

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-mózdkowa z

erythrokeratodermią

Erythrokeratodermia z ataksją

SCA34

SCA34

Spinocerebellar ataxia and erythrokeratodermia

Kod ORPHA

1955

Kod OMIM

133190

Kod ICD10

G11.1

Kod ICD11

8A03.16

*Źródło

orphanet