Opis choroby *

Definicja

A rare, genetic, slowly progressive neurodegenerative disease resulting from MGLUR1 deficiency characterized by global developmental delay (beginning in infancy), mild to severe intellectual deficit with poor or absent speech, moderate to severe stance and gait ataxia, pyramidal signs (e.g. hyperreflexia) and mild dysdiadochokinesia, dysmetria, tremors, and/or dysarthria. Oculomotor signs, such as nystagmus, strabismus, ptosis and hypometric saccades, may also be associated. Brain imaging reveals progressive, generalized, moderate to severe cerebellar atrophy, inferior vermian hypoplasia, and/or constitutionally small brain.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Autosomal recessive congenital cerebellar ataxia due to metabotropic glutamate receptor 1 deficiency
Autosomalna recesywna ataksja móżdżkowo-rdzeniowa typu 13
Autosomalna recesywna wrodzona ataksja móżdżkowa z powodu niedoboru receptora 1 metabotropowego glutaminianu
SCAR13
Autosomal recessive spinocerebellar ataxia type 13
SCAR13

Kod ORPHA
324262

Kod OMIM
614831

Kod ICD10
G11.1

Kod ICD11
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*Źródło

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