Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by intellectual deficit, choroideremia, horizontal nystagmus, severe myopia, acrokeratosis verruciformis-like skin abnormality, anhidrosis, and scapular winging. There have been no further descriptions in the literature since 1959.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3417

Kod OMIM
314500

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet