Opis choroby *

Definicja

Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome is a rare, genetic, congenital limb malformation syndrome characterized by bilateral short broad thumbs, short deviated index fingers, clinodactyly of the fifth fingers, broad, valgus-deviated halluces and laterally-deviated, overlapping second toe, associated with severe pectus excavatum and craniofacial dysmorphism (including brachycephaly, low anterior hairline, flat supraorbital ridges, telecanthus, upslanting palpebral fissures, maxillary hypoplasia, posteriorly rotated ears, microsomia and micrognathia). Radiological findings include thumb, index, and middle finger hyperphalangy, with severe delta phalanxes in affected fingers and halluces.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
369979

Kod OMIM
-

Kod ICD10
Q87.2

Kod ICD11
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*Źródło

orphanet