

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

Kod ORPHA	Kod OMIM	Kod ICD10
2878	171480	Q87.2

Kod ICD11
LD2H.Y

*Źródło

orphanet