Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by infantile or childhood onset of mild to profound developmental delay and intellectual disability in all affected individuals, as well as variable occurrence of epilepsy, autism spectrum disorder / behavioral issues, microcephaly, muscle tone abnormalities such as hypotonia and spasticity, dystonic, dyskinetic, or choreiform movement disorder, and cortical visual impairment. Brain MRI may reveal abnormal cortical development, hypoplastic corpus callosum, enlarged/dysplastic basal ganglia, and hippocampal dysplasia.

Dane

Klasyfikacja
Choroba

Synonimy
Związane z GRIN2B opóźnienie rozwoju, niepełnosprawność intelektualna i zaburzenia ze spektrum autyzmu

Kod ORPHA
589547

Kod OMIM
613970

Kod ICD10
G93.4

Kod ICD11
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*Źródło

orphanet