

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 mózdkowa - defekt poznawczy

Autosomalna recesywna ataksja mózdkowa 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](#)

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