



PROGRAM

OCTOBER 8 - 10, 2026

Welcome

WE'RE SO GLAD YOU ARE HERE

CTNNBI Connect & Cure is honored to welcome you to our 5th CTNNBI Conference. Whether you are a person living with CTNNBI syndrome, a family member, clinician, researcher, industry partner, volunteer, or supporter, we are grateful you are here.

This event is an opportunity to learn, connect, collaborate, and strengthen the relationships that help drive progress forward. Over the next several days, we will share knowledge, celebrate achievements, tackle challenges, and explore new possibilities for improving the lives of individuals and families affected by CTNNBI syndrome.

Our vision is a world where every family affected by CTNNBI syndrome has access to safe and effective treatments and a supportive community. While there is still important work to do, this gathering is a reminder that none of us walks this path alone, and together we can continue building a brighter future for everyone living with CTNNBI.

EVENT AT A GLANCE

THURSDAY

- On-site Research
- Community Activities
- Grab-and-go Lunch
- Registration

FRIDAY

- On-site Research
- Community Activities
- Grab-and-go Lunch
- Registration
- Community Dinner

SATURDAY

- Breakfast
- Conference Sessions
- Resource Rooms
- Lunch
- Signature Dinner & Dance Party



2025 IMPACT REPORT

The progress highlighted in this report is only possible because of the strength of our community. Behind every milestone are families who share their stories, researchers committed to discovery, and supporters who believe in our mission.



Conference Resources



MOBILE HUB

- Conference Information
- Q&A Form
- More Resources



TRANSLATIONS



Live translation is available for all conference presentations via Wordly.

Special thanks to our Translation Services Sponsor:



CONFERENCE ALBUM

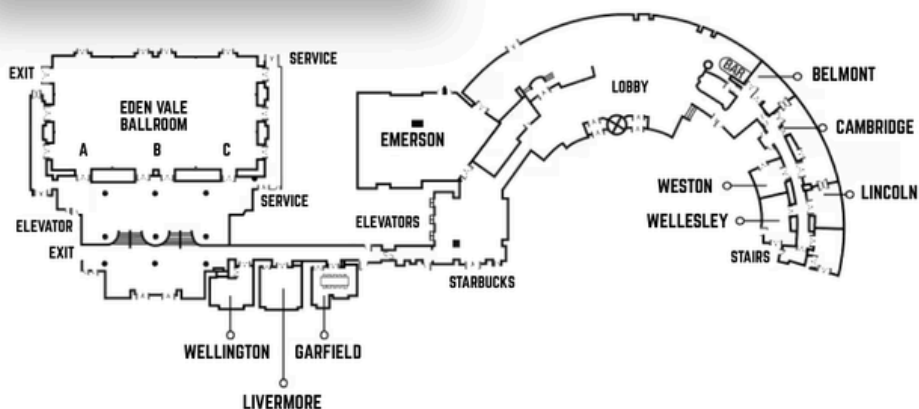
We'd love to see the conference through your eyes!

Take photos and videos throughout the weekend, then upload them to our shared conference photo album.

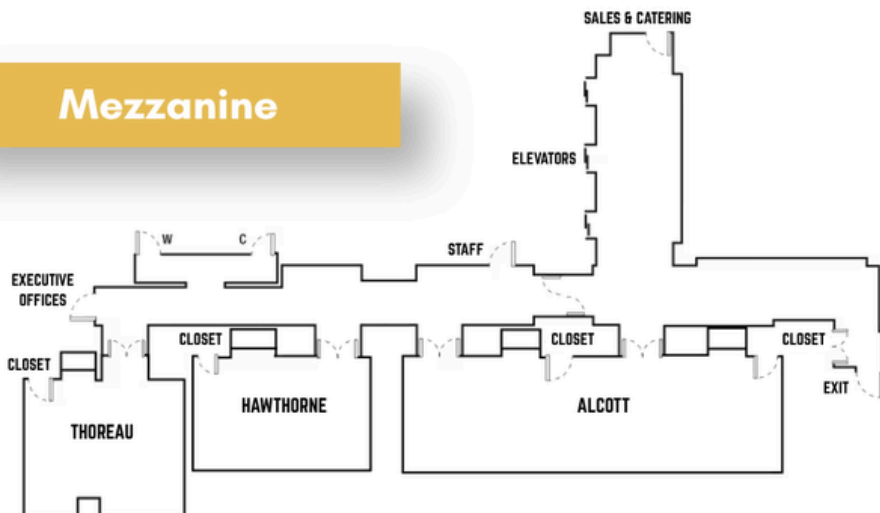


Hotel Map

1st Floor



Mezzanine



Thursday, Oct. 8



ACTIVITY STATIONS

Stop by for music, movement, art, coloring, bubbles, sensory activities, and some special visitors!

Children must be accompanied by an adult



8 - 10 AM & 1 - 3 PM



Cambridge, Lincoln, & Belmont



ON-SITE RESEARCH

BIOSAMPLE COLLECTION

In partnership with COMBINEDBrain



Walk-in 9AM - 5PM



Thoreau

NATURAL HISTORY STUDY CLINICAL EXAMINATIONS

In partnership with Boston Children's Hospital



Appointment only



Eden Vale Foyer

SPEECH STUDY

In partnership with Geisinger



Appointment only



Hawthorne



COMMUNITY CIRCLES

Facilitated conversation to connect and learn as a community

- Parents of 0-5 Year-Old Dragonflies
- Parents of Dragonflies Ages 6+
- Grandparents



10 - 11 AM



Cambridge, Lincoln, & Belmont



ORIENTATION FOR FAMILIES NEW TO CTNNB1

Meet other newly diagnosed families, ask questions, and build a foundation for your family's CTNNB1 journey



11 - 11:45 AM



Lincoln



LUNCH

Grab-and-go lunch

Generously sponsored by



SOPHIE'S HOPE
FOUNDATION



11:45 AM - 12:45 PM



Location to be announced



SPLASH ZONE

Swim, splash, and gather at the hotel pool



3:30 - 4:30 PM



Westin Waltham Pool



REGISTRATION

Check-in and receive conference materials



5 - 7 PM



Eden Vale Foyer

Friday, Oct. 9



ACTIVITY STATIONS

Stop by for music, movement, art, coloring, bubbles, sensory activities, and some special visitors!

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8 - 10 AM & 1 - 3 PM



Cambridge, Lincoln, & Belmont



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Hawthorne



SUPER SIBLING SOCIAL

A supportive gathering for brothers and sisters of individuals with CTNNB1 syndrome to connect, play games, and share their unique experiences.

Open to siblings of all ages. Children must be accompanied by an adult.



10:45 - 11:45 AM



Lincoln



LUNCH

Grab-and-go lunch

Generously sponsored by



11:45 AM - 12:45 PM



Location to be announced



REGISTRATION

Check-in and receive conference materials



3 - 5 PM



Eden Vale Foyer



COMMUNITY DINNER*

Gather together for an evening of connection and conversation before a full day of presentations

*This meal is self-pay



5 - 7 PM



Location to be announced

Saturday, Oct. 10

7:00 AM – 8:00 AM Breakfast

8:15 AM – 8:35 AM Opening Remarks

8:35 AM – 10:40 AM Session 1: Clinical Insights

8:35 AM – 9:05 AM KEYNOTE: Clinical features of CTNNB1 syndrome

Wendy Chung, MD, PhD, Boston Children's Hospital

9:05 AM – 9:20 AM CTNNB1 Disease Concept Model: Quantifying the patient experience

Sophie Marek, MS, CGC, COMBINEDBrain

9:20 AM – 9:35 AM Exploring the validity of EEG biomarkers in the CTNNB1 population: Findings from the 2025 EEG data collection

Nicolò Pini, PhD, Columbia University

9:35 AM – 9:50 AM Defining epilepsy and therapeutic targets in CTNNB1 syndrome: An integrated preclinical-clinical platform

Damon Page, PhD, Seattle Children's, University of Washington

9:50 AM – 10:05 AM Generating a plasma proteomic atlas of neurodegenerative rare disorders: A deeper dive into CTNNB1-related disorder biomarker discovery

Anna Pfalzer, PhD, Vanderbilt University

Ricardo Ramirez, PhD, RNR Consulting, LLC

10:05 AM – 10:20 AM From gut to brain: Understanding the microbiome in CTNNB1 syndrome

Mohammad Moshahid Khan, PhD, University of Tennessee

10:20 AM – 10:40 AM Session Q&A

10:40 AM – 10:55 AM Break

10:55 AM – 12:35 PM Session 2: Treatment Landscape & Preclinical Progress

10:55 AM – 11:10 AM Understanding your CTNNB1 variant

Emily Amerson, MS, CTNNB1 Connect & Cure

11:10 AM – 11:25 AM Therapeutics for CTNNB1 syndrome: A match made in mechanism

Soenika Gorrepati, Boston University School of Medicine

11:25 AM – 11:40 AM Identifying an efficacious therapeutic drug treatment for CTNNB1 syndrome

Michele Jacob, PhD, Tufts University

11:40 AM – 11:55 AM Development of RNA Boosters to enhance mRNA expression from genes associated with haploinsufficiency disorders

Dylan Miller, Doctoral Candidate, Johns Hopkins University

11:55 AM – 12:10 PM Targeting the β -catenin/cadherin axis in CTNNB1 syndrome: From small-molecule screening to multi-omic and cellular validation

Valter Tucci, FRISB, Italian Institute of Technology

12:10 PM – 12:35 PM Session Q&A

12:35 PM – 12:45 PM Break

12:45 PM – 1:00 PM Group Photo

1:00 PM – 2:00 PM Lunch

Sponsored by Special Needs Companies

2:10 PM – 3:25 PM Session 3: CTNNB1 in the Clinic

2:10 PM – 2:25 PM From flies to patients: The drug repurposing journey

Rachel Heilmann, PharmD, BCPS, RePURPOSEd Pharmacy Practice

2:25 PM – 2:40 PM CTNNB1 Drug Repurposing Project: Updates and observational study announcement

Kellan Weston, PhD, COMBINEDBrain

2:40 PM – 3:00 PM GAIN-CTNNB1: Updates from the clinical trial

Nina Žakelj, MD, University Children's Hospital Ljubljana

3:00 PM – 3:25 PM Session Q&A

3:25 PM – 3:40 PM Closing of Scientific Sessions

3:40 PM – 3:50 PM Break

3:50 PM – 5:00 PM Session 4: CTNNB1 Caregivers-Only Session

3:50 PM – 4:00 PM Launch of the CTNNB1 Patient Priority Survey

Kellan Weston, PhD, COMBINEDBrain

4:00 PM – 5:00 PM CTNNB1 Caregiver Workshop

Paige Ryan, DPT, Foundation First Physical Therapy

5:00 PM – 5:45 PM Break

5:45 PM – 8:30 PM "Wings in Motion" Dinner & Dance Party

Sponsored by Viralgen

8:00 AM – 5:00 PM Family Resource Rooms

We are happy to offer volunteer-led Resource Rooms for children and CTNNB1 adults who may not want to or be able to attend conference sessions. These rooms will be supported by parents, caregivers, and community volunteers, many with firsthand experience supporting children with CTNNB1 syndrome.

3 Ring Creations Circus Show: 11:30 AM – 12:00 PM

IncluYOUision Sports Adaptive Sports Session: 2:30 – 3:30 PM

Please note: This is not licensed childcare. Parents/guardians must remain nearby and available. Volunteer shifts will be organized throughout the day to help ensure consistent coverage and support.

KEEP THE MOMENTUM GOING

DONATE



CTNNB1 Connect & Cure is a volunteer-run Delaware nonprofit organization under Internal Revenue Code Section 501(c)(3).

Donate today to pave the way for a brighter tomorrow!



STAY CONNECTED



Follow our channels, tag us, and subscribe to our newsletter for the latest updates.



LEARN & EXPLORE



Browse our website for CTNNB1 information, research opportunities, educational resources, and more.



WAYS FOR CTNNB1 FAMILIES TO DRIVE PROGRESS

Research



- Complete the CTNNB1 Priority Survey
- Enroll in Citizen Health
- Enroll in Simons Searchlight
- Explore other research participation opportunities

Fundraising



- Create a Dragonfly Donations Page
- Set up a monthly donation and invite 5 friends or family members to do the same
- Share special campaigns (e.g., Silent Auction)
- Consider hosting a fundraising event

Volunteering



- Fill out the volunteer interest form
- Share the volunteer interest form with others who have relevant skills (they do not have to be a CTNNB1 parent)
- Apply for a leadership position

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SOPHIE'S HOPE
FOUNDATION



Gerald Cox Rare Care Consulting, LLC
The Rainone Family