

Opis choroby *

Definicja

An extremely rare, autosomal recessive, hereditary cerebellar ataxia disorder characterized by early onset of progressive, mild to moderate gait and limb ataxia, moderate to severe dysarthria, and nystagmus or saccadic pursuit, frequently associated with epilepsy, moderate intellectual disability, delayed speech acquisition, and hyporeflexia in the upper extremities. Hyperreflexia in the lower extremities may also be associated.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive spinocerebellar ataxia type
15
Autosomalna recesywna ataksja rdzeniowo-
mózdkowa typu 15
SCAR15
SCAR15
Salih ataxia

Kod ORPHA

404499

Kod OMIM

615705

Kod ICD10

G11.1

Kod ICD11

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[*Źródło](#)

orphanet