CTNNB1 CONNECT & CURE NEWSLETTER

Key Conference Takeaways

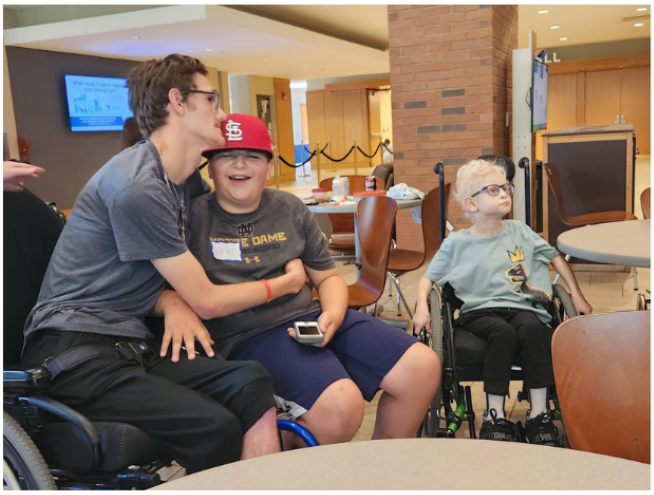
Hot News

Held First Natural History Study

Tufts/MIT Filed Patent for Treatment for CTNNB1

Strong small molecule candidate for treatment

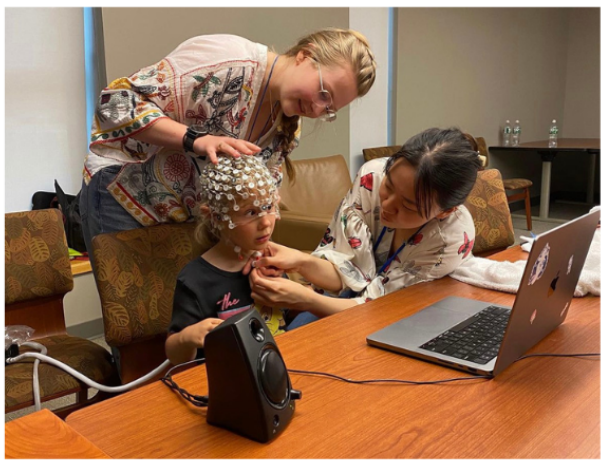
DNA/RNA Therapies underway



Natural History Study

CTNNB1 families traveled around the world and across the country to participate in our first in-person Natural History Study led by Principal Investigator Wendy Chung, MD, PhD and her team of clinicians from Columbia University Irving Medical Center.
The Natural History Study is an opportunity to advance the understanding of CTNNB1 syndrome through participation in clinical assessments to bring us closer to treatment and a clinical trial. This is something that CTNNB1 Connect & Cure plans to repeat yearly or bi-yearly to ensure meaningful endpoints are identified for measurement in future clinical trials. It also helps clinicians better understand how symptoms develop and change over time across various ages and mutations.

In total, 33 individuals affected by CTNNB1 syndrome participated in the study. Assessments included motor function (PT), neurological exam, research EEG, and neurocognitive exams. Additionally, 51 blood samples were collected by both Combined Brain and Simons Searchlight for our biorepositories for research. These samples are available to accredited researchers. Thanks to Simons Searchlight, 9 patient derived cell lines are already in the process of being created to test pharmacological treatments for CTNNB1 syndrome.
The study will move with Wendy Chung, MD, PhD to Boston Children's Hospital as she was recently named the hospital's next Chief of the Department of Pediatrics.



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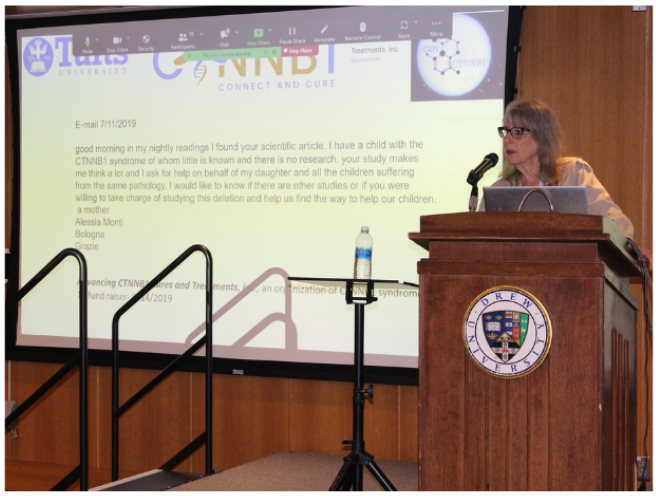
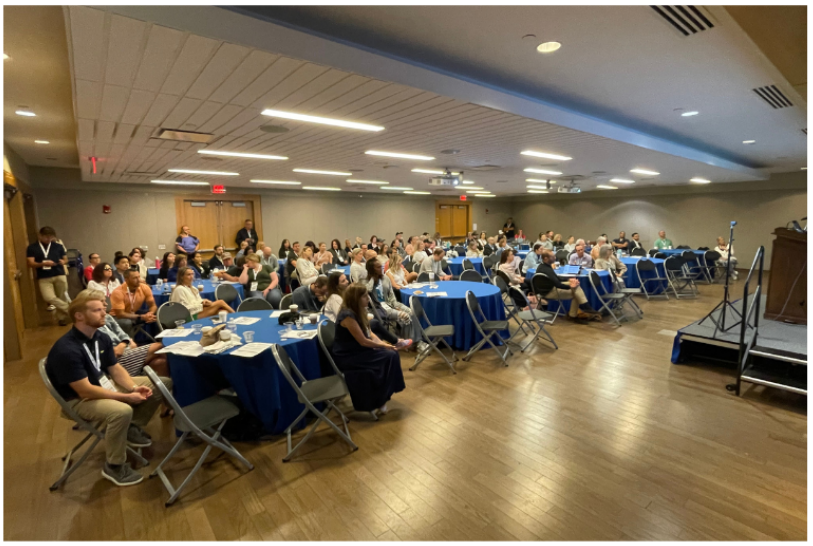


The great news is that while CTNNB1 Connect & Cure is busy filling our pre-clinical "toolbox" which includes – growing awareness of the disorder through initiatives like Cerebral Palsy clinic outreach and Probably Genetic free genetic testing, raising money for research and growing our patient registries with Simons Searchlight and Ciitizen, our researchers have developed a promising treatment for our patients. Michele Jacob, Ph.D and Jonathan Alexander, Ph.D at Tufts in collaboration with a medicinal chemist at MIT have filed a patent for method of treatment for CTNNB1 syndrome for a new drug they have developed. This drug brings the levels of beta-catenin (the protein encoded by the CTNNB1 gene and underproduced in our children) into the normal healthy range and leads to statistically significant improvements in motor and learning outcomes in our preclinical mouse model of CTNNB1 heterozygosity. The drug also increases the levels of beta-catenin in heterozygous CTNNB1 human cell models. Underway is additional testing in five CTNNB1 Syndrome patient derived cell lines to further prove that this drug could be a game changer for the CTNNB1 community. In addition, the team at Tufts, in collaboration with colleagues at Johns Hopkins, is exploring RNA and DNA based therapies for corrective outcomes and preliminary findings look promising.

Conference Key Takeaways

After two days of clinical assessments with the Columbia Team, 140 family members, friends, donors and researchers gathered at Drew University in Madison New Jersey on Saturday, June 24 for our CTNNB1 Connect & Cure Conference. Presenters in the morning session on translational research and clinical trial preparedness included Wendy Chung M.D., Ph.D Columbia University/Boston Children's Hospital, Gerald Cox, M.D., Ph.D Gerald Cox Rare Care Consulting & Boston Children's Hospital, Michele Jacob, Ph.D & Jonathan Alexander, Ph.D Tufts University, Mark Corbett B.Sc, Ph.D The University of Adelaide, Australia, Damjam Osredkar, M.D., Ph.D University Children's Hospital Ljubljana, CTNNB1 Foundation, Drew Scoles, M.D., Ph.D, Children's Hospital of Philadelphia and Mary Cobb from Global Genes. The afternoon session focused on the family experience with Luke Rosen, M.S. and Founder of KIF1A.org, Jennifer Siedman from Courageous Parents Network and Natasha and Tony King, Parents to Tony Jr. with CTNNB1 Syndrome.



CTNNB1 CONNECT AND CURE

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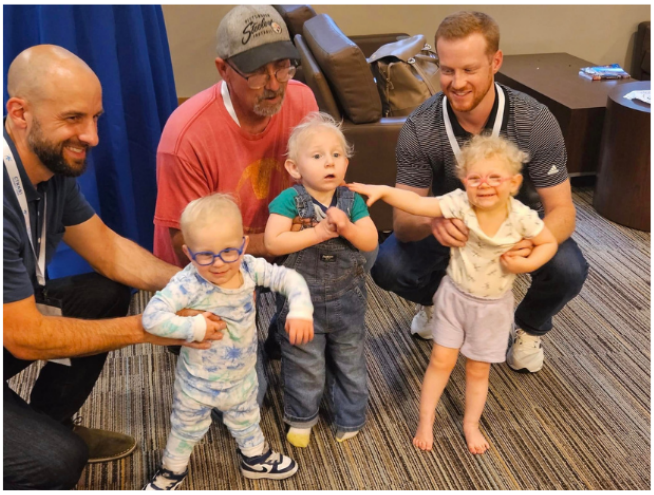

The key takeaway is that the research and findings thus far are very encouraging for the CTNNB1 community. What stands in the way is funding. Per the informative talk by Gerald F. Cox, MD, PhD, drug discovery and development is lengthy, expensive, and high risk. Our promising drug treatment still needs to be optimized, go through toxicology studies, be manufactured, obtain an IND from the FDA and approval from the Institutional Review Board to get into a human clinical trial.

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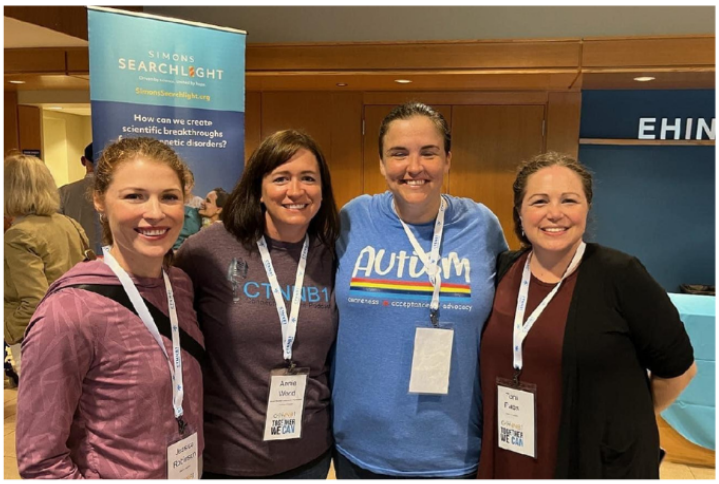

The CTNNB1 community is a powerful force in helping us get a treatment to clinical trial. As Wendy Chung, MD, PhD stated during our conference, we have become a highly engaged community with over 229 registered families in Simons Searchlight, our patient registry. We need to continue to expand our participation in Simons as well as our new registry Ciitizen. Growing our community is also a top priority. We are doing this through our Cerebral Palsy Outreach initiative as well as our partnership with Probably Genetic, free genetic testing. Together, we have the ability to raise awareness to the outside community with media outreach and the power of our voices. We need to be heard! Lastly is funding...every dollar counts. Each member of the CTNNB1 community plays an important part in helping us reach the goal line. This conference showed us the power of our community. Let's stay hopeful and engaged as we work together to make life better for our children and families.