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 (symphalangism), 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