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-móżdżkowa typu 15
SCAR15
SCAR15
Salih ataxia

Kod ORPHA
404499

Kod OMIM
615705

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet