Choroba Dyggve, Melchiora i Clausena
Kod Orpha: 239 Kod OMIM: 304950

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.