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Thumb stiffness-brachydactylyintellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Thumb</u> stiffness-brachydactyly-intellectual disability syndrome. ORPHA:1078

A rare, genetic, congenital limb malformation syndrome characterized by bilateral thumb ankylosis, type A brachydactyly and mild to moderate intellectual disability. Patients present thumb stiffness and abnormalities of the metacarpal bones, frequently associated with mild facial dysmorphism and signs of obesity. There have been no further descriptions in the literature since 1990.

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