Opis choroby *

Definicja

A rare genetic disease characterized by the association of unilateral or bilateral short fifth metacarpals (defined as a gap of 2 mm or more between the distal end of the fifth metacarpal bone and a tangential line connecting the distal ends of the third and fourth metacarpals), insulin resistance, and spherocytosis. Familial short stature has not been reported as part of the syndrome.

Dane

Klasyfikacja
Choroba

Kod ORPHA
66518

Kod OMIM
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Kod ICD10
E34.8

Kod ICD11
5A44

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*Źródło

orphanet