CTNNB1 Syndrome

Synonyms:

- CTNNB1 neurodevelopmental disorder (CTNNB1-NDD)
- Neurodevelopmental disorder with spastic diplegia and visual defects (NEDSDV)
- Severe intellectual disability-progressive spastic diplegia syndrome

Individuals with CTNNB1 Syndrome have a variant of the CTNNB1 gene, located on the 3rd chromosome. Any variation in the DNA sequence of this gene affects the body's production of a protein called beta-catenin. This protein is present in many types of cells and tissues. It plays an important role in cell adhesion, communication between cells, differentiation of cells, and the maintenance of tissue homeostasis. Though signs and symptoms can vary between individuals, the most common ones include: intellectual disability, developmental delays, abnormal muscle tone, vision impairments, sleep issues, and behavioral problems.

ABOUT US

CTNNB1 Connect & Cure is dedicated to finding treatments and a cure for CTNNB1 Syndrome while raising awareness and connecting families.

https://www.curectnnb1.org
info@curectnnb1.org

CONNECT WITH US

CTNNB1
CONNECT AND CURE

Research

COLUMBIA | COLUMBIA UNIVERSITY IRVING MEDICAL CENTER

Natural History Study: Principal Investigator Dr. Wendy Chung and her team of clinicians from Columbia University Irving Medical Center are gathering more information about CTNNB1 Syndrome and how it affects individuals over time. The first clinical assessments for this study will occur at the CTNNB1 Connect & Cure Research Conference in June 2023.

Tufts UNIVERSITY

Pharmaceutical Treatments: Dr. Michele Jacob and her team at Tufts University is analyzing 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.

CTNNB1 Foundation

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/.

Community & Partners

Even though CTNNB1 Syndrome is a rare disease—impacting an estimated 1 in 35,000—our community of children, families, friends, researchers, healthcare professionals and partners has created a network that extends beyond individual geographies and genetics.

SIMONS SEARCHLIGHT

Once Upon A GENE

NORD®

INVITAE

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COMBINED

Global Genes® Allies in Rare Disease

HAYSTACK PROJECT The Voices of Rare & Ultra Rare

CTNNB1 is the most common genetic cause of Cerebral Palsy.

No Cost Genetic Testing

https://www.curectnnb1.org/symptom-checker
(Currently available only in the U.S.)

probably genetic