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 Synonimy

Zespół wad wrodzonych Osebold-Remondini syndrome

Zespół Osebolda i Remondiniego

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 93382
 112910
 Q73.8

Kod ICD11 LD26.1

*Źródło

orphanet