JUNE 28 - 30, 2026

Conference Proceedings

TANGO2RESEARCH FOUNDATION

CONTENTS

ABOUT THE FOUNDATION

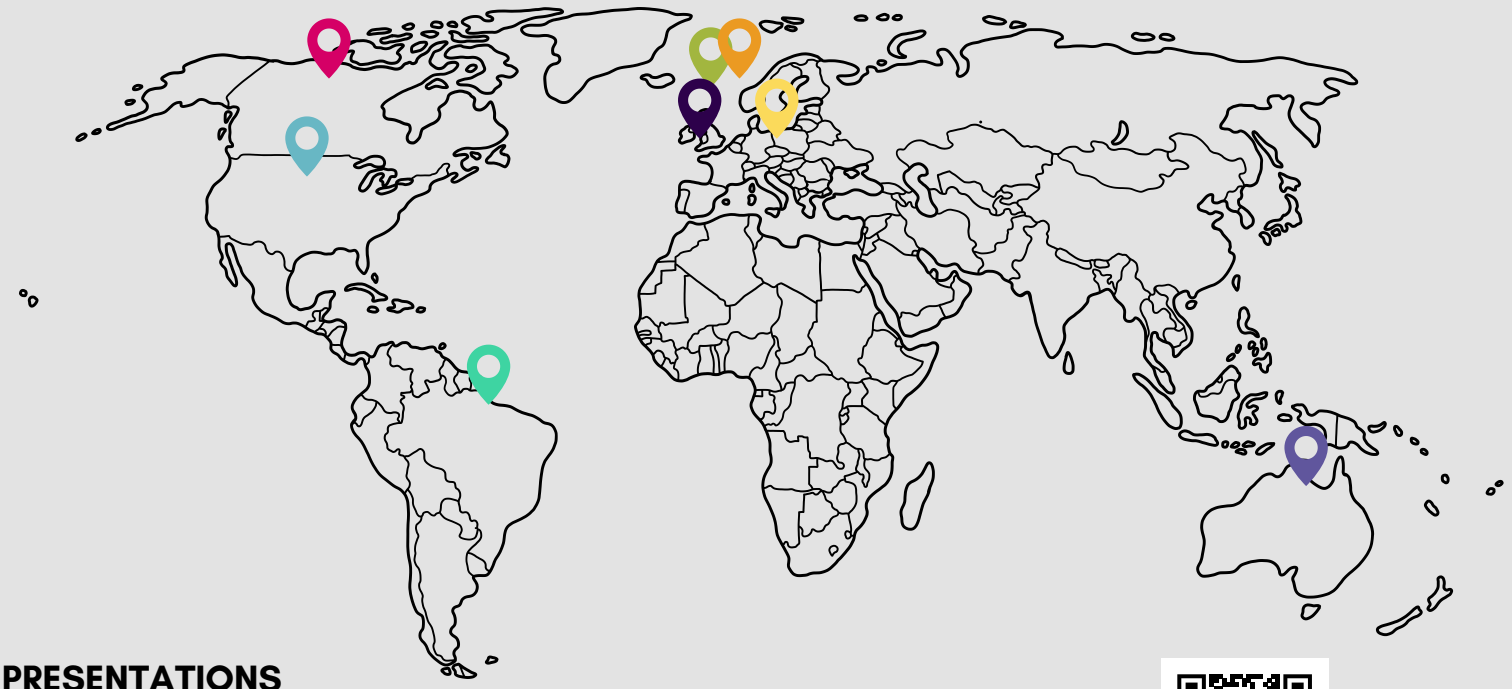
The TANGO2 Research Foundation (T2RF) is a 501(c)(3) nonprofit organization committed to accelerating research, promoting disease awareness, supporting families, and driving accurate, early diagnosis. Our goal is to enhance the lives of individuals affected by TANGO2 deficiency disorder (TDD) by supporting scientific research and fostering understanding of TANGO2 mutations.

2026 TANGO2 FAMILY CONFERENCE

The TANGO2 Research Foundation proudly hosted its 4th Biennial Family Conference at Disney's Coronado Springs Resort in Orlando, Florida. Over two and a half days, the conference featured scientific presentations, educational sessions, interactive workshops, and community-building activities designed to foster learning, collaboration, and engagement across the TANGO2 community. The program created opportunities to share the latest research advances, discuss emerging standards of care, highlight lived experiences, and strengthen partnerships among families, researchers, clinicians, and other stakeholders. Together, these collective efforts continue to accelerate progress toward improved treatments, better clinical care, and ultimately, a cure for TANGO2 deficiency disorder (TDD). We are grateful to everyone who contributed to the success of this year's conference. Thank you for joining us!

ATTENDEES

The 2026 TANGO2 Family Conference convened 195 participants from around the world, representing a diverse community of individuals committed to improving the lives of those affected by TANGO2 deficiency disorder. Attendees included individuals living with TANGO2, family members and caregivers, researchers, clinicians, industry representatives, and nonprofit partners. Caregivers shared powerful personal experiences that highlighted the realities of living with TANGO2 deficiency disorder and reinforced the critical role of patient and family voices in shaping research and care. Researchers, physicians, and other healthcare professionals presented the latest scientific discoveries, emerging therapeutic approaches, and clinical care advancements. By bringing together families, scientists, clinicians, and advocates in a shared learning environment, the conference fostered meaningful collaboration, strengthened partnerships, and reinforced a collective commitment to accelerating research, improving clinical care, and advancing the mission of the TANGO2 Research Foundation.



PRESENTATIONS

Scan the QR code to view recordings from the 2026 TANGO2 Family Conference on the TANGO2 Research Foundation YouTube channel.

BUILDING CONNECTIONS, ADVANCING DISCOVERY: HIGHLIGHTS FROM THE TANGO2 COMMUNITY

The 2026 TANGO2 Family Conference brought together families, clinicians, researchers, and advocates from around the world to connect, share knowledge, and advance progress in TANGO2 Deficiency Disorder (TDD). The conference featured a Poster Hall highlighting emerging research discoveries, clinical insights, and family-led projects, as well as the 2026 Emerging Researchers program, which recognized early-career scientists and clinicians advancing the future of TANGO2 research. The Shine a Light on TANGO2 Celebration brought the community together to honor the strength, resilience, and collective impact of families while celebrating progress toward improved treatments and a cure.

The conference also provided opportunities for families to directly contribute to research through onsite enrollment in the TDD Natural History Study, strengthening efforts to better understand disease progression and outcomes. Attendees also participated in Veggie Meter® readings, providing insights into nutrition-related measures while supporting ongoing efforts to better understand health and wellness in the TANGO2 community. Together, these activities reflected the power of collaboration among families, researchers, and clinicians to accelerate discovery and improve care for individuals living with TDD.

POSTER HALL

A key component of the conference was the 2026 poster program, which reflected the continued growth and engagement of the global TDD community. A total of 26 poster abstracts were accepted for presentation. Notably, submissions were contributed not only by members of the scientific and clinical communities, but also by members of the TANGO2 family community, underscoring the conference's commitment to inclusive, patient-centered collaboration and the value of lived experience in advancing rare disease research and advocacy.

SCAN TO VIEW PUBLISHED FAMILY AND SCIENTIFIC POSTER ABSTRACTS

SHINE A LIGHT ON TANGO2 CELEBRATION

The 2026 Shine a Light on TANGO2 Celebration at the TANGO2 Family Conference highlighted significant achievements in fundraising and research for TANGO2 Deficiency Disorder (TDD). The event celebrated the campaign's growth, raising over $100,000 with 410 donors and widespread international engagement of 50 participants. Special recognition goes to top fundraisers Veronica Jones-Swetek, The Galland Family, Matt Dort, Cynthia Garcia, and Lisa Wilson. Top teams were Caelan's Crew at Lorian BelAir, Team Ryan, Team Jake, Tyler's Crew, and Team Lorenzo. The event underscored the impact of community contributions on TDD studies, notably in personalized therapies and treatment advancements. A tribute to community resilience and research partnerships, the celebration reaffirmed TANGO2 Research Foundation's mission to fund impactful research and enhance patient outcomes.

2026 T2RF EMERGING RESEARCHERS

This year, the TANGO2 Research Foundation proudly recognized its second cohort of Emerging Researchers, honoring early-career investigators whose innovation, dedication, and scientific contributions are advancing the understanding of TANGO2 Deficiency Disorder. Through their research and commitment to improving patient outcomes, these recipients represent the next generation of leaders in TANGO2 research and bring renewed hope to individuals and families affected by this rare disease.

TANGO2 NATURAL HISTORY & NUTRITION STUDIES

Conference attendees had the opportunity to participate in onsite enrollment for the TANGO2 Natural History Study and nutrition assessments using the Veggie Meter®, a non-invasive measure of skin carotenoid levels. These research activities provide valuable data to advance understanding of the natural history of TANGO2 Deficiency Disorder and the role of nutrition in health outcomes. Individuals interested in participating after the conference may contact tango2research@bcm.edu for information about the TANGO2 Natural History Study or bose@montclair.edu to learn more about the TANGO2 Nutrition Study.

Mini scientific symposium sessions

Sunday, June 28th, 2026

TANGO2: From Mitochondrial Lumen Localization to Lipid-Chain Interactions

Agustin Lujan, MD, PhD

Agustín Luján, MD, PhD is a physician-scientist dedicated to advancing research in pediatric rare diseases, including conditions like TANGO2 deficiency. He holds a PhD in biological sciences and postdoctoral training in cell and molecular biology. At Hospital Sant Joan de Déu, he works as a researcher in the neuromuscular and arrhythmia/sudden death units, serving as a bridge between basic and clinical in this field. Dr. Lujan currently serves as a T2RF Research Committee member.

Session Highlights

Dr. Agustín Luján presented emerging research that advances the understanding of TANGO2 protein biology and its role in TANGO2 deficiency disorder. Building on previous work, he shared evidence demonstrating that TANGO2 localizes within the mitochondrial lumen, with the protein's LIL domain directing mitochondrial localization and the NRDE domain mediating acyl-CoA binding. His team also presented findings showing that TANGO2 forms oligomers, contains a molecular pocket capable of accommodating acyl-CoA, and functions as an acyl-CoA-binding protein, supporting a potential role in lipid metabolism. The presentation highlighted how disruption of TANGO2 alters lipid composition and metabolism, offering new insights into disease mechanisms and identifying promising directions for future therapeutic development. Dr. Luján concluded by discussing ongoing studies focused on biomarker discovery and key unanswered questions surrounding TANGO2 function, mitochondrial biology, and opportunities for targeted treatment strategies.

Unveiling Novel Insights into the Pathophysiology of TDD Using Integrated Functional and Multiomic Studies

Lina Ghaloul-Gonzalez, MD

Lina Ghaloul-Gonzalez, MD, is a clinical and metabolic geneticist and Mellon Scholar at UPMC Children's Hospital of Pittsburgh. She co-founded the Plain Community Translational Medicine Program at UPMC, which provides clinical care and research opportunities for patients from the Plain communities (Amish & Mennonite), primarily in western Pennsylvania. She leads multiple clinical and research initiatives within this population. She also serves on the Research Committee of the T2RF, and her laboratory is dedicated to understanding the function of TANGO2 and the pathophysiology of TDD.

Session Highlights

Dr. Lina Ghaloul-Gonzalez presented findings from integrated multiomic and functional studies that provide new insights into the molecular mechanisms underlying TDD. Her research identified a critical bottleneck in coenzyme A (CoA) metabolism that disrupts cellular energy production and lipid homeostasis. While vitamin B5 partially corrected metabolic abnormalities, combining vitamin B5 with the PanK activator PZ-3022 significantly improved mitochondrial function, highlighting the potential of combination therapies that target both CoA biosynthesis and utilization. These findings provide a deeper understanding of the molecular pathways disrupted in TDD and establish a foundation for identifying targeted therapeutic approaches to address the underlying metabolic dysfunction.

Metabolic Crisis Causes QT Prolongation in TANGO2 Deficiency Disorder

Lili Wang, PhD

Lili Wang, PhD is a Research Instructor in the Department of Medicine at Vanderbilt University Medical Center, where she investigates the arrhythmia mechanisms underlying genetic cardiac disorders. Her research centers on how metabolic stress disrupts cardiac electrophysiology, using hiPSC-derived cardiomyocyte models to dissect disease mechanisms at the cellular level. She will be presenting her work on how metabolic crisis drives QT prolongation in TDD Convening.

Session Highlights

This session presented new insights into the mechanisms underlying cardiac arrhythmias in TANGO2 deficiency disorder. TANGO2 loss disrupts long-chain fatty acid oxidation, leading to ATP depletion and impaired calcium handling during metabolic stress. These changes activate TRPM4 channels, causing abnormal electrical activity and arrhythmias. The findings identify TRPM4 as a promising therapeutic target and provide a mechanistic framework for how current and emerging treatments, including vitamin B-complex, verapamil, and TRPM4 inhibition, may mitigate cardiac complications.

Mini scientific Symposium Sessions

Sunday, June 28th, 2026

Understanding TDD via Genotype-Phenotype Correlations in 22q11.2 Deletion Syndrome

Donna McDonald-McGinn, MS, CGC & Beata Nowakowska, PhD

Donna McDonald-McGinn, MS, LCGC, is the Director of the 22q Center and Chief of the Genetic Counseling Section at Children's Hospital of Philadelphia, and a Professor of Clinical Pediatrics at the University of Pennsylvania. She leads global initiatives as Chair of the 22q11.2 Society and a Founding Board member of the International 22q11.2 Foundation. With over 380 original articles and a textbook, she specializes in chromosome 22q11.2 deletions and variants, including patients with concurrent TANGO2 Deficiency Disorder. She also serves on T2RF's Scientific Advisory Board.

Beata Nowakowska, PhD, is researcher in the Department of Medical Genetics at the Institute of Mother and Child in Warsaw, Poland. She has also served as a Trustee of the 22q11.2 Society and 2024 Program Chair. Dr. Nowakowska has published research exploring pre- and post-natal diagnosis of TANGO2 Deficiency Disorder and genotype-phenotype correlations with TDD and 22q11.2 Deletion Syndrome. Her research aims to uncover how genetic links could offer insights into the biology of TDD, bringing us closer to better understanding, earlier diagnosis, and smarter treatment strategies.

Session Highlights

22q11.2 deletion syndrome (22q11.2DS) affects approximately 1 in 2,000 births and is a leading cause of congenital heart disease, cleft palate, developmental delay, and schizophrenia. Disease severity varies with deletion size and can be modified by pathogenic variants on the remaining chromosome, unmasking recessive disorders such as TDD. In a cohort of over 1,700 individuals with 22q11.2DS, TDD was identified in approximately 1 in 160 patients. Because TDD is treatable, newborn screening for both conditions could enable earlier diagnosis and improve outcomes.

The Future of TDD Gene Therapy - AAV Development

Vandana Gupta, PhD

Vandana Gupta, PhD works on neuromuscular disorders, with a particular focus on the molecular mechanisms underlying genetic muscle diseases, gene discovery, and therapeutic development.

Her research integrates genetics, developmental biology, and translational approaches to uncover disease mechanisms & develop targeted therapies for rare neuromuscular disorders. She currently serves on T2RF's SAB.

Session Highlights

This presentation explained that because TDD is caused by loss of function variants in a single gene, it is a strong candidate for gene replacement therapy. An AAV mediated gene therapy is currently being designed to restore TANGO2 expression through systemic delivery. Supporting evidence from a related mouse model demonstrates that AAV gene replacement improved survival, muscle growth, motor function, and tissue distribution without causing abnormal pathology in other organs. In parallel, the laboratory is screening nearly 1,000 FDA approved mitochondrial targeted compounds in zebrafish models exposed to fasting and exertion, which mimic clinical triggers of TDD. Several promising compounds have already been identified for additional testing to determine whether they can prevent or reverse disease onset. Both gene therapy and small molecule approaches show promise, and future treatment strategies may need to be tailored to disease severity, affected tissues, and individual patients, with combination therapies potentially offering the greatest benefit.

Higher-ordered Organization of TANGO2 May Be a Prerequisite for Its Function

Michael Sacher, PhD

Michael Sacher, PhD, studied biochemistry at McGill University and completed postdoctoral research at Yale University, where he discovered the TRAPP complexes involved in membrane trafficking. At Concordia University, his independent laboratory linked TRAPP mutations to rare diseases. Recently, his lab has focused on TANGO2 function and TANGO2 Deficiency Disorder, discovering that vitamin B5 has beneficial effects in fruit fly models and human cells. His team continues to investigate TANGO2 function to identify further treatments for TDD.

Session Highlights

TANGO2 functions as a metabolic adaptation factor linking peroxisomal and mitochondrial lipid metabolism to maintain energy homeostasis during metabolic stress. Under conditions requiring metabolic flexibility, such as fasting, exercise, or illness, TANGO2 binds long-chain acyl-CoAs, promotes metabolite-dependent oligomerization, and supports efficient fatty acid oxidation. TANGO2 deficiency disrupts lipid utilization and alters lipid metabolic pathways, contributing to energy failure, fasting-induced metabolic crises, rhabdomyolysis, and cardiac arrhythmias. Although TANGO2 localizes differently in yeast and mammals, its conserved function is demonstrated by rescue of yeast lipid utilization defects with human TANGO2. High-dose vitamin B5 may compensate by expanding CoA-dependent metabolic capacity, while vitamin B9 may reduce arrhythmia risk by supporting mitochondrial function and limiting oxidative stress. These findings identify TANGO2 as a key regulator of lipid metabolism and metabolic resilience.

Plenary Sessions

Monday, June 29th, 2026

Opening Keynote
Pat Furlong, BSN, MS

Patricia (Pat) Furlong, BSN, MS is the Founding President of Parent Project Muscular Dystrophy (PPMD), the leading U.S. nonprofit dedicated to ending Duchenne muscular dystrophy. Following the diagnosis of her two sons with Duchenne in 1984, Pat transformed personal tragedy into a lifelong mission to accelerate research, improve care, and advance treatments for individuals living with rare diseases.

Session Highlights

Founded in 1994 to address critical gaps in care and research, Parent Project Muscular Dystrophy (PPMD) has transformed the Duchenne muscular dystrophy landscape through coordinated patient advocacy, research investment, and policy initiatives. By establishing patient registries, advancing standards of care, creating certified care centers, and securing more than $650 million in federal research funding, PPMD helped expand a therapeutic pipeline of over 30 treatments, including five FDA-approved therapies. Their experience demonstrates how sustained collaboration among patients, clinicians, researchers, and policymakers can accelerate progress in rare diseases, a model that offers valuable insights for advancing TANGO2 research, care, and treatment development.

From Observation to Impact: TANGO2 Natural History Study Updates - *Developing Acute Care Clinical Guidelines*
Samuel Mackenzie, MD, PhD & Jonathan Scaccia, PhD

Samuel Mackenzie, MD, PhD is an assistant professor at the University of Rochester where he focuses on developing gene-targeted therapeutics for pediatric neuromuscular conditions. He has been a long-standing advocate for the TANGO2 community and currently serves as chair of the T2RF Scientific Advisory Board (SAB) and as a member of the Board of Directors.

Jonathan Scaccia, PhD, is a community psychologist and implementation scientist specializing in systems change, health equity, and AI-enhanced dissemination. He is the founder of PubTrawlr and This Week in Public Health, where he develops innovative tools that translate complex research into accessible, actionable insights for practitioners and communities. His work focuses on bridging research, practice, and policy to improve health outcomes and strengthen community-driven systems.

Session Highlights

This presentation highlighted the TANGO2 community's strong readiness to partner in research, with families prioritizing studies on biomarkers, TANGO2 biology, mitochondrial function, and improved management of neurological and metabolic symptoms. It also described the development of the first consensus-based TANGO2 clinical care guidelines using a multidisciplinary Modified Delphi process. The guidelines provide standardized recommendations for managing acute metabolic crises, including rapid molecular testing, early dextrose-containing IV fluids, modified arrhythmia management, and routine vitamin B5 and B9 supplementation, demonstrating how community engagement and expert consensus can accelerate improvements in TANGO2 care.

From Observation to Impact: TANGO2 Natural History Study Updates - *Nutrition Matters: Supporting Health Day to Day*
Mousumi Bose, PhD

Mousumi Bose, PhD is the mother of Ilan Betzer (2010-2011), who was born with Zellweger spectrum disorder, a rare genetic disorder of peroxisome biogenesis. As a result of her personal experience with her son, Dr. Bose re-focused her career goals to study rare disease. Over the last decade, Dr. Bose has conducted research and published multiple papers on the role of the family caregiver in the management of rare pediatric disorders. Currently, Dr. Bose holds a faculty position in the Department of Nutrition and Food Studies at Montclair State University in New Jersey, studying quality of life, patient-focused drug development, dietary assessments, and health equity issues in rare disease communities. Dr. Bose is passionate about learning what is important to rare disease families and using that information to improve their lives.

Session Highlights

This presentation investigated the relationship between caregiver-reported vitamin B5 and B9 intake and TANGO2 Deficiency Disorder symptom frequency and severity. The cross-sectional study evaluated 20 participants using 24-hour dietary recall interviews and symptom surveys. Results showed mean daily intakes of approximately 380 mg for vitamin B5 and 8,400 µg for vitamin B9, with supplements contributing most of the overall intake. Higher B5 intake was associated with decreased overstimulation severity and improved socio-emotional scores on the Developmental Profile 4 assessment. Higher B9 intake was associated with decreased severity of ataxia and exotropia. The study identified predicted daily dose ranges of 155 to 250 mg for B5 and 2 to 70 mg for B9 to guide clinical trial design. Despite limitations such as a small sample size and caregiver-reported data, this research provides a foundation for future dose-response trials and B-vitamin guidelines.

From Observation to Impact: TANGO2 Natural History Study Updates - *Looking Ahead: What Long-Term Follow-Up Tells Us*

Christina Miyake, MD, MS, MPH

Christina Miyake, MD, MS, MPH is a pediatric cardiologist and Director of the Cardiovascular Genetics Arrhythmia Program at Baylor College of Medicine. She is a member of the T2RF Scientific Advisory Board and lead investigator of the TANGO2 Natural History Study. She discovered the role of B vitamins & TANGO2 deficiency disorder. Dr. Miyake's ultimate goal is to improve TDD management and outcomes while working toward effective treatments and a cure.

Session Highlights

This presentation summarized findings from a natural history study of TANGO2 Deficiency Disorder. Interviews, questionnaires, medical records, and targeted testing are all part of the data collected. It describes common early signs such as motor, speech, developmental, cognitive, and communication delays, as well as seizures, movement disorders, and TANGO2 "spells", which are often triggered by illness, fasting, heat, or activity. The study reports that most participants experience spells, while 68% have had at least one metabolic crisis. Observational data suggest that daily B5 (pantothenic acid) and B9 (folic acid) may help prevent metabolic crises, though it's effect on other symptoms remain unknown. Also discussed were routine recommendations, emergency guidelines for a metabolic crises, and how to monitor and support patients with TANGO2 deficiency disorder.

You Spoke, We Listened: Community Priorities and a Transparent Look at Rare Disease Research Funding

Deena Chisholm, MPH, CHES & Courtney Clyatt, MPH

Deena Chisholm, MPH, CHES serves as the T2RF's Research Director. Deena plays a pivotal role in developing and implementing the foundation's strategic plans for research and grant activities, ensuring alignment with the Foundation's objectives. She also builds and maintains relationships with a diverse array of stakeholders, such as researchers, clinicians, patient families, and the broader research community. Deena oversees the continuous evaluation and refinement of the foundation's research operations ensuring that the Foundation consistently operates at the forefront of the field.

Courtney Clyatt, MPH is a Senior Program Officer for Engagement at the Patient-Centered Outcomes Research Institute (PCORI). She came to PCORI with more than 10 years of experience in public health and project management. In her position, she has played a vital role in the Engagement Awards program and, specifically, the Pipeline to Proposal Awards, which funded community-building and engagement projects. Currently, her work has been focused on helping smaller organizations to build capacity to engage in patient-centered comparative effectiveness research.

Session Highlights

This joint presentation introduced the principles of patient-centered comparative clinical effectiveness research (CER), including PCORI's focus on addressing questions that matter most to patients, families, and caregivers. The PICOTS framework was introduced as a tool to guide the conference PCOR workshop and support the development of meaningful, patient-centered research questions. Findings from the TANGO2 Research Landscape & Priority Survey, which collected 179 global responses across five languages, identified biomarkers, TANGO2 biology, mitochondrial function, and neurological outcomes as top community priorities. While families expressed strong interest in partnering in research, they emphasized the need for flexible participation options, clear communication, and meaningful engagement. These efforts reinforce a shift from conducting research on families to conducting research with families as essential partners in advancing TANGO2 research.

Conference Workshops

This conference was partially funded through a Patient-Centered Outcomes Research Institute (PCORI) Eugene Washington Engagement Award (EASCS-41293). Funding from this award will support the development of a TDD globally connected, patient-centered, research-engaged community of families, researchers, clinicians, and stakeholders actively involved throughout the research process, from concept development to dissemination.

The TANGO2 Research Foundation remains committed to supporting TANGO2 families, raising awareness, promoting earlier diagnosis, and advancing innovative TDD research.

We extend our sincere gratitude to the T2RF PCOR Convening Advisory Board (CAB) members for generously contributing their time, expertise, and leadership to this initiative.


Kasha Morris

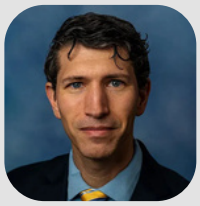

Debbie DeLoach


Amanda Hull, PhD


Seema Lalani, MD


Mathew Edick, PhD


Lina Gonzalez, MD


Joshua Meisner, MD, PhD


Chaya Murali, MD


Jon Scaccia, PhD


Alison Bell, MPH


Sam Mackenzie, MD, PhD


Sarah Sandkuhler, MD, PhD

Monday, June 29th, 2026 and Tuesday, June 30th, 2026

TDD PCOR WORKSHOPS 1 & 2 - *Researching the core: Unveiling What the Community Values Most*

Session Highlights

As part of the TANGO2 Research Foundation's PCORI Eugene Washington Engagement Award, conference participants took part in interactive workshops designed to strengthen capacity for patient-centered comparative effectiveness research (CER). Families, researchers, clinicians, and other stakeholders worked collaboratively to apply the PICOT framework (Population, Intervention, Comparison, Outcome, and Time) to develop research questions addressing the community's highest-priority research needs. The workshops emphasized identifying outcomes that matter most to individuals living with TANGO2 deficiency disorder and their families, while ensuring future research reflects real-world priorities and patient-centered principles.

Across all workshop groups, five overarching themes consistently emerged as the community's highest priorities for future research: optimizing vitamin treatment, measuring meaningful patient-centered outcomes, achieving earlier diagnosis, improving quality of life, and advancing the understanding of TANGO2 disease biology. These priorities and PICOT examples will help inform the Foundation's future research agenda and contribute to the development of a TDD patient-centered comparative effectiveness research framework and guidebook.

Scan to view
The TDD Research Toolbox

Scan to join the discussions on
The Research Learning Network

Plenary Sessions

Monday, June 29th, 2026

Elizabeth Long, PhD (Moderator)

Dylan Simon, MS

Ron Bartek, MA

Julie Tierney, JD

Bridging the Gap Between TDD Community Needs, Research and Regulatory Agendas

Elizabeth Long, PhD is Director of Research and Evaluation of the Evidence-to-Impact Collaborative's Research Translation Platform where she oversees the evaluation and research activities of research-policy bridging models. These include a science communication model (the SciComm Optimizer for Policy Engagement) and a partnership model (the Research-to-Policy Collaboration model). Her training background is in the etiology and prevention of substance use disorders, which led to her aspirations of informing large-scale impact through evidence-based policies.

Dylan Simon currently serves as the Senior Director of Policy for the EveryLife Foundation for Rare Diseases. Since joining the EveryLife Foundation, he has focused on newborn screening and diagnostic policy issues, as well as annual appropriations efforts for the rare disease community. Dylan's work includes leadership with the community on the passage of landmark state legislation providing for expedient implementation of RUSP conditions. Prior to joining EveryLife, Dylan interned for the Senate HELP Committee and worked at advocacy organizations.

Ron Bartek is Co-Founder & President of FARA, a leading rare disease advocacy organization focused on treatments and cures for Friedreich's ataxia. He's widely recognized as a national leader in rare disease advocacy, patient engagement, and research acceleration. Ron currently serves on the Board of Directors of Critical Path Institute and previously served as Director and Chair of the (NORD). He's held numerous leadership and advisory roles across the rare disease ecosystem, including service on the NIH/NCATS National Advisory Council & NIH Neurological Institute National Advisory Council. In recognition of his longstanding impact on the rare disease community, he was named one of the FDA Office of Orphan Products Development's "30 Heroes Changing the Lives of Rare Disease Patients."

Julie Tierney, JD is a Principal at Leavitt Partners in Washington, D.C., where she advises stakeholders across the life sciences ecosystem, including companies, researchers, and patient organizations, on U.S Food and Drug Administration (FDA) regulatory strategy and policy. Before joining Leavitt Partners, she held a number of senior roles during an almost two decade tenure at the FDA, including FDA Chief of Staff, and Deputy Director of FDA's Center for Biologics Evaluation and Research. While at FDA, Julie advanced cross cutting policy and regulatory initiatives to promote innovation for patients with rare and serious diseases, with a strong focus on cell and gene therapies. She is committed to patient focused regulatory science and partnering with advocacy organizations to help translate scientific advances into meaningful therapies. Julie received her Bachelor of Arts from Johns Hopkins University with a double major in History and Biology and her Juris Doctor from Georgetown University Law Center.

Session Highlights

Supported by the EveryLife Foundation for Rare Diseases, this session highlighted the critical role of patient advocacy in advancing rare disease research, policy, and therapeutic development. Dylan Simon introduced frameworks for effective policymaker engagement, including the Research-to-Policy Collaboration and SCOPE models, emphasizing that patients and families are essential partners across the rare disease ecosystem. He described how lived experience informs research priorities, clinical trial design, regulatory decision-making, and patient-focused drug development initiatives. Ron Bartek shared the Friedreich's Ataxia Research Alliance (FARA) as a model for building strategic partnerships that transformed a limited research landscape into a robust therapeutic pipeline through sustained collaboration among families, researchers, government, industry, and advocacy organizations. Julie Tierney concluded the session by providing perspectives on advocacy within the regulatory environment and strategies for engaging effectively with the FDA.

The session underscored that meaningful collaboration among patients, families, researchers, clinicians, industry, policymakers, and regulators is essential to accelerating therapeutic development and expanding access to treatments for individuals living with rare diseases. Attendees were also introduced to advocacy resources developed by the EveryLife Foundation to support continued learning and empower families to become effective advocates. As a next step, the TANGO2 community will build on these efforts by engaging the U.S. Food and Drug Administration (FDA) in a patient listening session to ensure the lived experiences, priorities, and treatment needs of individuals with TANGO2 Deficiency Disorder help inform future drug development and regulatory decision-making.

Panel Sessions

Monday, June 29th, 2026

Erin Kaiser, MA, CCC-SLP

Amy Clay

Kasha Morris

Laura Moore

Giving Voice to TANGO2: Speech Challenges, Strategies, and Hope

Erin Kaiser, MA, CCC-SLP has two decades of experience in school-based speech-language pathology and specializes in treating complex disabilities, including autism, intellectual disabilities, and other genetic disorders. Her clinical focus centers on implementing multimodal communication strategies, including comprehensive AAC evaluations, as well as staff and parent training. Driven by a personal connection to the TANGO2 community formed early in her career, Erin is a fierce advocate for neurodiverse learners in Colchester, CT, where she works to educate families and community members on multimodal communication and acceptance for all forms of communication. Every child deserves a voice!

Amy Clay resides in Rochester, NY, with her husband Stan, daughter Katie, and dog Cody. She serves as a T2RF Regional Coordinator and actively participates in the Outreach and Fundraising Committees. Professionally, she is an IT Director for the Catholic Health System and also serves as a board member and Treasurer for the Rochester Special Hockey Icecats. In her spare time, she supports her daughter's DJ endeavors as a chauffeur and groupie for DJ IceCat.

Kasha Morris is the mother of Ryan, a TANGO2 warrior, and Co-Founder and Secretary of the TANGO2 Research Foundation. She serves on the Foundation's Research Committee and plays a leading role in family engagement and community support initiatives. Through her advocacy, Kasha works to bridge the gap between scientific research and the everyday experiences of families affected by TANGO2 Deficiency Disorder, helping to advance awareness, collaboration, and hope for the global TDD community.

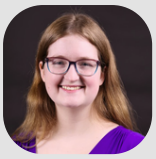
Laura Moore wears many hats, as many TANGO2 parents do. She is the mother of three children, including her youngest daughter, Ellie, who was diagnosed with TANGO2 Deficiency Disorder in 2017. A dedicated homeschool parent and caregiver, Laura balances her family's needs with her role as Financial Director for the City of Tahoka, Texas. Laura holds a Bachelor of Science in Accounting Control Systems and a Bachelor of Arts in Interior Design from the University of North Texas. She and her husband, Blake, have been married for 20 years and live in Littlefield, Texas, with their three children: Denton, Reese, and Ellie.

Session Highlights

This panel combined clinical expertise with lived experience to highlight communication challenges and successes across the TANGO2 community. Speech-language pathologist Erin reviewed common communication impairments associated with TANGO2 deficiency disorder, including speech delay, dysarthria, and cognitive impairment, and discussed individualized interventions such as augmentative and alternative communication (AAC) systems and voice preservation technologies. Families shared their personal journeys, including Katie's successful treatment for velopharyngeal insufficiency (VPI), which significantly improved her speech and confidence, and Ryan's notable gains in speech clarity following optimized vitamin B therapy. Laura described Ellie's communication journey, demonstrating how early adoption of high-tech AAC, consistent caregiver modeling, and community support fostered increasing independence and communication skills. Collectively, the presentations underscored the importance of individualized care, emerging technologies, and family-centered approaches in empowering individuals with TANGO2 deficiency disorder to communicate effectively and participate fully in everyday life.

Introduction to AAC

AAC Manufacturers & Support Organizations



Small Creatures, Big Questions: Studying TANGO2 Deficiency Disorder Mechanism and Therapeutics Using C. elegans

Sarah Sandkuhler, MD/PhD Cand.

Sarah Sandkuhler, MD/PhD candidate is a student in the Pathology department at the University of Rochester School of Medicine and Dentistry. She is working on her thesis in the lab of SamuelMackenzie, studying TANGO2 Deficiency Disorder in a C. elegans (worm) model. Her clinical interests include eurogenetics and the development of improved gene replacement therapeutic strategies. She currently serves on the T2RF Early Detection and Diagnostic Committee and was a 2024 T2RF Emerging Researcher Award recipient. When not in the lab, she can be found swing dancing, singing in choir, and spending time with her cat, Beatrice.

Session Highlights

This presentation highlighted the use of the nematode C. elegans to investigate disease mechanisms and identify therapeutic candidates for TANGO2 Deficiency Disorder. Researchers generated worms lacking both TANGO2 homologs and screened 958 mitochondria-targeted compounds, identifying 88 candidates for further evaluation of survival, movement, reproduction, and other disease-related phenotypes, with cross-species validation underway. Additional studies found no evidence that TANGO2 functions as a heme transport protein, suggesting previously observed heme abnormalities are secondary rather than causative. Unbiased metabolomic analyses are also underway to identify metabolic pathways disrupted by TANGO2 deficiency. Together, these complementary approaches are advancing our understanding of TANGO2 biology while accelerating therapeutic discovery. Together, these studies provide new insights into TANGO2 biology while prioritizing promising compounds and pathways for future therapeutic development.

Small Molecule Screening for Therapeutic Development in TDD

Vandana Gupta, PhD

Dr. Gupta works on neuromuscular disorders, with a particular focus on the molecular mechanisms underlying genetic muscle diseases, gene discovery, & therapeutic development. Her research integratesgenetics, developmental biology, and translational approaches to uncover disease mechanisms and develop targeted therapies for rare neuromuscular disorders. She currently serves on T2RF's SAB.

Session Highlights

This session presented emerging therapeutic approaches for TANGO2 deficiency disorder (TDD), a multisystem condition characterized by developmental delays, rhabdomyolysis, and cardiac arrhythmias triggered by metabolic stress. Using a mouse model, systemic AAV9 gene therapy restored muscle growth, body mass, and motor function for up to eight months without observed toxicity. Complementary high-throughput screening of 998 FDA-approved mitochondrial compounds in a tango2 mutant zebrafish model identified candidate drugs that rescued stress-induced disease features. Together, these studies highlight the potential of combining gene replacement and targeted small molecule therapies to develop effective treatments for TDD.

Cardiac Findings in TANGO2 Deficiency Disorder

Agustin Lujan, MD, PhD

Agustín Luján, MD, PhD is a physician-scientist dedicated to advancing research in pediatric rare diseases, including conditions like TANGO2 deficiency. He holds a PhD in biological sciences and postdoctoral training in cell and molecular biology. At Hospital Sant Joan de Déu, he works as a researcher in the neuromuscular and arrhythmia/sudden death units, serving as a bridge between basic and clinical research in this field. Agu currently serves as a T2RF Research Committee member.

Session Highlights

This presentation summarized cardiac monitoring and clinical observations in individuals with TANGO2 Deficiency Disorder, highlighting the importance of ongoing surveillance for life-threatening arrhythmias. Among 12 patients evaluated, cardiac findings included prolonged QT intervals, Brugada-pattern electrocardiograms, bradycardia, and severe sinus pauses requiring pacemaker implantation in one individual. Since the introduction of vitamin B-complex therapy, no cardiac arrhythmias have been observed in this cohort. These findings underscore the value of individualized cardiac monitoring to guide timely intervention and support continued efforts to expand international clinical collaboration, provider education, and standardized cardiac care for individuals with TANGO2 Deficiency Disorder.

Plenary Sessions

Tuesday, June 30th, 2026

Elucidating the Cardiac Channelopathy in TANGO2 Deficiency Disorder

Weiyi Xu, PhD

Weiyi Xu, PhD 2024 T2RF Emerging Researcher awardee is an instructor in the Molecular and Human Genetics Department at Baylor College of Medicine (BCM). He did his PhD in electrophysiology at University of Hong Kong, and then received postdoctoral training specialized in cardiology and genetics in BCM. He is pursing an academic career path in genetic cardiac arrhythmias, and enjoys conducting translational research where findings in the laboratory can have an immediate impact on patient care in the clinic. Outside the lab, he enjoys playing basketball and hanging out with his 2-year-old son.

Session Highlights

This presentation explained research into the cardiac complications of TANGO2 Deficiency Disorder (TDD), focusing on why patients develop life-threatening heart rhythm abnormalities during metabolic crises. Dr. Xu discussed that the heart's electrical activity depends on ion channels and how disruptions can cause long QT syndrome and dangerous arrhythmias. Dr. Xu, also describes using a "heart-in-a-dish" model of TDD to study these mechanisms. The research shows that vitamin B9 (folate) can stop arrhythmias in TDD cells, although it does not correct the underlying prolonged QT interval. The findings suggest that arrhythmias result from both a vulnerable electrical substrate (reduced Kv7.1 ion channel activity causing long QT) and a separate trigger, potentially involving overactive KATP channels. The presentation concludes by highlighting ongoing efforts to identify targeted treatments, acknowledging collaborators, funding sources, and the TANGO2 patient community whose experiences motivate the research.

Cerebellar Contributions to Movement Challenges in TANGO2 Deficiency Disorder

Lauren Peace, PhD Cand.

Lauren Peacoe, BS, PhD Candidate is a Neuroscience PhD student at Baylor College of Medicine in Houston, Texas. Growing up alongside two brothers with a rare disorder, Lauren has always been passionate about understanding how genetic variations can disrupt brain development and activity. She conducts her thesis work in the Sillitoe Lab, where discoveries continue to establish the cerebellum, a brain region that coordinates movement and balance, as an origin of diseases like dystonia, ataxia, and tremor. Using a mouse model, Lauren studies how loss of TANGO2 alters cerebellar structure and function to influence movement, with the goal of identifying ways to improve the lives of those with TDD. Beyond her research, Lauren enjoys sharing her love of science through teaching, and she volunteers with children and families affected by special needs in her community.

Session Highlights

This presentation explored the neurological basis of movement disorders in TANGO2 Deficiency Disorder, focusing on the role of cerebellar dysfunction. Because many individuals with TDD experience multiple movement disorders, this study investigates whether abnormalities within the cerebellum contribute to these symptoms. Using a mouse model that reproduces the genetic loss of TANGO2, researchers observed movement abnormalities similar to those seen in patients. Electrophysiological recordings from awake mice demonstrated that Purkinje cells, the principal computational cells of the cerebellum, exhibited significantly more irregular complex spike firing compared with controls. These findings suggest that altered cerebellar signaling may contribute directly to the motor symptoms observed in TDD. Future work aims to determine how different patterns of cerebellar activity relate to specific movement disorders and how the brain encodes the complex neurological features of the disease.

TANGO2 PubNav: Building Exploration, Understanding and Community Around TDD Published Literature

Mike Morris

Mike Morris is dad to TANGO2 warrior Ryan and Co-founder of the TANGO2 Research Foundation. His background is in technology and entrepreneurship. Mike currently serves as the T2RF President and is a member of the Board of Directors and Research Committee.

Session Highlights

This presentation introduced PubNav, an AI-powered platform that centralizes all published TANGO2 Deficiency Disorder literature in a single, accessible repository for families, clinicians, and researchers. Each publication includes AI-generated plain-language summaries, podcasts, videos, slide decks, and infographics to improve accessibility. An integrated AI chat feature enables users to explore individual studies, compare findings, identify research trends, and ask questions across the entire body of TANGO2 literature. The platform also supports community discussion by allowing families, clinicians, and researchers to discuss publications directly alongside each article. The presentation concluded with a live demonstration showcasing the platform's features and functionality.

Plenary Sessions

Tuesday, June 30th, 2026

Behind Every TDD Warrior Is a Caregiver, Rare and Resilient: Tools & Strategies to Cultivate Hope, Community, & Well-Being

Julie Wells & Kasha Morris

Julie Wells has spent over 32 years in the non-profit sector working on community-based issues, centering marginalized populations and access to opportunities as the focal point of her impact. In 2024, she joined Give an Hour, a national mental health nonprofit, as Director of Strategic Relationships. In this role, she travels nationwide providing training and support to diverse communities, including individuals affected by rare diseases. She also has a personal connection to the rare disease community through her own experience with genetic heart failure, which similarly affects her sister, nieces, and nephew.

Kasha Morris is the mother of Ryan, a TANGO2 warrior, and Co-Founder and Secretary of the TANGO2 Research Foundation. She serves on the Foundation's Research Committee and plays a leading role in family engagement and community support initiatives. Through her advocacy, Kasha works to bridge the gap between scientific research and the everyday experiences of families affected by TANGO2 Deficiency Disorder, helping to advance awareness, collaboration, and hope for the global TDD community.

Session Highlights

This presentation explored the mental health challenges experienced by individuals and families affected by TANGO2 Deficiency Disorder, emphasizing how rare disease profoundly shapes well-being through medical uncertainty, complex care, prolonged advocacy, trauma, chronic stress, grief, and social isolation. Speakers highlighted that emotional needs evolve across the diagnostic and caregiving journey and that fostering psychological safety, connection, and hope is essential to long-term resilience.

The presentation also emphasized practical strategies to promote well-being, including self-compassion, emotional regulation, nervous system regulation, self-care, and peer connection. Recognizing an unmet need for rare disease-specific mental health resources, the TANGO2 Research Foundation partnered with Give an Hour to develop the TDD Mental Health Toolkit, providing individuals and caregivers with practical, evidence-informed resources to support emotional well-being throughout the disease journey.

To view TANGO2 Research Foundation resources, scan the QR codes below:

TDD Mental Health Toolkit

Family Resources

YouTube Channel

Panel Sessions

Tuesday, June 30th, 2026

Kuntal Sen, MD

Kimberly Houck, MD

Robin Mitchell, MHSA

Understanding the TANGO2 Brain: Development, Injury, & Lived Experience

Kuntal Sen, MD is the co-director of the Neurogenetics Clinic in the Division of Neurogenetics & Neurodevelopmental Pediatrics at Children's National Hospital. He is one of the few physicians in the country with dual training in neurology and clinical genetics which affords him a unique expertise to diagnose and manage patients with complex monogenic neurological disorders. Dr. Sen's clinical expertise is centered on providing long-term, holistic, & family-centered care for patients with mitochondrial diseases and other ultra-rare neuro-metabolic disorders. Dr. Sen's research focus is on multi-modal neuroimaging and neuromonitoring in inborn errors of metabolism.

Kimberly Houck, MD is a Pediatric Neurologist at Texas Children's Hospital and Assistant Professor in the Department of Pediatrics, Division of Neurology and Developmental Neuroscience at Baylor College of Medicine (BCM) in Houston. Her clinical practice focuses on medical and surgical management of refractory epilepsy, with special interest in genetic and structural causes. She is a member of the multidisciplinary TANGO2 research team at BCM and has contributed to natural history study publications, review articles, and practice guidelines.

Robin Mitchell, MHSA is mom to Hanna (TANGO2 warrior) and Benjamin (TANGO2 sibling) and a 2023 retiree from a large health insurance company in Michigan. Robin's currently a volunteer with the TANGO2 Research Foundation, serving as the chair of the Outreach Committee and the Plymouth Canton Literacy Council.

Session Highlights

Dr. Sen reviewed the neurodevelopmental features and future neuroscience research priorities for TANGO2 Deficiency Disorder. Data from the natural history study indicate that while early infancy is often typical, developmental delays commonly emerge around 12 months, with metabolic crises contributing to developmental regression and persistent cognitive, speech, language, motor, and gait impairments. He emphasized the need for multidisciplinary care and outlined future research focused on neurobehavioral characterization, TANGO2 spells, advanced neuroimaging, and structured transition to adult care.

Dr. Houck expanded on the neurological manifestations of TANGO2 Deficiency Disorder, highlighting the marked clinical variability in seizure presentation and management. She reviewed current treatment approaches, emphasized that the ketogenic diet is contraindicated, and underscored the importance of preventing metabolic crises through trigger management, adequate sleep, and vitamin B5 and B9 supplementation to improve neurological and cardiac outcomes.

Complementing these clinical presentations, Robin Mitchell shared her daughter Hanna's journey living with TANGO2 Deficiency Disorder, offering a caregiver's perspective on the lifelong neurological, developmental, and caregiving challenges faced by affected families. Robin described Hanna's remarkable clinical stability since initiating a vitamin B-complex regimen, noting that she has experienced no metabolic crises or seizures during this period. Her family's experience reinforced the potential impact of proactive clinical management while underscoring the importance of multidisciplinary care, thoughtful transition planning, and continued family support to improve long-term outcomes and quality of life for individuals with TANGO2 Deficiency Disorder.

Panel Sessions

Tuesday, June 30th, 2026

Chaya Murali, MD (Moderator)

Amanda Hull, PhD

Cheyenne Beach, MD

Tyson Swetek

Veronica Swetek

Advocating for Your Warrior: A Panel Perspective

Chaya Murali, MD is a pediatric geneticist at Texas Children's Hospital in Houston, Texas. Her research is focused on quality of life and lived experiences in people with genetic diagnoses and their families. Dr. Murali spearheaded a study of quality of life in the TDD community in 2019, and she currently serves as part of the TDD PCOR stakeholders team and Research Committee. In addition to her work as a physician, Dr. Murali is a published personal essayist, and some of her writing can be viewed at www.chayanautiyalmurali.com

Amanda Hull, PhD (DPsych Ed) is from Suffolk in England. She and husband, Daniel, have three boys; Joe, Sebby and Walter. Sebby is a 15-year-old diagnosed with TANGO2 Deficiency Disorder. Amanda has worked as an Educational Psychologist for the past 20 years, and Daniel is a farmer. They live on a farm and have an active lifestyle with all of them enjoying being outdoors and doing adventurous activities.

Cheyenne Beach, MD directs the Pediatric Electrophysiology program at Yale New Haven Children's Hospital. In addition to seeing patients in both inpatient and outpatient settings, she performs procedures to restore more normal heart rhythms for children with arrhythmias and adults with congenital heart disease. She cares for patients with TDD and other rare diseases and wants to help patients, families, and other providers in their rare disease journeys. Cheyenne currently serves as a T2RF SAB member.

Tyson Swetek is dad to TANGO2 warrior Thea. He currently serves as a member of the T2RF Research Committee. His family served as the 2025 Shine a Light on TANGO2 Ambassador Family, helping elevate awareness with their story.

Veronica Jones-Swetek is a first grade teacher with over 20 years of experience in education. She is also the mother of an 11-year-old daughter who was diagnosed with TANGO2 Deficiency Disorder at age two. Through the TANGO2 Research Foundation, Veronica has served as a former board member and currently helps lead the family community as an administrator of the foundation's Facebook group. She is also an active member of the Outreach committee and a Regional Coordinator where she works to connect and support families navigating a TANGO2 diagnosis.

Session Highlights

This panel presentation focused on advocacy, parent empowerment, and emergency preparedness for individuals with TANGO2 Deficiency Disorder. Speakers emphasized strategies for advocating across healthcare, educational, and community settings, highlighting the importance of carrying a physician-endorsed TANGO2 Emergency Protocol Letter and utilizing the International 24/7 TANGO2 Clinical Consult Hotline during medical emergencies. Family speakers shared real-world examples of effective advocacy, demonstrating how persistence can overcome barriers to accessing necessary medical care and how emergency preparedness, including providing treatment guidelines and connecting local physicians with TANGO2 clinical experts, can facilitate timely, evidence-based care during a metabolic crisis.

The panel also highlighted resources available through the TANGO2 Research Foundation, including the Clinical Consult Hotline, Parent & Caregiver Guidebook, "Elevator Speech" cards, and educational materials on mental health and genetics. Additional resources from the Courageous Parents Network were presented to strengthen caregiver confidence, advocacy skills, and emotional support. The session concluded with Emily Perl Kingsley's Welcome to Holland, a reflection on the unexpected journey of raising a child with special needs and the resilience families develop along the way.

24/7 TANGO2 Clinical Consult Hotline: 1 (860) 598-0955

CONTINUING THE JOURNEY TOGETHER

Building Community Beyond the Conference

In addition to the scientific and educational program, the conference featured Family Experiences Through the Years, a breakout session highlighting the diverse experiences of families at different stages of the TANGO2 journey. Families shared their perspectives, challenges, milestones, and lessons learned, offering valuable support and insights to others navigating diagnosis, care, and life with TANGO2 Deficiency Disorder. The conference also offered a variety of optional community activities designed to foster connection, peer support, and celebration, including a pre-conference gathering for angel families, a community walk/run, informal coffee conversations for moms and dads, a creative gathering for siblings and friends, and other social events. Together, these opportunities created welcoming spaces for families to build relationships, exchange support, celebrate their community, and strengthen connections with one another.

Closing Remarks

The 2026 TANGO2 Family Conference reflected the strong momentum across research, clinical care, patient advocacy, and community engagement. It provided an important opportunity for families, clinicians, researchers, and industry partners to connect, share knowledge, and reinforce their shared commitment to improving the lives of individuals affected by TANGO2 Deficiency Disorder. The TANGO2 Research Foundation is deeply grateful to all presenters, attendees, volunteers, sponsors, and partners whose dedication and support made this meeting possible.

Looking ahead, the Foundation is actively building on this momentum through new grant submissions, the development of expanded research initiatives, and the launch of additional patient- and family-centered research opportunities designed to further engage the TANGO2 community as partners in discovery. These next steps will help accelerate scientific progress, deepen understanding of the disorder, and support the translation of research into meaningful clinical impact.

Conference presentation recordings are available on the TANGO2 Research Foundation YouTube Channel, providing continued access to the valuable educational content shared throughout the meeting. We also encourage you to stay connected by following the TANGO2 Research Foundation on social media for the latest research updates, educational resources, community events, and opportunities to get involved. We look forward to continuing this momentum and welcoming you to the next TANGO2 Family Conference.

We gratefully acknowledge the following sponsors for their support of the 2026 TANGO2 Family Conference

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WWW.TANGO2RESEARCH.ORG