Opis choroby *

Definicja

A rare syndrome characterised by mesomelic shortening and bowing of the limbs, camptodactyly, skin dimpling and cleft palate with retrognathia and mandibular hypoplasia. It has been described in a brother and sister born to consanguineous parents. Transmission is autosomal recessive.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mesomelic dysplasia, Kozlowski-Reardon type
Zespół Reardona, Halla i Slaney'a
Mesomelic dysplasia, Reardon type
Reardon-Hall-Slaney syndrome

Kod ORPHA
2631

Kod OMIM
249710

Kod ICD10
Q78.8

Kod ICD11
LD24.A

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*Źródło

orphanet