

Opis choroby *

Definicja

A rare autosomal recessive cerebellar ataxia characterized by early onset of slowly progressive cerebellar atrophy, clinically manifesting with extremity and truncal ataxia, global developmental delay, intellectual impairment, nystagmus, dysarthria, intention tremor, and pyramidal signs, among others.

Dane

Klasyfikacja

Choroba

Synonimy

SCAR17

Ataksja rdzeniowo-mózdkowa autosomalna
recesywna typu 17

SCAR17

Spinocerebellar ataxia autosomal recessive type
17

Kod ORPHA

453521

Kod OMIM

616127

Kod ICD10

G11.1

Kod ICD11

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*Źródło

orphanet