

## 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

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