Opis choroby *

Definicja

A rare subtype of autosomal dominant cerebellar ataxia type 1 (ADCA type 1) characterized by the onset in infancy of cerebellar ataxia, neonatal hypotonia (in some), mild developmental delay and, in later life, intellectual disability. Less common features include dysarthria, dysmetria and dysmorphic facial features (long face, bulbous nose long philtrum, thick lower lip and pointed chin).

Dane

Klasyfikacja
Choroba

Kod ORPHA
314647

Kod OMIM
614756

Kod ICD10
G11.0

Kod ICD11
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*Źródło

orphanet