Opis choroby *

Definicja

An autosomal dominant cerebellar ataxia type I that is characterized by papulosquamous, ichthyosiform plaques on the limbs appearing shortly after birth and later manifestations including progressive ataxia, dysarthria, nystagmus and decreased reflexes.

Dane

Klasyfikacja
Choroba

Synonimy
Erythrokeratodermia with ataxia
Ataksja rdzeniowo-móżdżkowa z erytrokeratodermią
Erytrokeratodermia z ataksją
SCA34
SCA34
Spinocerebellar ataxia and erythrokeratodermia

Kod ORPHA
1955

Kod OMIM
133190

Kod ICD10
G11.1

Kod ICD11
8A03.16

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*Źródło

orphanet