Opis choroby *

Definicja

A rare autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome characterized by early-childhood onset of cerebellar ataxia associated with generalized tonic-clonic epilepsy and psychomotor development delay, dysarthria, gaze-evoked nystagmus and learning disability. Other features in some patients include upper motor neuron signs with leg spasticity and extensor plantar responses, and mild cerebellar atrophy on brain MRI.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive spinocerebellar ataxia type 12
Autosomalna recesywna ataksja rdzeniowo-móżdżkowa-12
SCAR12
SCAR12

Kod ORPHA
284282

Kod OMIM
614322

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet