

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

*Źródło

orphanet