Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Finger hyperphalangy-toe anomalies-severe pectus excavatum syndrome. ORPHA:369979

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.