Opis choroby *

Definicja

Spectrin-associated autosomal recessive cerebellar ataxia is a rare, genetic neurological disease, due to SPTBN2 mutations, characterized by global development delay in infancy, followed by childhood-onset gait ataxia with limb dysmetria and dysdiadochokinesia, mild to severe intellectual disability, development of cerebellar atrophy, and abnormal eye movements (including a convergent squint, hypometric saccades, jerky pursuit movements and incomplete range of movement).

Dane

Klasyfikacja
Choroba

Synonimy
Ataxie spinocérébelleuse à début infantile avec retard psychomoteur
Autosomalna recesywna ataksja móżdżkowa - defekt poznawczy
Autosomalna recesywna ataksja móżdżkowa związana ze spektryną typu 1
SPARCA
SPARCA1
Autosomal recessive spinocerebellar ataxia type 14
Infantile-onset spinocerebellar ataxia-psychomotor delay syndrome
SCAR14
SPARCA
SPARCA1
Spectrin-associated autosomal recessive cerebellar ataxia type 1

Kod ORPHA
352403

Kod OMIM
615386

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet