Opis choroby *

Definicja

A rare, genetic, autosomal recessive cerebellar ataxia disease characterized by nonprogressive cerebellar ataxia, with onset in infancy, manifesting with delayed motor and speech development, gait ataxia, dysmetria, hypotonia, increased deep tendon reflexes, and dysarthria. Additional variable manifestations include moderate nystagmus on lateral gaze, mild spasticity, intention tremor, short stature and pes planus. Brain imaging reveals cerebellar vermis atrophy.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive spinocerebellar ataxia type 6
Autosomalna recesywna ataksja rdzeniowo-móżdżkowa-6
SCAR6
SCAR6

Kod ORPHA
284332

Kod OMIM
608029

Kod ICD10
G11.0

Kod ICD11
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*Źródło

orphanet