

Opis choroby *

Definicja

Intellectual disability-strabismus syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by moderate to severe intellectual disability and esotropia. Other associated features may include growth failure (underweight, failure to thrive, short stature), microcephaly, tone abnormalities (hypotonia, spasticity), epilepsy, behavioral problems (hyperactivity, aggressiveness), and/or abnormal brain morphology, including arachnoid cyst, cerebral atrophy, mild ventriculomegaly, abnormal CNS myelination or corpus callosum agenesis.

Dane

Klasyfikacja

Choroba

Kod ORPHA

363528

Kod OMIM

615286

Kod ICD10

H50.8

Kod ICD11

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*Źródło

orphanet