Opis choroby *

Definicja

A rare disorder characterised by a slowly progressive pure cerebellar ataxia associated with dysarthria. It has been described in 53 individuals from 26 families of Canadian origin. The mode of transmission is autosomal recessive. Positional cloning has led to the identification of several <i<SYNE1</i> gene mutations.

Dane

Klasyfikacja
Choroba

Synonimy
ARCA1
ARCA1
Autosomalna recesywna ataksja móżdżkowa typu 1
SCAR8
Autosomal recessive cerebellar ataxia type 1
SCAR8

Kod ORPHA
88644

Kod OMIM
610743

Kod ICD10
G11.2

Kod ICD11
-

---

*Źródło

orphanet