CTNNB1
CONNECT AND CURE

Dear newly diagnosed family,

Welcome to the CTNNB1 family. Though none of us would have chosen to be here, our community will support, care, teach, inspire, and walk with you on this journey. We have all experienced the world-shifting moment that we received a CTNNB1 diagnosis and the emotions that precede and follow it. Though your life may now look very different than you imagined, there will be unique blessings sprinkled in and beautiful connections to be made, should you open yourself to them.
Most importantly, please remember that you are not alone.
So what now? Below are some steps and information that many of our CTNNB1 families have found helpful.

LEARN

The first step in processing a CTNNB1 diagnosis is learning about it. CTNNB1 Syndrome was first discovered in 2012. There are currently about 430 documented cases, though its actual prevalence is estimated to be more than 200,000 cases worldwide. The syndrome is caused by a variation of the CTNNB1 gene, which affects the body's production of a protein called beta-catenin. This protein is important for embryonic development and cell function. Most documented variants are "de novo," meaning new or not inherited. CTNNB1 Syndrome shares many symptoms with Cerebral Palsy. In fact, it is the most frequent genetic variant among those diagnosed with Cerebral Palsy. The features of CTNNB1 Syndrome vary by the individual, but can include some or all of the following: developmental delay, movement disorders, speech/language difficulties, behavior problems, altered muscle tone, sleep issues, microcephaly, impaired vision, and feeding difficulties. Published studies about CTNNB1 Syndrome can be found at www.curectnnb1.org/research/related-studies/. Therapies can vary widely and you may find that you often know more about what works for your child than your medical team does. Our parents' Facebook page (www.facebook.com/groups/787268954682708/) is especially helpful in sharing this kind of information.

BE COUNTED

We are asking all CTNNB1 families to register for Simons Searchlight and Ciitizen (Invitae). These databases collect information and provide an understanding of what CTNNB1 Syndrome looks like across all ages and developmental milestones, a crucial step in the process of finding treatments and ultimately a cure! Please see the steps below.

INVITAE ciitizen

Offered only in the U.S. for now
1. Create an account at: https://www.ciitizen.com/rarenetwork/CTNNB1Connect.
2. Verify your identity by uploading your child's birth certificate and your driver's license.
3. Verify the diagnosis by uploading your child's genetic report or select the name of the lab that did the work.
4. Add the names of institutions and providers visited to obtain records from.

SIMONS SEARCHLIGHT

1. Create an account at: https://research.simonssearchlight.org/account/create.
2. Upload your child's genetic report.
3. Participate in a short call with a genetic counselor to discuss your child's medical history and development.
4. Provide a blood sample if you are interested.

Please email questions to coordinator@simonssearchlight.org.

CURRENT RESEARCH

Natural History Study: This study, funded by the U.S.-based CTNNB1 Connect & Cure charity, is led by Principal Investigator Dr. Wendy Chung and her team of clinicians from Columbia University Irving Medical Center. It is an opportunity for patients and families from the CTNNB1 community to advance clinical understanding of CTNNB1 Syndrome and bring us closer to clinical trials and treatment. The first clinical assessments for this study will occur at the CTNNB1 Connect & Cure Research Conference in June 2023.

Pharmaceutical Treatments: CTNNB1 Connect & Cure is funding the work of Dr. Michele Jacob and her team at Tufts University to find and assess small-molecule treatments that correct reduced beta-catenin levels. Data from completed in vivo mouse models and current in vitro human cell models will provide the foundation needed to get to clinical trials.

Gene Replacement Therapy: The European-based CTNNB1 Foundation is funding research to create a gene replacement therapy. For more information, please visit ctnnb1-foundation.org/.

MAKE A DIFFERENCE

The largest impacts our families can make are by spreading awareness and by fundraising. Because CTNNB1 Syndrome is newly discovered and very rare, it is up to us to make it known. One population that we need to reach are individuals who have been diagnosed with Cerebral Palsy. We want to make pediatricians and pediatric specialists aware of CTNNB1 Syndrome and the need for earlier, accurate diagnosis through genetic testing. We also need funding to keep the research moving forward. Several CTNNB1 families have organized wonderful fundraising events. You can find more information about getting involved at https://www.curectnnb1.org/get-involved/.

GET CONNECTED

@ctnnb1connectcure CTNNB1 Connect & Cure @ctnnb1cc

In addition to following/subscribing to our accounts above, please listen and subscribe to our CTNNB1 Connect & Cure Podcast, hosted by CTNNB1 mom and board member, Annie Wood. We also highly recommend the award-winning rare disease podcast Once Upon a Gene, hosted by CTNNB1 mom and board member, Effie Parks.

Together, we can move mountains and create better lives for our loved ones with CTNNB1 Syndrome. We are sorry you have found yourselves here in this frightening and unexpected place, as we all were at diagnosis, but please know that we welcome your family into the CTNNB1 family with true understanding and warm embrace. Our board member, Ashley (ashley@curectnnb1.org) would love to connect with you and answer any questions that you may have, as we know all of this is a lot to take in.

Sincerely,
The CTNNB1 Connect & Cure Team