CTNNB1 SYNDROME
FAMILY RESOURCE GUIDE

Awareness, Resources and Connections

CTNNB1 SYNDROME AWARENESS WORLDWIDE

Founded in 2017, CTNNB1 Syndrome Awareness Worldwide (CSAW) is a 501(c)3 non-profit organization dedicated to raising awareness of CTNNB1 Syndrome, providing information about symptoms, diagnosis, treatments and therapies, and connecting affected families. Find more information at www.ctnnb1.org.

www.ctnnb1.org

info@ctnnb1.org

facebook.com/ctnnb1org

@ctnnb1org

1-877-739-7789

https://podcasts.apple.com/us/podcast/once-upon-a-gene/id1485249347

parents-only facebook group
facebook.com/groups/787268954682708

Funding Research

ADVANCING CTNNB1 CURES AND TREATMENTS

Advancing CTNNB1 Cures and Treatments (ACCT) is a 501(c)3 organization incorporated in 2019 with the sole focus of funding research that will develop treatments, and potentially a cure for CTNNB1 Syndrome. Find more information at www.curectnnb1.org.

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Registry

SIMONS SEARCHLIGHT

All CTNNB1 Families are highly encouraged to participate in the Simons Searchlight Registry and Natural History Study at www.simonssearchlight.org.

facebook.com/groups/ctnnb1

CTNNB1 Syndrome Frequently Asked Questions (FAQ)

What is CTNNB1 Syndrome? The CTNNB1 gene provides the blueprint for beta-catenin, a protein that allows for cell specialization, cell division/growth, cell adhesion, and inter-cell communication. CTNNB1 Syndrome refers to deletion, partial deletion or mutation of the gene.

What are its symptoms? CTNNB1 Syndrome symptoms range from mild developmental delays to severe physical and intellectual disabilities, including some, all, or (possibly) none of the following: global developmental delay, spasticity, hypotonia, microcephaly, amblyopia, strabismus, and hyperopia. Many with CTNNB1 Syndrome have repetitive behaviors, sensory processing issues, a high startle reflex and feeding difficulty.

How is it diagnosed? CTNNB1 Syndrome is definitively diagnosed using Whole Exome Sequencing (WES). As of late 2019, there are fewer than 200 people known to have been diagnosed with CTNNB1 Syndrome. Prior to a CTNNB1 diagnosis, many are diagnosed as having Autism Spectrum Disorder or a variant of Cerebral Palsy. As genetic testing becomes more widely available, more cases are being newly diagnosed, or reassessed.

Is there a cure? While there is no cure for CTNNB1 Syndrome at this time, each of its associated symptoms may be treated. Those with hypotonia, for instance, report good results with physical (physio) therapy. Speech-Language Pathology and Occupational Therapy are frequently needed for gross motor and fine motor delays. Equine therapy is also beneficial for core strength.

Is there CTNNB1-specific research underway? ACCT has identified two leading scientists, Wendy K. Chung, M.D., PH. D at Columbia University and Michele H. Jacob., PH.D. at Tufts University who will be studying our specific gene and developing a registry and natural history database with Simons Searchlight.

I**s other scientific research underway?** With the advent of diagnostic gene technology, there is emerging research targeted at the CTNNB1 gene, the beta-catenin protein, the Wnt pathway, and related disorders under which those with CTNNB1 Syndrome may have been diagnosed previously such as Cerebral Palsy and Autism. Researchers into that broad scope include Dr. Michael Kruer of the University of Arizona College of Medicine; and Dr. Raphael Bernier at the University of Washington Bernier Lab TIGER (The Investigation of Genetic Exome Research) Study, among others.

How does CSAW advance its mission of connecting families? CSAW maintains a multilingual website with a parents' forum, a public Facebook page, Instagram, Twitter and Linked-In accounts, a toll-free telephone number, and an online newsletter. We will add a YouTube channel in 2019. We sponsor regional meet-ups for affected families. Our symbol is the dragonfly, and we have designated **July 25th**, which is National Dragonfly Day for many countries, as World CTNNB1 Syndrome Awareness Day. The first CTNNB1 Syndrome national event is slated for 2020. We also point people to the parents-only facebook group at facebook.com/groups/787268954682708.

Do CSAW and ACCT work with other groups? Recognizing that each rare disease has a limited constituency, we align with organizations that work on behalf of rare diseases overall to advance public and scientific awareness of the breadth of rare disease. These include the Unique (rarechromo.org), National Organization of Rare Disorders, (NORD), Global Genes, Simons Searchlight, and CRISPR Cas9 Genetic Disability.