

Mission

CTNNB1 Connect and Cure is a nonprofit organization dedicated to finding treatment options and a cure for CTNNB1 Syndrome while improving the lives of our patients and community.

Vision

To create a world where every family affected by CTNNB1 Syndrome has access to safe and effective treatments and a supportive community.

Core Values

Transparency
Teamwork
Integrity

Better care today
while we fight for a
brighter tomorrow



 www.currectnnb1.org

 221 W 9th St, Ste 421
Wilmington, DE 19801

 info@currectnnb1.org



Tethered Spinal Cord Syndrome (TSCS)





What is CTNNB1?

CTNNB1 is a gene located on the 3rd chromosome that encodes for a protein called beta-catenin that acts as a key mediator in Wnt signaling and cell to cell adhesion. Mutations or variants in CTNNB1 lead to a broad spectrum of symptoms, including:

Tethered spinal cord (TSCS)

- Cognitive impairment
- Microcephaly
- Behavioral challenges
- Sleep disturbances
- Limited speech / nonspeaking
- Epilepsy
- Truncal hypotonia
- Peripheral spasticity
- Dystonia
- Strabismus
- Refractive errors
- Familial exudative vitreoretinopathy
- Congenital heart defects
- Osteopenia and scoliosis
- Feeding difficulties
- Gastrointestinal problems



Given the implications for patient care, we recommend considering spinal imaging in individuals with CTNNB1 neurodevelopmental disorder, particularly in patients with progressive spasticity or bladder dysfunction.

(Cornaz Buros. S., et al)

TSCS & CTNNB1: Relevant Research

Cornaz Buros S, Eggli S, Mirosevic S, Scherer C, Osredkar D, Schucht P, Petutschnigg T, Klein A, Grunt S, Lutz K. **Tethered cord in CTNNB1 neurodevelopmental disorder, a not-so-rare comorbidity: Results of a parent survey and a clinical example.** *Neuropediatrics*. 2025 Sept;56(S 01), S1–S24. doi: 10.1055/s-0045-1812147.

Sudnawa KK, Garber A, Cohen R, Calamia S, Kanner CH, Montes J, Bain JM, Fee RJ, Chung WK. **Clinical phenotypic spectrum of CTNNB1 neurodevelopmental disorder.** *Clin Genet*. 2024 May;105(5):523-532. doi: 10.1111/cge.14487. Epub 2024 Jan 21. PMID: 38247296; PMCID: PMC11872165.



Watch the latest CTNNB1 Conference Presentation about Tethered Spinal Cord Syndrome

