

## **SYNGAP1-related NSID**

### ➤ **What is SYNGAP1-related NSID?**

- SYNGAP1-related non-syndromic intellectual disability (NSID) is a condition that primarily affects the central nervous system. It is characterized by moderate to severe intellectual disability that is usually apparent in the first few years of life. Some affected people may also experience seizures and/or autism spectrum disorder and may also have attention deficits, impulsivity, and/or mood disorders.

### ➤ **What are the signs of SYNGAP1-related NSID?**

- Delayed development of speech and motor skills (sitting, standing, walking)
- Weak muscle tone (hypotonia)
- Developmental regression
- Hyperactivity
- Autism Spectrum Disorder
- Recurrent seizures (epilepsy)

### ➤ **What is SYNGAP1-related NSID a result of?**

- A mutation in the SYNGAP1 gene. The protein from the SYNGAP1 gene (SynGAP) plays an important role in nerve cells in the brain. It is found at the junctions between nerve cells (synapses) and it helps regulate changes in the synapses that are critical for learning and memory.
- SYNGAP1 gene mutations prevent the production of functional SynGAP protein from one copy of the gene, reducing the protein's activity in cells. Studies show that a reduction of SynGAP activity can have multiple effects in nerve cells, including pushing synapses to develop too early.
- The resulting abnormalities disrupt the synaptic changes in the brain that underlie learning and memory, leading to cognitive impairment and other neurological problems (SYNGAP1-related intellectual disability)

### ➤ **What are some complications of SYNGAP1-related NSID?**

- Decreased muscle tone (hypotonia)
- Movement disorders, ataxia or tremor
- Behavior disorders, including aggression, inattention, impulsivity, hyperactivity or features of autism spectrum disorder
- High pain thresholds
- Eating difficulties and sleeping problems

Resources/References:

<https://rarediseases.info.nih.gov/diseases/12558/syngap1-related-non-syndromic-intellectual-disability>

<https://medlineplus.gov/genetics/condition/syngap1-related-intellectual-disability/>

<https://www.chop.edu/conditions-diseases/syngap1-related-disorders>