



2025 TANGO2

RESEARCH SYMPOSIUM

Program

FRIDAY, OCTOBER 24TH 2025

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INTRODUCTION

Welcome to the 3rd Biannual TANGO2 Research Symposium

We are honored to bring together researchers, clinicians, families, caregivers, and advocates dedicated to advancing knowledge and strengthening connections within the TANGO2 community. Today's program, led by leading experts and supported by Foundation grants, features information-rich sessions highlighting the latest research findings, clinical insights, and lived experiences.

Through presentations and interactive discussions, we aim not only to share discoveries but also to foster collaboration and reinforce the global network committed to improving care and shaping brighter futures for those affected by TANGO2 deficiency disorder.

Thank you for joining us—and please share this invitation with others who may wish to be part of this important effort.

The 2025 Research Symposium Planning Committee,

Mike and Kasha Morris
Kara Cameron
Giorgio Pochettino
David Longman
Tyson Swetek

Joshua Meisner, MD, PhD
Lina Gonzalez, MD
Seema Lalani, MD
Agustin Lujan, MD, PhD

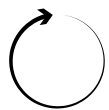
Deena Chisholm, MPH, CHES
Ann Geffen, MA, JD
Destiny Braden



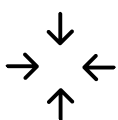
OUR MISSION

To lead the way in finding a cure for TANGO2 deficiency disorder.

OUR VISION



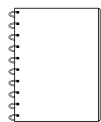
Accelerate research and therapeutic discoveries



Advocate and promote awareness



Support and connect families



Drive early and accurate diagnosis

OUR STAFF



Ann Geffen, MA, JD
Executive Director



Deena Chisholm, MPH, CHES
Director, Research Engagement



Destiny Braden
Development & Operations
Manager



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WORDLY TRANSLATION & CLOSED CAPTIONING

Vertaling en Geslote Ondertiteling | Përkhimi dhe Titrat e Mbyllur | الترجمة والترجمة المغلقة | অনুবাদ এবং বন্ধ
ক্যাপশনিং | Превод и Затворен Субтитър | 翻译和闭幕字幕 | 翻译和字幕 | 翻譯和封閉字幕 | Prijevod i
Zatvoreno Titlovanje | Překlad a uzavřené titulky | Oversættelse og Lukket Tekstning | Vertaling en Gesloten
Ondertiteling | Tölge ja Suletud Subtiitrid | Käännös ja Suljetut Tekstit | Traduction et Sous-titrage |
Traduction et Sous-titrage | Übersetzung und Untertitelung | Μετάφραση και Κλειστή Υποτιτλισμός |
અનુવાદ અને બંધ કેપ્શનિંગ | אנוואד וכיתובים סגורים | अनुवाद और बंद कैप्शनिंग | Fordítás és Zárt Felirat | Terjemahan
dan Teks Tertutup | Aistriúchán agus Foirmeacha Dúnta | Traduzione e Sottotitolazione | 翻訳とクローズド
キャプション | 번역 및 폐쇄 자막 | Tulkojums un Aizvērtie Subtitri | Vertimas ir Uždaryti Subtitrai | Terjemahan
dan Sari Tertutup | Terjemahan dan Sari Tertutup | Oversættelse og Lukkede Tekster | ترجمه و زیرنویس بسته
| Tłumaczenie i Napisy | Tradução e Legendagem | Tradução e Legendagem | અનુવાદ અਤੇ ઘંટ વૈપક્ષનિંગ |
Traducere și Subtitrare | Превод и Закрываете Субтитры | Превод и Затворени Титлови | Preklad a
uzavreté titulky | Prevod in Zaprti Podnapisi | Traducción y Subtitulación | Traducción y Subtitulación |
Tafsiri na Maandishi ya Sauti | Översättning och Slutna Texter | Pagsasalin at Saradong Subtitles |
மொழிபெயர்ப்பு மற்றும் மூடப்பட்ட கப்சனினிங் | παραλλαγή κλειστής υποτίτλων | Çeviri ve Kapalı Altyazı |
Переклад і Закриті субтитри | ترجمه اور بند کیپشننگ | Dịch và Phụ Đề | Cyfieithu a Chyfandir Cau



SCAN THE QR CODE

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



TDD Research Toolbox



Family Resources



YouTube Channel

Symposium Agenda

All times are listed in Eastern Standard Time (EST)

TIME	PRESENTATION	SPEAKER
9:00 AM	Founder's Welcome	Mike and Kasha Morris
9:05 AM	From Updates to Action: Building the TANGO2 Research Foundation Roadmap	Deena Chisholm, MPH, CHES
9:20 AM	From DNA to Day-to-Day: TANGO2 Patient Outcomes, A Natural History Study Update	Sergio Espinosa-Alvarez, MD
9:50 AM	Clinical Updates in TANGO2 Deficiency Disorder: Movement Disorders & Leanings From a Newly Diagnosed 61-Year-Old Patient	Samuel Mackenzie, MD, PhD
10:20 AM	Bread, Beer and Rare Disease: Using the Yeast TANGO2 Homolog Tng2p to Understand TDD	Michael Sacher, PhD
	Break	
11:00 AM	Cardiac Monitoring in TANGO2 Warriors	Georgia Sarquella-Brugada, MD, PhD
11:30 AM	TANGO2 and the Cerebellum: Linking Brain Circuits to Motor Disorders	Lauren Peacoe, BS, PhD (cand)
12:00 PM	High-throughput Drug Screening for TANGO2 Deficiency Disorder Using a Cross-Models Approach	Sarah Sandkuhler, MD, PhD (cand) & Vandana Gupta, PhD
12:30 PM	Genotype and Phenotype Patterns in TANGO2 Deficiency Disorder	Nikhita Gogate, MS, PhD (cand)
1:00 PM	T2RF Next Steps & Adjournment	Ann Geffen, JD, MA

SPEAKERS



Georgia Sarquella-Brugada, MD, PhD, is the Head of Arrhythmia, Inherited Cardiac Diseases and Sudden Death Unit, at the Hospital Sant Joan de Déu in Spain. She is also a Professor of Medicine in University of Barcelona and University of Girona in addition to the Director of Research in Arrhythmias and Inherited Cardiac Diseases, at the Institut de Recerca Sant Joan de Déu.



Vandana Gupta, PhD's laboratory at Brigham and Women's Hospital in Boston Massachusetts studies the genetics of neuromuscular diseases. Her research group has identified several new genes in neuromuscular diseases and developed animal models to test novel therapeutics and study disease pathophysiology. Dr. Gupta also currently serves as T2RF Scientific Advisory Board (SAB) member.



Samuel Mackenzie, MD, PhD, is a physician scientist at the University of Rochester Medical Center where he treats patients with genetic neuromuscular diseases and oversees a lab comprised of smart people who have decided to dedicate their days looking at microscopic worms. In his free time, he likes to think about TANGO2 Deficiency Disorder. Dr. Mackenzie currently serves as the chair of the T2RF's SAB.



Michael Sacher, PhD, received his terminal degree in biochemistry from McGill University followed by postdoctoral work in the Department of Cell Biology at Yale University. His work led to the discovery of the TRAPP complexes that are critical for moving proteins between specific cellular compartments. More recently, his laboratory became interested in TANGO2 and they established several model systems including fruit flies and human fibroblasts to aid in their work. These studies have led to the discovery that vitamin B5 alleviates cellular defects associated with non-functional TANGO2. Dr. Sacher currently serves as a member of the T2RF's Board of Directors & SAB.



Sarah Sandkuhler is an MD/PhD 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 Samuel Mackenzie, studying TANGO2 Deficiency Disorder in a *C. elegans* (worm) model. Her clinical interests include neurogenetics and the development of improved gene replacement therapeutic strategies. When not in the lab, she can be found swing dancing, singing in choir, and spending time with her cat, Beatrice.

SPEAKERS



Lauren Peacoe, is a Neuroscience PhD student at Baylor College of Medicine in Houston, Texas. She conducts her research in the Sillitoe Lab, where discoveries continue to establish the cerebellum as an origin of diseases including dystonia, tremor & ataxia. Motivated by a deep passion for understanding rare neurodevelopmental disorders, her current work uses a TANGO2 knockout mouse model to investigate how loss of TANGO2 alters cerebellar structure and function in TDD. By combining in vivo electrophysiology, behavioral analysis, and circuit-level interventions such as deep brain stimulation, Lauren aims to uncover the brain mechanisms underlying motor dysfunction in TDD, while also exploring therapeutic strategies that may improve the lives of affected individuals. Outside of the lab, Lauren engages in science education, and she volunteers with children and families affected by special needs in her community.



Sergio Espinosa-Alvarez, MD, is a Medical Graduate and Research Coordinator for the TANGO2 Natural History Study. With a background in pediatric cardiology, Sergio focuses on patient outcomes and improving quality of life for those affected by a heart condition.



Nikhita Gogate, MS, PhD (cand), in Genetics & Genomics at Baylor College of Medicine studies rare disease genomics and genotype-phenotype relationships through integrative clinical-genomic analyses. She completed a clinical translational certificate program (CTR-CAQ) under Dr. Seema Lalani, advancing a clear path from bench to bedside. Her work on TDD evaluates 120 individuals from the natural history study, applies phenotypic analysis to delineate subgroups and identifies an association between c.460G>A (p.G154R) and metabolic crises. Outside the lab, Nikhita enjoys cooking and trips to national parks.





TANGO2 Deficiency Disorder Clinician Consult Hotline

+1 (860) 598-0955

The TANGO2 Hotline provides peer-to-peer life-saving consultation to healthcare professionals treating patients with TANGO2 Deficiency Disorder (TDD). This includes patients that are hospitalized, currently in crisis, or at risk of crisis.

This international resource is available 24 hours/7 days a week.

CALL NOW



TANGO2
Emergency Protocol



TANGO2
Clinician Consult
Hotline



For Healthcare
Professionals

GRANT CYCLE 6

Now Open!



LETTERS OF INTENT DUE: 11/07/2025 5PM EST

SINGLE INSTITUTION AWARD - UP TO \$25,000 (USD)

MULTI-INSTITUTION AWARD - UP TO \$50,000 (USD)

PLUS AN OPPORTUNITY FOR ADDITIONAL FUNDING
WITH A COMPETITIVE ONE YEAR RENEWAL

TANGO2 NATURAL HISTORY STUDY PARTICIPANTS NEEDED

WHY IS PARTICIPATION CRITICAL?

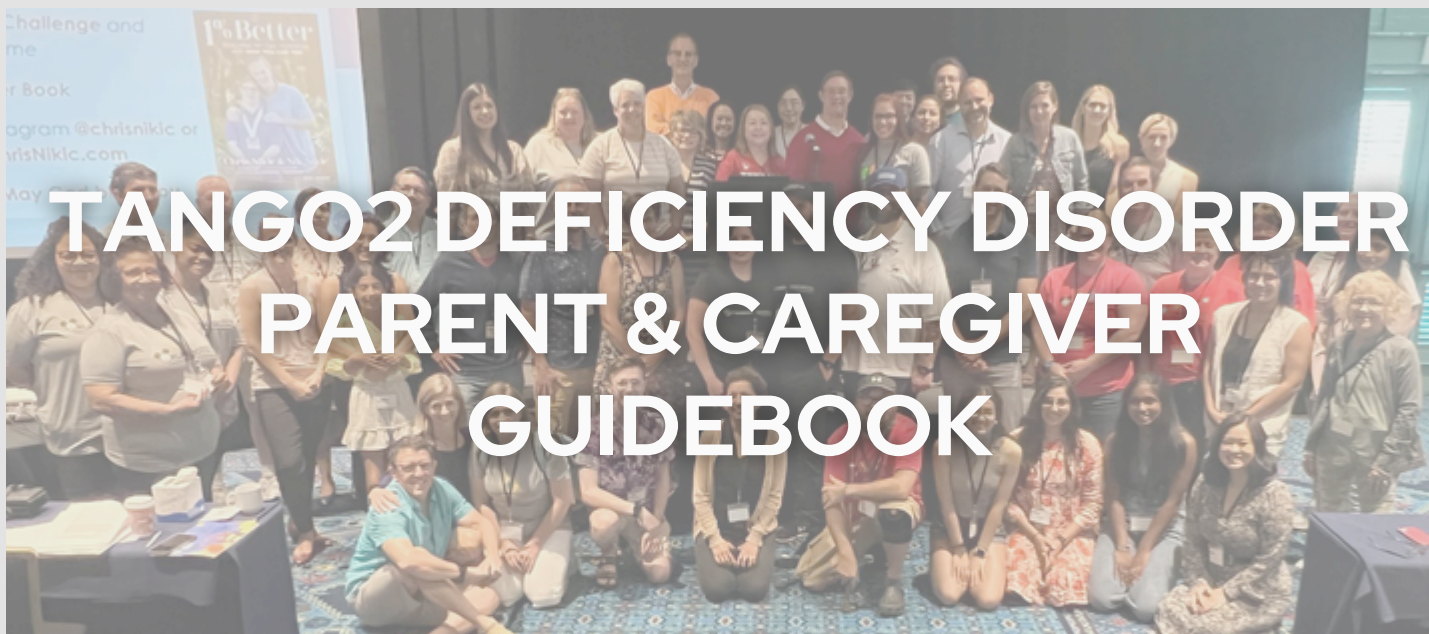
Supported by the TANGO2 Research foundation, this study aims to enhance our understanding of TANGO2 deficiency disorder progression over time, offer opportunities to improve clinical care, establish comprehensive treatment guidelines and ultimately find a cure.

WHO CAN PARTICIPATE?

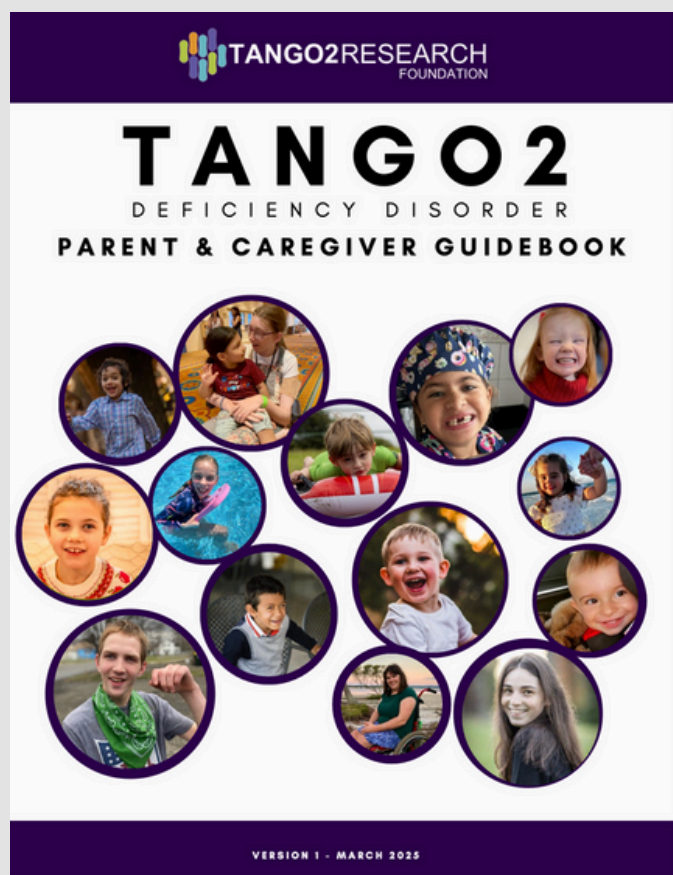
Any individual diagnosed with TANGO2 deficiency disorder and/or 22q11.2 Deletion Syndrome.

**FOR MORE INFORMATION AND TO ENROLL,
EMAIL TANGO2.RESEARCH@BCM.EDU.**





The Outreach Committee of the TANGO2 Research Foundation, comprised of parent & caregiver volunteers, is proud to share a resource we wish we had when our own children were first diagnosed. A TANGO2 Deficiency Disorder (TDD) diagnosis can feel overwhelming, bringing uncertainty and more questions than answers. This guidebook is designed to ease that first step by providing clear, accessible information about TDD, personal reflections from caregivers, guidance on treatment options such as B vitamins, and links to trusted resources. Most importantly, it serves as a reminder that you are not alone. Created especially for newly diagnosed families, this guide offers knowledge, hope, & connection to a community that understands your journey.



**SCAN TO SIGN UP FOR A FREE
DIGITAL COPY OF OUR GUIDEBOOK**

Save the Date!

June 28 - 30, 2026

Disney's Coronado Springs Resort
Orlando, Florida





WWW.TANGO2RESEARCH.ORG