Opis choroby *

Definicja

A rare, genetic primary bone dysplasia of the spondylo-epi-metaphyseal dysplasia (SEMD) group characterized by progressive short-trunked dwarfism, protruding sternum, microcephaly, intellectual disability and pathognomonic radiological findings (generalized platyspondyly with double-humped end plates, irregularly ossified femoral heads, a hypoplastic odontoid, and a lace-like appearance of iliac crests)

Dane

Klasyfikacja
Choroba

Kod ORPHA
239

Kod OMIM
304950

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet