Opis choroby *

Definicja

A rare hereditary ataxia characterized by an early onset symptomatic generalized epilepsy, progressive cerebellar ataxia resulting in significant difficulties to walk or wheelchair dependency, and intellectual disability.

Dane

Klasyfikacja
Choroba

Synonimy
SCAR23
Spinocerebellar ataxia autosomal recessive type 23
SCAR23
Spinocerebellar ataxia autosomal recessive type 23

Kod ORPHA
404493

Kod OMIM
616949

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet