Opis choroby *

Definicja

A rare hereditary ataxia characterized by unusual facies (i. e. gross, rough and abundant hair, mild palpebral ptosis, thick lips, and down-curved corners of the mouth), dysarthria, delayed psychomotor development, scoliosis, foot deformities, and ataxia. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja
Choroba

Kod ORPHA
1185

Kod OMIM
271270

Kod ICD10
G11.8

Kod ICD11
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*Źródło

orphanet