Cryptomicrotia-brachydactyly-excess fingertip arch syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cryptomicrotia-brachydactyly-excess fingertip arch syndrome. ORPHA:1547

Cryptomicrotia - brachydactyly - excess fingertip arch syndrome describes a combination of malformations that include bilateral cryptomicrotia, brachytelomesophalangy with short middle and distal phalanges of digits 2 through 5, hypoplastic toenails and excess fingertip arch patterns, and has been reported in one family (mother and son). Cryptomicrotia - brachydactyly - excess fingertip arch syndrome is thought to follow an autosomal dominant transmission. There have been no further descriptions in the literature since 1988.