Symbrachydaktylia dłoni i stóp
Kod Orpha: 1570 Kod OMIM:

Opis choroby *

Definicja
Symbrachydactyly of hands and feet is a rare, non-syndromic limb reduction defect disorder characterized by unilateral or bilateral brachydactyly, cutaneous syndactyly and global hypoplasia of the hand and/or foot, with underlying muscles, tendons, ligaments and bones being affected but without other associated limb anomalies. Patients typically present short, stiff, webbed or missing fingers and/or toes which are often replaced with small stumps (nubbins) with residual nails.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
De Smet-Fabry-Fryns syndrome
Zespół De Smeta, Fabry'ego i Frynsa

Kod ORPHA
1570

Kod OMIM
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Kod ICD10
Q73.8

Kod ICD11
LB75.2

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.