Opis choroby *

Definicja

A rare primary bone dysplasia disorder characterized by brachymesophalangy with mesomelic short limbs, and carpal and tarsal bone abnormalities. In general, the affected individuals are of slightly short stature and normal intelligence. The syndrome has been described in a kindred with seven affected members from three generations. Transmission appears to be autosomal dominant.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Osebold-Remondini syndrome
Zespół Osebolda i Remondiniego

Kod ORPHA
93382

Kod OMIM
112910

Kod ICD10
Q73.8

Kod ICD11
LD26.1

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*Źródło

orphanet