Opis choroby *

Definicja

Phocomelia-ectrodactyly-deafness-sinus arrhythmia syndrome is characterised by phocomelia (involving arms more severely), ectrodactyly, ear anomalies (bilateral anomalies of the pinnae), conductive deafness, dysmorphism (long and prominent philtrum, mild maxillary hypoplasia) and sinus arrhythmia. It has been described in four patients (a father and his son and a mother and her daughter) from two unrelated families.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Phocomelia-ectrodactyly-hearing loss-sinus arrhythmia syndrome
Zespół Stolla, Lévy'ego i Francforta
Stoll-Lévy-Francfort syndrome

Kod ORPHA
2878

Kod OMIM
171480

Kod ICD10
Q87.2

Kod ICD11
LD2H.Y

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*Źródło

orphanet