

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Piussan-Lenaerts-Mathieu syndrome Zespół Piussana, Lenaerts i Mathieu

Kod ORPHA	Kod OMIM	Kod ICD10
1078	188201	Q87.2

Kod ICD11

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*Źródło

orphanet