

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