

Opis choroby *

Definicja

A rare dysostosis with brachydactyly characterized by variable combinations of features of brachydactyly types A2 (such as delta-shaped middle phalanx of the second finger or toe) and D (short, broad distal phalanx of the thumb) and other types of brachydactyly (sympalangism), as well as unique features (dislocatable thumbs, lateral deviation of second toes with elevation of first toes). There have been no further descriptions in the literature since 1989.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Brachydactyly, Smorgasbord type Brachydaktylia, typ Smorgasborda

Kod ORPHA	Kod OMIM	Kod ICD10
93397	-	Q73.8

Kod ICD11
LD26.1

*Źródło

orphanet