September 2025

ROSAMUND STONE ZANDER TRANSLATIONAL NEUROSCIENCE CENTER

Boston Children's Hospital

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL

Rosamund Stone Zander Translational Neuroscience Center Newsletter

Welcome to the first edition of our new Rosamund Stone Zander Translational Neuroscience Center newsletter! We will release these newsletters on a quarterly basis: September, December, March, and June.

We're excited to launch this inaugural edition—a space to keep you informed and connected with the latest updates, important announcements, and highlights from across our RSZ TNC community.

We hope this becomes a helpful and engaging way to stay in the loop! If you have something you want to share in our next newsletter, be sure to fill out our RSZ TNC Quarterly Newsletter Submission Form or email us at tnc@childrens.harvard.edu with any questions.

In This Issue

Featured Initiative - Accelerating Clinical Trial Readiness Innovations for Monogenic Neurodevelopmental Disorders (ACTION)

The Rosamund Stone Zander Translational Neuroscience Center team was delighted to be highlighted in the recent publication of Brain and Life magazine for our partnerships with rare disease patient advocacy organizations to advance translational research for rare disorders.

Led by the Translational Genomic Medicine Core, including Director Dr. Maya Chopra, Fellow Dr. Cagri

Bayraktutan, and genetic counselors Abigail Sveden and Zoë Frazier, the ACTION Initiative is a 3-year longitudinal natural history protocol for rare monogenic neurodevelopmental disorders (NDDs). The goal of this project is to build clinical trial readiness through the generation of validated endpoints and biomarkers for such conditions, leveraging the expertise of the Human Neurobehavioral Core Service led by Dr. Ellen Hanson and the Clinical Neurophysiology Research Core, led by Dr. April Levin. The initial cohorts of the ACTION Initiative will be Kleefstra Syndrome, in partnership with Dr. Siddharth Srivastava, and MED13L, in partnership with Dr. Sara Trowbridge with enrollment beginning earlier this summer.

The MED13L cohort kicked off enrollment with data collection for nearly 20 individuals during the MED13L Research and Family Meetup from July 9-11, 2025. This conference data collection showcased the power of collaboration across RSZ TNC cores, along with patient advocacy group partnership.

New Clinical Trials & Research Studies

Jaguar Gene Therapy in SHANK3 Haploinsufficiency

PI: Dr. Siddharth Srivastava, IRB-P00049346

This is a Phase 1/2, first in human, open-label, dose-escalation study to evaluate the safety, tolerability, and clinical activity of a single dose of JAG201. It is administered via intracerebroventricular (ICV) injection in pediatric and adult participants with SHANK3 haploinsufficiency, which results from SHANK3 loss of function pathogenic variants and chromosomal deletions encompassing the SHANK3 gene.

The study will include 5 periods: Pre-Screening, Screening, Gene Therapy Administration and Perioperative Management, Initial Follow-Up, and Long-Term Follow-Up. In total, all participants will be followed for safety and tolerability for at least 5 years after the date of treatment with JAG201. We are seeking to enroll pediatric participants for the initial cohort, ranging from ages 2 to 9 years.

Read more study details on ClinicalTrials.gov

JAGUAR GENE THERAPY

Achievements & Celebrations

Fellows of the Child Neurology Society (FCNS) Program

The Child Neurology Society (CNS) recognizes 40 CNS members' outstanding achievements and exemplary service who have been accepted into the Fellows of the Child Neurology Society (FCNS) program. Notably, 3 of the 40 members accepted are from the Department of Neurology at Boston Children's Hospital.

Established in 2024, the FCNS program is a prestigious distinction to recognize esteemed members and honor their extraordinary professional achievements and contributions. Fellows are recognized as child neurology leaders who have made outstanding contributions to the field of child neurology and the CNS. RSZ TNC Affiliated Faculty awarded this honor include:

2025 Fellows

- Alexander Cohen, MD, PhD
- Scott Pomeroy, MD, PhD
- Mustafa Sahin MD, PhD

Congratulations on Recent Weddings!

Raymond Belanger Deloge and his wife, Michelle Leonard, tied the knot on June 14, 2025, in Salem, MA amongst their family and friends.

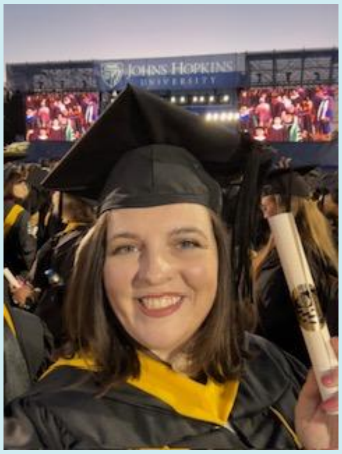
Zoë Frazier married her husband, Eric Link, on June 21, 2025, in Phippsburg, ME surrounded by their closest family and friends.

Congratulations on Recent Graduations!

Christine Barthelemy, Regulatory Specialist, graduated with her Master of Science in Regulatory Affairs and Health Policy from Massachusetts College of Pharmacy and Health Sciences.

Kate Pawlowski, Clinical Research Manager and Director of DDM Clinical Research Labs, graduated with her Master of Science in Research Administration from Johns Hopkins University.

Christine Barthelemy, MS



Kate Pawlowski, MS

New Staff & Interns

RSZ TNC New Employees

Brynn Heslin
Genetic Counseling Assistant
Tuberous Sclerosis Complex (TSC), CDKL5, Kleefstra Clinics
Supports the Neurology Department Genetic Counselors

Maggie Allen
Clinical Research Assistant I
Spinal Muscular Atrophy (SMA), Myotonic Dystrophy Type 1, and Duchenne Muscular Dystrophy (DMD)

Sydney Klau
Clinical Research Assistant I
PTEN Hamartoma Tumor Syndrome Natural History Study and Clinical Trial

Elizabeth Kolln
Clinical Research Assistant I
Down Syndrome Program and Research Scholars Program
Down Syndrome CAPE and Birthday Studies

Sahana Raman
Clinical Research Assistant I
Tuberous Sclerosis Complex (TSC) Natural History Study and Clinical Trial

Marena Neggers, MPH, CCRP
Regulatory Compliance Specialist
RSZ TNC Regulatory Team

Jillian O'Toole, MS, GC
Genetic Counselor
Tuberous Sclerosis Complex (TSC), Kleefstra, Rett Syndrome, CDKL5 Clinics
MED13L, CAGS, ReNU Syndrome Clinical and Research Efforts
CHD/ASD Sequencing Initiative Research Project

Helen Olson
Clinical Research Assistant I
CDKL5 and Rett Syndrome Natural History and Interventional Studies

RSZ TNC Summer Internships

Kaya Nichols studies neuroscience at Princeton University and is an aspiring doctor. At the TNC, she's been working with the Community Partnership Program to identify and reach out to potential health center and college program partners in an effort to diversify research and healthcare. Additionally, she has been developing her coding skills in the Cohen Lab, where she's analyzing perinatal stroke data and assisting in the current fMRI neurofeedback research study.

Damian Hoxhalli is a senior at Boston College High School who plans on studying sociology on the pre-med track in college. He dreams of pursuing his education at Tufts University with his application being due this upcoming cycle. At the TNC, Damian has been working with the PMS study, entering data into REDCap as well as creating/organizing subject binders. From this work, he has learned to keep himself organized and to pay attention to every single detail within a study. Damian also helps enter data for the PTEN and TSC studies.

RSZ TNC Partnerships

The Rosamund Stone Zander Translational Neuroscience Center has announced two new research partnerships with organizations focused on rare disease studies.

The iDefine-The Kleefstra Syndrome Foundation and the MED13L Foundation have each made commitments to natural history studies in collaboration with the RSZ TNC. Both will last over a three-year timespan to help create a better understanding of these rare diseases.

These studies are a part of the RSZ TNC's Accelerating Clinical Trial Readiness Innovations for Monogenic Neurodevelopmental Disorders (ACTION) Initiative, an umbrella program designed to establish clinical trial readiness for rare NDDs, leveraging shared RSZ TNC resources and pilot funding.

Led by Principal Investigator Dr. Maya Chopra, Clinical Geneticist and Director of the Translational Genomic Medicine Core of the RSZ TNC at Boston Children's Hospital, the ACTION Initiative provides scalable infrastructure for standardized data collection from individuals with NDDs, customized to the specific needs of individual disorders.

THE MED13L FOUNDATION

IDEFINE
The Kleefstra Syndrome Foundation

Recent Conferences & Events

Global Genes RARE Drug Development Symposium

The RARE Drug Development Symposium (RDDS) was hosted by Global Genes in partnership with the Rosamund Stone Zander Translational Neuroscience Center on September 3-4, 2025, in Boston, MA at the Joseph B. Martin Conference Center at Harvard Medical School.

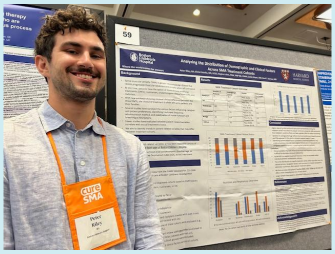
This symposium was designed to equip advocates with the knowledge, skills, and connections needed to navigate early-stage research with confidence, as well as offer practical insights to accelerate progress in research strategies and activities.

During this 1.5-day event, attendees had a mix of main stage sessions, targeted breakouts, group presentations, and hands-on workshops.

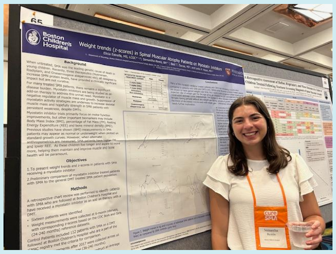
CureSMA Conference

Clinical Research Assistants, Peter Riley, Samantha Remis, and Gerald Mastellone shared their posters at the recent 2025 Annual Cure SMA Conference in Anaheim, CA.

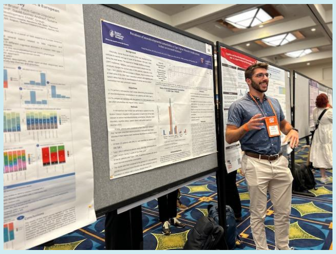
CureSMA picks the five best posters, and Gerald Mastellone's poster, "Prevalence of neurodevelopmental comorbidities in Type 1 Spinal Muscular Atrophy patients: **Is there an increased incidence?**" won an award. Gerald had the opportunity to give a five minute presentation to the entire conference group.



Peter Riley



Samantha Remis



Gerald Mastellone

Bottom Line Career Insights Event

Bottom Line is a non-profit organization that partners with first-generation students from low-income backgrounds to help them graduate high school, get into college, graduate college, and successfully launch careers.

On August 14, 2025, members of the RSZ TNC participated in a Bottom Line Career Insights Event, where they met with students virtually to give an introduction and overview of clinical research and the RSZ TNC, followed by a Career Panel, and a Key Skill Activity: Being a Clinical Research Coordinator.

Throughout this session, students were able to learn more about different roles within clinical research, as well as target key skills including communication, critical thinking, teamwork, and career and self-development, to help them find careers and build upon their professional development.

Brainstorming Bench to Bedside in Fragile X Syndrome

On May 22, 2025 the RSZ TNC hosted a focused one-day meeting aimed at addressing key challenges in the research and clinical translation of Fragile X Syndrome. The event brought together researchers, clinicians, patient advocacy groups, and families to share their experiences and collaboratively identify strategies to overcome barriers to effective translation.

Keynote presentations included Dr. Mike Tranfaglia, Director of the FRAXA Research Foundation, who provided a comprehensive overview of the history of Fragile X research and emerging therapeutic strategies. Dr. Elizabeth Berry-Kravis of Rush University reviewed the trajectory of clinical trials in Fragile X, highlighting lessons learned from past efforts and detailing recent advances. Professor Mark Bear of MIT presented an in-depth discussion of the mGluR Theory of Fragile X and the current status of translational research based on this model.

The meeting concluded with a powerful session featuring parents who shared firsthand experiences with clinical trial preparation and offered valuable recommendations for improving patient-centered approaches. Building on the success of this event, plans are underway to establish an annual workshop to foster collaboration and strengthen the Fragile X research and clinical community in the Boston area.

New Grants

RDCRN Project, Developmental Synaptopathies Associated with TSC, PTEN, SHANK3, and SynGAP1 Pathogenic Variants

We are pleased to announce that Dr. Mustafa Sahin has been awarded an NIH grant of over $8.6 million for the third five-year cycle of the Rare Disease Clinical Research Network (RDCRN) project, “Developmental Synaptopathies Associated with TSC, PTEN, SHANK3, and SynGAP1 Pathogenic Variants”.

This grant, which now involves 13 sites across the United States, aims to continue the comprehensive characterization of individuals with these rare diseases, focusing on cognition, communication, sleep, sensory deficits, and neuropsychiatric symptoms. The project also seeks to develop neurophysiological biomarkers, support pilot projects through strategic disorder-specific priorities, and foster a new generation of skilled investigators. Additionally, efforts to expand the research activities and effective dissemination of information, and plan for the consortium’s sustainability beyond RDCRN funding will also be a main focus of the next five years. This initiative represents a significant step forward in both rare disease research and patient advocacy.

IDefine and MED13L Foundation Fund RSZ TNC for Natural History Studies

Boston Children’s Hospital’s Rosamund Stone Zander Translational Neuroscience Center will be receiving a $300,000 grant from IDefine - The Kleefstra Syndrome (KS) Foundation– and a $250,000 grant from the MED13L Foundation for groundbreaking natural history studies under the ACTION Initiative. This commitment marks a major milestone for families affected by KS and MED13L Syndrome, rare neurodevelopmental disorders (NDDs), underscoring IDefine, the MED13L Foundation, and Boston Children’s shared mission to drive forward treatments for KS, MED13L Syndrome and other rare NDDs. In the longitudinal studies under the ACTION Initiative, clinical and biological data from 30 individuals with KS and 30 individuals with MED13L Syndrome will be collected over three years to better understand disease progression, identify potential biomarkers, and determine meaningful clinical trial endpoints.

Buffalano Family Supports Chopra-Amiel-Gordon Syndrome (CAGS) Clinic Launch

The Chopra-Amiel-Gordon Syndrome (CAGS) research team, led by Dr. Maya Chopra, received a $300,000 gift from the Buffalano Family, the third installment of gifts to support work for CAGS. This year’s generous gift supports the launch of a multidisciplinary CAGS clinic – a coordinated effort of neurogenetics, neurology, immunology, and nephrology specialists to care for individuals with suspected or confirmed CAGS. As the first clinic in the world dedicated to CAGS, the clinic will be a transformational resource for families searching for care tailored to their child’s specific needs. The first CAGS Clinic will be held on August 29, and slots will be available monthly thereafter.

Cyclin-Dependent Kinase-Like 5 (CDKL5) Pilot Grant Program Gives Award

Jed Hubbs, PhD and Elizabeth Buttermore, PhD were one of four Awardees of the 2025 CDKL5 Pilot Grant Program. They will be receiving $149,050 for their one-year research proposal, *Development of a tau hyperphosphorylation assay in CDKL5 deficiency disorder iPSC-derived neurons*.

The CDKL5 Pilot Grant Program is directed by the Loulou Foundation through its collaboration with the

CDKL5 Program of Excellence at the Orphan Disease Center of the University of Pennsylvania Perelman School of Medicine. The goal of the Program is to advance basic and therapeutic research on CDKL5 Deficiency Disorder, a rare neurodevelopmental disorder affecting thousands of patients across the world.

Recent Publications

Nature Genetics

Base editing of trinucleotide repeats that cause Huntington's disease and Friedreich's ataxia reduces somatic repeat expansions in patient cells and in mice

Nat Genet. 2025 May 26. doi: 10.1038/s41588-025-02172-8. Epub ahead of print. PMID: 40419681.

Read the paper

eNeuro

Syngap+/- CA1 Pyramidal Neurons Exhibit Upregulated Translation of Long MRNAs Associated with LTP

eNeuro. 2025 May 19;12(5):ENEURO.0086-25.2025. doi: 10.1523/ENEURO.0086-25.2025. PMID: 40295099; PMCID: PMC12091090.

Read the paper

Sage Open Pediatrics

Development of a Patient and Caregiver-Centered Pediatric Disease Concept Model for Kleefstra Syndrome

Sage Open Pediatrics. 2025 May 5;12.; doi:10.1177/30502225251336880

Read the paper

Nature Communications

Non-muscle myosin II inhibition at the site of axon injury increases axon regeneration

Nat Commun. 2025 Mar 26; doi: 10.1038/s41467-025-58303-6. PMID: 40140393

Read the paper

Translational Psychiatry

Shared early molecular mechanisms revealed in P301S and 5xFAD Alzheimer's disease mouse models

Transl Psychiatry. 2025 Mar 26; doi: 10.1038/s41398-025-03321-7. PMID: 40140365

Read the paper

Frontiers in Neuroscience

Harnessing the potential of human induced pluripotent stem cells, functional assays and machine learning for neurodevelopmental disorders

Front Neurosci. 2025 Jan 8; doi: 10.3389/fnins.2024.1524577. PMID: 39844857

Read the paper

Rosamund Stone Zander Translational Neuroscience Center Clinical Research Unit

Clinical research visit space now available at 2 BP

The **Rosamund Stone Zander Translational Neuroscience Center**
Clinical Research Unit (CRU) is located at
2 Brookline Place on the 7th floor

The CRU facilitates neurodevelopmental focused studies at no cost for RSZ TNC-affiliated investigators in collaboration with our partners in the Brain Mind Behavior Center including **Developmental Medicine, Genetics & Genomics, Neurology and Psychiatry**.

Apply here

CRU features:

- Wheelchair Accessible **Exam rooms** (3) with beds, otoscopes; 1 with oxygen and suction
- **Consult Room** (1) with adult and child-sized tables
- **IDS Pharmacy** is on-site 2 days per week with availability to be on-site other days with advance preparation
- **Height & Weight rooms** including scale for wheelchair users
- **Research Lab** for sample processing; -20 & -80 freezer space for same-day storage; & centrifuges

Types of Studies eligible:

- Pharmaceutical trials
- Device studies
- Behavioral intervention trials
- Natural history/outcome studies

Questions? Contact us at tnc@childrens.harvard.edu

Trainee Trialist Education Program

The goal of this initiative is to provide practical clinical research training to residents and fellow trainees within TNC affiliated departments/divisions who have an interest in clinical trials in neurodevelopmental disorders. Training is provided either individually or in a group session (depending on the numbers participating) and includes being paired with PIs/study teams for observation activities.

The program aims to provide trainees with a strong foundation to build upon so they're equipped with the skills necessary for roles in clinical research trials upon their graduation.

If you are interested in learning more about this program, please contacttnc@childrens.harvard.edu.

Regulatory Tips

Following a monitoring visit (whether it's an internal visit or a sponsor visit), address monitor comments and findings within 1 month of receipt. Timely responses help ensure good clinical practice and regulatory compliance.

Did you know that CRIT has new guidance on answering IT-related questions in CHeRP when submitting a new protocol? See here for more information.

For any regulatory needs, please contactsuzanne.lemoine@childrens.harvard.edu and someone from the regulatory team will follow up with you.

Watch for the "Regulatory Tips" section in future newsletters. These tips are geared towards investigators and their study teams involved in clinical research.

New Trainees Selected for Clinical Psychology Advanced Graduate Practicum

Five new trainees have been selected to participate in Boston Children's Hospital's Advanced Graduate Psychology Practicum. This Practicum, facilitated by the RSZ TNC, is an integrated program with in-service training at five BCH sites. The program provides in-depth training in neuropsychological and diagnostic assessment, differential diagnosis, treatment planning, and multidisciplinary collaboration in pediatric care.

The participating sites are:

- Arnett Laboratory
- Developmental Medicine Center
- Faja Laboratory
- Human Neurobehavioral Core Service (HNBCS)
- Cardiac Neurodevelopmental Program

This year's trainees are:

Diana Laenen
Developmental Medicine Center

Brittney Kleinfelter
Faja Lab

Mikaela de Lemos
Arnett Lab

Julia Kausel
Arnett Lab

Ryan Sunda
Human Neurobehavioral Core Service (HNBCS)

Boston Children's Hospital Anniversaries

Allyson McCrary, who joined the Clinical Research Operations (CRO) team in the RSZ TNC in June 2025 as Clinical Research Program Manager, recently celebrated 20 years working at Boston Children's Hospital as of July 2025.

Allyson feels fortunate to have had the opportunity to learn, grow, and succeed at BCH and looks forward to marking future anniversaries with both the hospital and the RSZ TNC!

RSZ TNC Community

Is your birthday included in our RSZ TNC birthday list?

Send us an email at tnc@childrens.harvard.edu and we'll add you to our birthday list, where we send out monthly emails so you can make sure you don't miss your team members' birthdays!

We want to see photos of your pets!

Bring in photos of your pets and add them to our RSZ TNC pet wall, which is located in the RSZ TNC administrative suite on the 7th floor of 2BP.

Stay Connected with the RSZ TNC

External Site: Rosamund Stone Zander Translational Neuroscience Center/ BCH Intranet: RSZ TNC

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