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 Synonimy

Zespół wad wrodzonych Mesomelic dysplasia, Kozlowski-Reardon type

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

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 2631
 249710
 Q78.8

Kod ICD11 LD24.A

*Źródło

orphanet