What is CTNNB1 Syndrome?

CTNNB1 Syndrome is a rare neurodevelopmental disease caused by changes to a gene called CTNNB1.

The CTNNB1 gene provides instructions for making a protein called beta-catenin.

Beta-catenin 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.

Hallmarks of CTNNB1 Syndrome are intellectual disability, developmental delays, abnormal muscle tone, vision impairments, sleep issues, and behavioral problems.

There is a broad range of severity and not all symptoms listed here are experienced by all who are diagnosed.

Symptoms

- Global Delays
- Microcephaly
- Sensory Issues
- Hypotonia
- Hypertonia
- Strabismus
- Sleep Issues
- Dystonia
- Seizures
- Hyperopia
- IUGR
- Speech/language difficulties
- FEVR
- Chronic constipation
- Dysphagia
- GERD
- Anxiety
- Autism
- Tethered Cord
- Frequent ear infections
- Missing teeth
- ADHD
- OCD
- Apnea
- Persistent cough
- Asthma
- Vesicoureteral reflux

CTNNB1
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