

GRIN2B-RELATED NEURODEVELOPMENTAL DISORDER SYMPTOMS

For patients and families

Information About the Respondents*

61 families hailing from 17 different countries. Of the respondents, 57% were Male and 43% were Female and consisted of the following age ranges:

- 30% 0-4 years of age
- 48% 5-10 years of age
- 15% 11-14 years of age
- 7% 15-18 years of age

**Hypotonia
(low muscle
tone)**

90%

**Most Common
Symptoms**

90% or more of the respondents reported the following symptoms & characteristics:

**Gross and Fine
motor delays
and disabilities**

**Cognitive
delays and
disabilities**

**Non-verbal
and/or
limited speech
communication**



Additional Symptoms:

Below are additional symptoms or behaviors noted by parents of children with GRIN2B.

Listed from MOST common to LEAST common. Not all symptoms are present in each child and symptoms may manifest and/or disappear at different stages of development.

Developmental/Behavioral Symptoms & Characteristics

- Self care assistance
- Sensory seeking behaviors
- Behavioral problems (anxiety, mood swings, etc.)
- Problems sleeping
- Tires easily
- Arm/hand flapping/other abnormal movements
- Likes strange noises
- Drooling
- Poor eye contact
- Sensory avoiding behaviors
- Self harming behaviors
- Repetitive movements and/or tics
- Short-term memory issues
- Non-ambulatory
- Hand wringing
- Echolalia

Seizures

About 1/3 of the surveyed individuals HAVE experienced seizures. The following seizure types have been observed:

- Complex
- Unknown seizure type
- Grand Mal
- Simple
- Spasm
- Absence
- Atonic
- Drop/Gelastoc
- Multifocal Generalized Epilepsy

About 2/3 of the surveyed individuals HAVE NOT experienced seizures. Of those, just under half show irregular brain activity on their EEG.

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Additional Symptoms continued:

Other Neurological Symptoms and Characteristics:

- Clumsy
- Small head size
- Cerebral Palsy
- High muscle tone (Hypertonia)
- Large head size
- Brain malformations

Vision Symptoms and Characteristics:

- Strabismus
- Nearsighted
- Nystagmus
- Farsighted
- Cortical blindness/cortical vision impairment
- Exotropia

Feeding/Gastrointestinal Symptoms and Characteristics:

- Constipation
- Reflux
- Can eat only soft/pureed food
- Chokes or gags easily
- Tube fed
- Diarrhea
- Vomits easily

Other Physical Issues:

- Difficulty gaining weight
- Hypermobility/loose joints
- Recurrent chest infections
- Scoliosis
- Short stature
- Hip dysplasia

Other Medical Diagnoses Respondents Received:

- Intellectual Disability
- Epilepsy
- Autism Spectrum Disorder
- Attention Deficit /
Hyperactivity Disorder



**This survey was created by GRIN2B Foundation's Board of Directors who are all parents of affected children and are not medical professionals. The survey was completed by 61 parents. No medical professionals contributed to the data, though the GRIN2B Foundation MAB did review the final list of symptoms. As new cases of GRIN2B-Related Neurodevelopmental Disorder are diagnosed, our knowledge of the disorder will improve. Therefore, the information included in this document is considered the best available at the time of publication. August 2019*