Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphyc 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

Synonimy
Hunter-Thompson-Reed syndrome
Zespół Huntera, Thompsona i Reeda

Kod ORPHA
1390

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet