



SYNGAP1-RELATED INTELLECTUAL DISABILITY

TREATMENT OF MANIFESTATIONS:

DD/ID are managed as per standard practice. No guidelines are available regarding choice of specific antiepileptic drugs (AEDs).

In about 50% of patients, the epilepsy responds to a single antiepileptic drug (AED); in the remainder it is pharmacoresistant.

CHILDREN MAY QUALIFY FOR AND BENEFIT FROM INTERVENTIONS USED IN TREATMENT OF ASD

Consultation with a developmental pediatrician may guide parents through appropriate behavioral management strategies and/or provide prescription medications when necessary. Nasogastric/gastrostomy feeding may be required for individuals with persistent feeding issues.

SUGGESTIVE FINDINGS

SYNGAP1-related intellectual disability (SYNGAP1-ID) should be considered in individuals with developmental delay or intellectual disability with or without:

Generalized epilepsy;
and/or
Autism spectrum disorder (ASD)

DEVELOPMENTAL DELAY AND INTELLECTUAL DISABILITY

The great majority of affected children present with developmental delay or intellectual disability that is typically moderate to severe but can be mild. Early motor development is characterized by hypotonia.

The average age at walking was 26 months (range: 10.5 months to 5 years). A subset of these children had an ataxic gait that remained stable or improved over time.

DEVELOPMENTAL DISABILITY / INTELLECTUAL DISABILITY EDUCATIONAL ISSUES

AGES 0-3 YEARS

Referral to an early intervention program is recommended for access to occupational, physical, speech, and feeding therapy.

In the US, early intervention is a federally funded program available in all states.

In the US, an IEP based on the individual's level of function should be developed by the local public school district. Affected children are permitted to remain in the public school district until age 21.

Discussion about transition plans including financial, vocation/employment, and medical arrangements should begin at age 12 years.

Developmental pediatricians can provide assistance with transition to adulthood.

AGES 3-5 YEARS

In the US, developmental preschool through the local public school district is recommended.

Before placement, an evaluation is made to determine needed services and therapies and an individualized education plan (IEP) is developed.

AGES 5-21 YEARS

AUTISM SPECTRUM DISORDER (ASD) AND OTHER BEHAVIORAL ABNORMALITIES.

The occurrence of ASD could be as high as 50%. This includes stereotypic behaviors such as hand flapping, obsessions with certain objects, and poor social development. In addition, inattention, impulsivity, self-directed and other-directed aggressive behavior, elevated pain threshold, hyperacusis, and sleep disorders have been observed.

OTHER ASSOCIATED FEATURES INCLUDE THE FOLLOWING:

Acquired microcephaly observed in a minority of affected individuals

Eye abnormalities including strabismus

Musculoskeletal disorders including hip rotation or dysplasia, kyphoscoliosis, and pes planus Hypertrichosis (predominantly on the limbs and lower spine) occasionally described

Gastrointestinal dysfunction (including constipation requiring medical intervention) frequently reported; swallowing difficulties rarely reported

Craniofacial features. Although some authors have suggested a subtle but consistent facial appearance (almond-shaped palpebral fissures, mildly myopathic and open-mouthed appearance) [Parker et al 2015], it is unclear if these changes are distinct enough to allow a clinician to suspect the condition in a child.



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