Opis choroby *

Definicja

A rare, autosomal recessive, congenital, cerebellar ataxia disorder characterized by hypotonia from birth, marked psychomotor delay and prominent cerebellar dysfunction (manifesting with nystagmus, intention tremor, dysarthria, ataxic gait and truncal ataxia), described in an isolated population of the Grand Cayman Island. Cerebellar hypoplasia, observed on CT scan, may be associated.

Dane

Klasyfikacja
Choroba

Synonimy
Cayman ataxia
Ataksja Caymana

Kod ORPHA
94122

Kod OMIM
601238

Kod ICD10
G11.0

Kod ICD11
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*Źródło

orphanet