Opis choroby *

Definicja

Camptodactyly syndrome, Guadalajara type 2 is an extremely rare multiple congenital anomaly syndrome characterized by distinctive intrauterine growth retardation, skeletal dysplasia with multiple malformations including camptodactyly of all fingers, bilateral hallux valgus, short second, fourth and fifth toes, hypoplastic patella, microcephaly, low-set ears, short neck, cuboid-shaped vertebral bodies, pectus excavatum, hip dislocation, and hypoplastic pubic region and genitalia. Camptodactyly syndrome, Guadalajara type 2 has been described in two sisters and is most likely transmitted in an autosomal recessive manner. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1326

Kod OMIM
211920

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet