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

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CTNNB1
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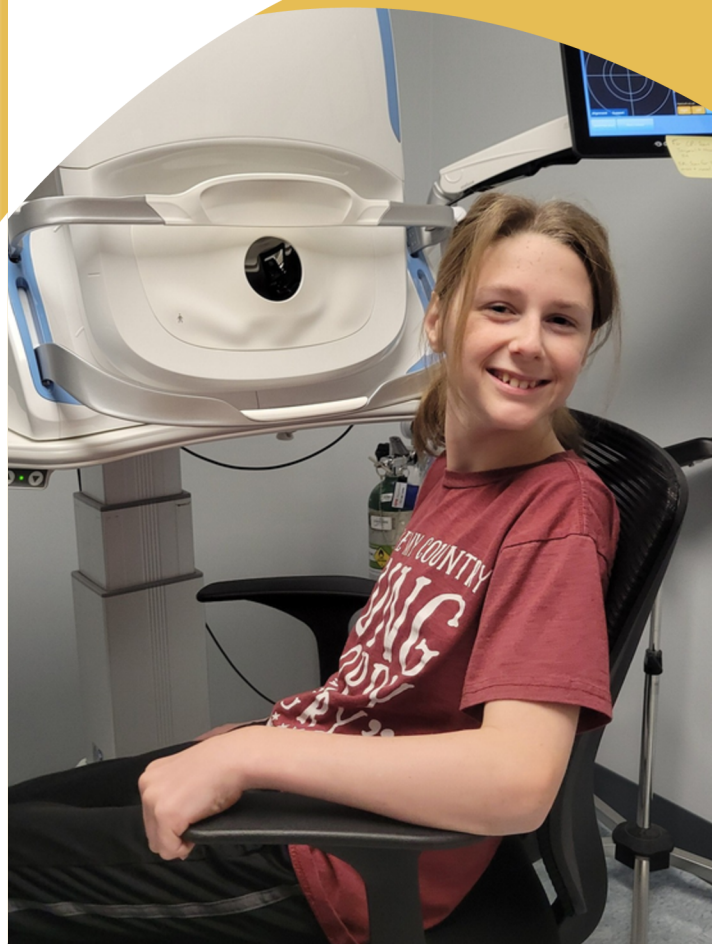
Familial Exudative Vitreoretinopathy (FEVR)



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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:

Familial exudative vitreoretinopathy

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

"Patients in our cohort require treatment for FEVR despite repeatedly normal dilated examinations in clinic over multiple years."
(Requejo Figueroa, G.A., et al)

FEVR & CTNNB1: Relevant Research

Requejo Figueroa GA, Morgan DJ, Jardine GJ, Young MP, Hwang ES. **Utility of Fluorescein Angiography for Early Detection of Familial Exudative Vitreoretinopathy in Neurodevelopmental Disorder With Spastic Diplegia and Visual Defects Due to CTNNB1 Variants**. J Pediatr Ophthalmol Strabismus. 2025 May-Jun;62(3):166-172. doi: 10.3928/01913913-20241122-01. Epub 2024 Dec 30. PMID: 39749995; PMCID: PMC12212438.

Bedoukian EC, Forbes G, Scoles D. **Vitreoretinopathy in Asymptomatic Children With CTNNB1 Syndrome**. JAMA Ophthalmol. 2024;142(9):874–878. doi:10.1001/jamaophthalmol.2024.2847

Huang L, Lu J, Wang Y, Sun L, Ding X. **Familial Exudative Vitreoretinopathy and Systemic Abnormalities in Patients With CTNNB1 Mutations**. Invest Ophthalmol Vis Sci. 2023 Feb 1;64(2):18. doi: 10.1167/iovs.64.2.18. PMID: 36790797; PMCID: PMC9940768.

CTNNB1 Connect & Cure. **"CTNNB1 and the Eyes | Drew Scoles, MD, PhD."** *YouTube*, 14 Aug. 2025, https://youtu.be/-lBv_13o6ME?si=zr5Ot05G7XxF4RxK