

GRIN2B-related neurodevelopmental disorder

Description

GRIN2B-related neurodevelopmental disorder is a condition that affects the nervous system. Neurodevelopmental disorders result from impaired growth and development of the central nervous system, which includes the brain and spinal cord, and the nerves connecting them. These disorders often affect learning ability, memory, and behavior and can be associated with other neurological problems.

Individuals with *GRIN2B*-related neurodevelopmental disorder have mild to profound intellectual disability and delayed development of speech and motor skills, such as sitting and walking. Some affected individuals never develop speech or the ability to walk on their own. Many people with this condition have weak muscle tone (hypotonia), which can contribute to the problems developing motor skills and lead to difficulty eating. Some affected individuals have abnormal muscle stiffness (spasticity), which can also cause problems with movement.

Recurrent seizures (epilepsy) occur in about half of people with *GRIN2B*-related neurodevelopmental disorder. About one-quarter of affected individuals have features of autism spectrum disorder, which is characterized by impaired communication and social interaction. Affected individuals may also be hyperactive, impulsive, or easily distractible, and some are described as being overly friendly. Sleeping difficulties can also occur in this condition.

Less common features of *GRIN2B*-related neurodevelopmental disorder include structural brain abnormalities, an unusually small head size (microcephaly), impaired vision, and involuntary muscle movements.

Frequency

The prevalence of *GRIN2B*-related neurodevelopmental disorder is unknown. Fewer than 100 cases have been reported in the medical literature.

Causes

GRIN2B-related neurodevelopmental disorder is caused by mutations in a gene called *GRIN2B*. This gene provides instructions for making a protein called GluN2B, which is found in nerve cells (neurons) in the brain primarily during development before birth. This protein is a part of specialized protein structures called NMDA receptors, which are

involved in normal brain development, changes in the brain in response to experience (synaptic plasticity), learning, and memory.

Some *GRIN2B* gene mutations lead to production of a nonfunctional GluN2B protein or prevent the production of any GluN2B protein from one copy of the gene in each cell. A shortage of this protein may reduce the number of functional NMDA receptors, which would decrease receptor activity in cells. Other mutations lead to production of abnormal GluN2B proteins that likely alter how the NMDA receptors function; some mutations reduce NMDA receptor signaling while others increase it. Researchers are unsure how abnormal activity of NMDA receptors prevents normal growth and development of the brain or why too much or too little activity lead to similar neurological problems in people with *GRIN2B*-related neurodevelopmental disorder.

[Learn more about the gene associated with GRIN2B-related neurodevelopmental disorder](#)

- GRIN2B

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Most cases of this condition result from new (de novo) mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) in an affected individual's parent or in early embryonic development. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- EIEE27
- Epileptic encephalopathy, early infantile, 27
- GRIN2B encephalopathy
- GRIN2B related syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Developmental and epileptic encephalopathy, 27 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4015316/>)
- Genetic Testing Registry: Intellectual disability, autosomal dominant 6 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151411/>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 27; DEE27 (<https://omim.org/entry/616139>)
- INTELLECTUAL DEVELOPMENTAL DISORDER, AUTOSOMAL DOMINANT 6, WITH OR WITHOUT SEIZURES; MRD6 (<https://omim.org/entry/613970>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28GRIN2B%5BTIAB%5D%29+AND+%28%28encephalopathy%29+OR+%28neurodevelopment%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

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