GRIN2B-related neurodevelopmental disorder

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GRIN2B gene

GRIN2B gene

glutamate ionotropic receptor NMDA type subunit 2B

Normal Function

The GRIN2B gene provides instructions for making a protein called GluN2B. This protein is found in nerve cells (neurons) in the brain, primarily during development before birth. The GluN2B protein is one component (subunit) of a subset of specialized protein structures called NMDA receptors. There are several types of NMDA receptors, made up of different combinations of proteins.NMDA receptors are glutamate-gated ion channels. When brain chemicals called glutamate and glycine attach to the receptor, a channel opens, allowing positively charged particles (cations) to flow through. The flow of cations activates (excites) neurons to send signals to each other. The cation flow also plays a role in the process by which the neurons mature to carry out specific functions (differentiation). NMDA receptors are involved in normal brain development, changes in the brain in response to experience (synaptic plasticity), learning, and memory.

Health Conditions Related to Genetic Changes

GRIN2B-related neurodevelopmental disorder

Several dozen mutations in the GRIN2B gene have been found to cause GRIN2B-related neurodevelopmental disorder, which is characterized by intellectual disability and delayed development of speech and motor skills. Other neurological problems that commonly occur in this disorder include seizures, weak muscle tone (hypotonia), movement disorders, and behavioral problems.Many 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 reduce receptor activity in cells. Other mutations lead to the 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.

More About This Health Condition

Autism spectrum disorder

MedlinePlus Genetics provides information about Autism spectrum disorder

More About This Health Condition

Other Names for This Gene

GluN2B glutamate [NMDA] receptor subunit epsilon-2 glutamate receptor ionotropic, NMDA 2B precursor glutamate receptor subunit epsilon-2 glutamate receptor, ionotropic, N-methyl D-aspartate 2B hNR3 N-methyl D-aspartate receptor subtype 2B NMDAR2B NR2B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of GRIN2B

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL-D-ASPARTATE, SUBUNIT 2B; GRIN2B

Gene and Variant Databases

NCBI Gene

ClinVar

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Genomic LocationThe GRIN2B gene is found on chromosome 12.

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