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 wrodzonyc	Synonimy h De Smet-Fabry-Fryns syndrome Zespół De Smeta, Fabry'ego i Frynsa		
Kod ORPHA 1570	Kod OMIM -	Kod ICD10 Q73.8	
Kod ICD11 LB75.2			
<u>*Źródło</u>			
orphanet			