

Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by slowly progressive night blindness, skeletal abnormalities (sloping shoulders, joint hyperextensibility, minor radiological anomalies) and characteristic facial features (periorbital anomalies, malar flatness, retrognathia). Additional manifestations include myopia and extinguished electroretinograms. There have been no further descriptions in the literature since 1979.

Dane

Klasyfikacja

Zespół wad wrodzonych Hunter-Thompson-Reed syndrome
Zespół Huntera, Thompsona i Reeda

Synonimy

Kod ORPHA

1390

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet