor of their daughter Iris and all those affected by GM1 gangliosidosis. Christine received the Sanofi TORCH award in 2017 for outstanding patient advocacy and she also received the WORLD Symposium Patient Advocacy Leadership Award in 2023. Founding and running Cure GM1 is a true labor of love to help bring a treatment to all those affected by GM1 gangliosidosis.

# Daisy NG-MAK

## topic

**BURDEN OF CAREGIVING IN GM1 GANGLIOSIDOSIS**

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*Friday September 20,2024*


Daisy Ng-Mak is a Business Partner of the Health Economics & Value Assessment (HEVA) Rare Disease at Sanofi.

Daisy is a researcher and a leader with extensive experience in strategizing health economics and outcomes research and developing market access and reimbursement strategies in the United States, Europe, and Asia.

Daisy received her Doctorate Degree (PhD) in Epidemiology and 2 Masters Degrees (MPhil in Epidemiology, MS in Biostatistics) from Columbia University in the City of New York.

# David LAW

## topic

**CAREGIVING AND COMMUNITY**

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Dr. David Law is the father of Violet Aurora and Derek Sison Law. Violet was diagnosed with late-infantile GM1 gangliosidosis and passed away at age 4. He is an internal medicine physician who trained at the UC Davis Medical Center and Harbor-UCLA Medical Center, and currently practices hospital medicine and palliative care medicine in southern California. He has been married to his wife, Dr. Veena Sison, a developmental pediatrician, since 2014 and together they enjoy traveling, attending live music events, and are the co-founders of 3rd Call Productions, an award-winning independent film studio in Los Angeles. They use the majority of their time now advocating for rare disease awareness and research.

# Dawn BLESSING

## topic

**CLINICAL TRIALS, PRECLINICAL RESEARCH, AND NATURAL HISTORY**

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***Friday September 20,2024***

Dawn Blessing is Chief Business Officer at Neucore Bio and was recently Vice President of Scientific Strategy and Corporate Development at Zogenix. Prior to Zogenix, she served as Senior Vice President Corporate Development at Modis Therapeutics and previously Vice President, Corporate Development at Audentes Therapeutics. She has 30 years of experience in biotechnology finance, business development, and alliance management. Over this period, Ms. Blessing has focused on programs for rare diseases and the application of genetic information to drug development. Before joining Audentes, Ms. Blessing served as Senior Director, Business Development and Alliance Management at 23andMe, and Director of Business Development at BioMarin Pharmaceutical.

# James WILSON

## topic

**GEMMA BIOTHERAPEUTICS GM1 AAV GENE THERAPY**

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James M. Wilson, MD, PhD, is a Professor in the Perelman School of Medicine at the University of Pennsylvania where he has led an effort to develop the field of gene therapy. His research career spanning over 40 years has focused on rare diseases and ways to treat them by gene therapy. Dr. Wilson has published over 600 papers and is named on more than 1200 patents worldwide. The Wilson lab identified a new type of vector based on novel isolates of adeno-associated viruses which have become best in class for gene therapy. More recently Dr. Wilson's laboratory has focused on improved vectors for gene therapy and clinical applications of genome editing and mRNA therapy.

# Jean JOHNSTON

## topic

**GM1 GANGLIOSIDOSIS TYPE II: RESULTS OF A 10-YEAR PROSPECTIVE NATURAL HISTORY STUDY**

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Jean Johnston is an Associate Investigator and Study Coordinator at the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH). Mrs. Johnston holds degrees in Public Health, Nursing, and Clinical Research Management. Mrs. Johnston joined Dr. Cynthia Tifft's study on the "Natural History of Glycosphingolipid Storage Disorders and Glycoprotein Disorders" in 2012. Mrs. Johnston played an integral role in writing the first-in-human gene therapy study for patients with Type I and II GM1 in 2019. She is currently managing both the natural history study and the "Phase 1/2 Study of Intravenous Gene Transfer With an AAV9 Vector Expressing Human Beta-galactosidase in Type I and Type II GM1 Gangliosidosis" study.

# Jennifer SIEDMAN

## topic

**COURAGEOUS PARENTS NETWORK: WHAT DO I SAY NOW? HOW TO ANSWER DIFFICULT QUESTIONS?**

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A bereaved mom, Jennifer brings her experience as a teacher and development professional to her role in engaging patient and caregiver organizations, industry partners and healthcare providers. A rare disease mom, she serves as President of Ben's Dream: Sanflippo Research Foundation and worked with researchers and patient advocacy groups worldwide to fund and advance gene therapies to the clinical trial stage. In addition to co-authoring Supporting Families Considering Participation in a Clinical Trial, and NeuroJourney.org she has been recognized with a Bosto Celtics Heroes Among Us award, Global Genes RARE Champion of Hope and Sano Torch Award. Jennifer received her M.Ed in middle school education from Lesley University.

# Kathleen KIRBY

## topic

**CURE GM1 DATA SHARING INITIATIVE**

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Kathleen Kirby has over 30 years of drug development experience in Clinical Development Operations and Program Management. She is well versed in strategic planning and preparation of product / clinical development plans, and establishes a bridge between program development, clinical strategy and clinical trial execution in early phase start up companies focusing on gene therapies, rare diseases, patient recruitment strategies and natural history studies. Kathleen made significant contributions during early phase development of SKYSONA (eli-cel) in Cerebral Adrenoleukodystrophy and ZYNTEGLO (beta-cel) in Beta Thalassemia. Kathleen supports Cure GM1 and other rare disease foundations.

# Kensho IWANAGA

## topic

**RESPIRATORY CARE FOR GM1 GANGLIOSIDOSIS**

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Kensho Iwanaga is a pediatric pulmonologist who cares for infants, children, adolescents, and young adults with acute and chronic respiratory conditions. He earned his medical degree at the University of California, Irvine. He completed general pediatric residency training at UCSF Benioff Children's Hospital Oakland and pediatric pulmonology fellowship training at Seattle Children's Hospital.

# Kylie HARRISON

## topic

**ADVOCACY AND PATIENT-LED EFFORTS**

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Kylie lives in Colorado with her 4 daughters and husband, Kyle. In June of 2020 her two oldest daughters, Kinley and Kennedy, were diagnosed with Juvenile GM1. Since learning of her daughter's diagnosis, she and Kyle have dedicated much of their time to raising awareness and funding to advance treatments for all children with GM1. Kylie recently became a Certified Nursing Assistant and is now paid to take care of Kinley and Kennedy. The Harrison family has been fundraising to support research and development of treatment options for this horrific disease since the girls were diagnosed in 2020. To date they have raised over $350,000 for the foundation.

# Maria KEFALAS

## topic

**THE PATH TO APPROVAL: WHAT I LEARNED GETTING THE WORLD'S MOST EXPENSIVE DRUG APPROVED**

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CURE GM1

*Friday September 20,2024*

Maria Kefalas became a patient advocate after her late daughter Calliope was diagnosed with metachromatic leukodystrophy (MLD) in 2013. Her work has been featured in STAT, Slate, The Philadelphia Inquirer, and CBS Sunday Morning with Jane Pauley. She has spoken at the FDA, NORD, and the NIH about the promise of gene therapy. Her most recent book is a memoir titledHarnessing Grief: A Mother's Quest for Meaning and Miracles. On the second anniversary of Cal's death, the FDA approved Lenmeldy, a gene therapy to treat children with MLD.

# Mary MCDIRMID

## topic

**SUPPORTING YOUR FAMILY WITH A SPECIAL NEEDS MASTER PLAN**

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When Mary isn't helping families navigate the world of special needs financial planning, she is being a wife to Jay and a mom to Charlie and Ruth. Not to mention her leadership role as founder of All Needs Planning and a volunteer advocate for kids with rare diseases. She takes the title "Mom Boss" seriously, just ask her!

# Neena NIZAR

## topic

**ULTRA RARE: PATHWAY TO TREATMENTS**

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Dr. Neena Nizar serves as Founder and Executive Director of the Jansen's Foundation,. Nizar is a TEDx speaker, a blogger, and a disability and inclusion advocate. Nizar is also a rare disease patient herself and a mother to two boys with Jansen's Disease, and advocates for the authentic engagement of patient and caregiver voices in the drug development process for rare diseases. She is WebMD Healthcare Hero for 2023, serves on the Mayor's Commision for Disability and was recently featured in People Magazine and York Times for her work in rare disease drug development and patient advocacy. She is the Director of Patient Advocacy Strategy at ICON Plc, a global CRO. In this role she works to ensure patients are centered in the drug development process.

# Precilla D'SOUZA

## topic

**INTRAVENOUS DELIVERY OF AAV9-GLB1 GENE THERAPY FOR GM1 GANGLIOSIDOSIS AND THE SAFETY OUTCOMES**

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Precilla D'Souza is a Pediatric Nurse practitioner at the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH). She is an Associate Investigator of the Natural History of Neurodegeneration in the Glycosphingolipid and Glycoprotein Disorders and Associate Investigator for IND18831, A Phase 1/2 study of Intravenous Gene Transfer with an AAV9 Vector Expressing Human β-galactosidase in Type I and Type II GM1 Gangliosidosis. Dr. D'Souza has more than 30 years of experience in healthcare, including 22 years as aPediatric Nurse Practitioner in inpatient care, primary care, surgery, and pediatric clinical research settings. She joined the National Institute of Mental Health in 2010 as an associate investigator multiple trials including autism spectrum disorder, PANDAS, the early detection of autism in toddlers, and Phelan McDermid Syndrome. In 2016, she joined NHGRI as a primary care provider for the NIH Undiagnosed Disease Program and associate investigator for studies on the natural history of glycosphingolipid and glycoprotein disorders and the AAV9 gene therapy for GM1 patients. Dr. D'Souza received her Doctorate in Nursing Practice (DNP) fromthe University of Maryland School of Nursing in 2018.

# Rafael ESCANDON

## topic

**GENE THERAPY: FROM THE BASICS TO ITS APPLICATION IN GM1**

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Rafael “Rafa” Escandon has worked in global clinical research and development and patient advocacy in the biotechnology industry for three decades and is currently a consultant to the biotechnology industry. His approach is to be highly considerate of the ethical implications of conducting and participating in clinical research, especially with advanced therapies in rare conditions, as well as in vulnerable and developing-world populations. He has been a member of the NYU Grossman School of Medicine’s Department of Bioethics Pediatric Gene Therapy Medical Ethics Working Group (PGTME) since 2019, and is a contributing graduate faculty member in Pediatric Bioethics at the Children’s Mercy Bioethics Center in Kansas City Missouri. Rafa has also served as adjunct faculty at the University of California Berkeley’s Clinical Research Management Program and as an educator in human rights and justice. He holds graduate degrees from the University of Maryland at Baltimore, Walden University of Minnesota, and the Johns Hopkins Bloomberg School of Public Health, and is a certified healthcare ethics consultant.

# Rebecca STOCKLEY

## topic

**GM1 FAMILY SERIES SPEAKING COACH**

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*Friday September 20,2024*

Rebecca is an executive coach, working with people to optimize their communication and their presentations. She is also one of the most sought-after improv teachers in the United States. In addition to working with numerous top universities and theater schools, her corporate clients include Pixar Animation Studios, Lucasfilm, and Electronic Arts. As a pioneer in the innovative field of Applied Improv, Rebecca was named an Apple Distinguished Educator in 2005. A founding member of BATS Improv, Rebecca was the first dean of the BATS School of Improv, celebrating over 35 years of improvisation leadership. With a BFA from the University of Washington's Professional Actor Training Program, she continues to be an active performer.

# Sharon KING

## topic

**IMPOSSIBLE TO POSSIBLE: BRINGING LIGHT TO POSSIBILITY**

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Sharon is a passionate patient advocate who has united public officials, researchers, biotech and industry representatives, and other patient advocates to catalyze rare disease treatment development and critical public policies. Sharon is Manager of Advocacy and Community Engagement at Aldevron, a leading manufacturer of biologics. There, she works to educate, connect, and improve understanding across the stakeholder community in the biotech space. Using her experience from more than two decades of community service, Sharon co-founded and serves as president of Taylor's Tale, the public charity named for her late daughter to raise awareness and funding for CLN1 disease (Batten disease) research and advocate for the needs of rare disease patients. Since 2006, Sharon has worked with other dedicated volunteers to build a grassroots organization into a leading force against rare disease. She is a state-appointed member of the North Carolina Advisory Council on Rare Diseases. The law that established the Council – the nation's first – is named for her daughter. Sharon received her bachelor's degree from Meredith College. She and her husband live close to their children and grandchildren in Charlotte.

# Sonal PATEL

## topic

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Sonal Patel is a pediatric gastroenterologist and parent of a child with late-infantile metachromatic leukodystrophy. She has extensive experience with enteral feeding and feeding difficulties.

# Timothy STUHLMILLER

## topic

**XCURES PLATFORM FOR PATIENT-CENTRIC DECENTRALIZED RESEARCH IN RARE DISEASE**

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Timothy J. Stuhlmiller, PhD, is VP of Scientific and Medical Affairs with xCures, a health data technology company for patient-driven decentralized clinical research. Tim received his PhD from Yale in Molecular Biology and postdoctoral training at UNC Pharmacology applying systems biology to cancer. His industry experience includes clinical development, competitive market analysis, and strategic asset positioning. At xCures, Tim leads clinician engagement, supports xCures clinical programs, and leads academic and non-profit partnerships to advance scientific interests.

# Walter ACOSTA

## topic

**TREATMENT OF GM1 GANGLIOSIDOSIS BY AAV SYSTEMIC EXPRESSION OF LECTIN-ENHANCED ENZYME**

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Dr. Acosta is a molecular and cell biologist with a deep-rooted passion for developing innovative therapies for rare genetic diseases. He has pioneered the use of plant lectins as novel drug delivery systems, leading to significant preclinical advancements for treatment of lysosomal storage disorders. As CSO of BioStrategies, Dr. Acosta leads the development of a robust product pipeline, securing multiple NIH grants to conduct groundbreaking research. His expertise spans from fundamental research to clinical translation, with a focus on transforming scientific discoveries into life-changing treatments for patients with unmet medical needs.”

# GM1 FAMILY SERIES

Stephanie
HOUGH

Olivia
CROSBY

Maria
BACON

Martha
LOUDERBACK