Opis choroby *

Definicja

A rare, genetic, spondyloepimetaphyseal dysplasia disease characterized by short-limbed short stature (more pronounced in lower limbs) associated with characterisitic facial dysmorphism (i.e. relative macrocephaly, frontal bossing, midface hypoplasia, depressed nasal root, small upturned nose, prognathism) and abnormal radiological findings, which include abnormal vertebral bodies (particularly in the lumbar region), striated metaphyses, generalized mild osteoporosis, and delayed ossification of the carpal bones. Progressive coxa vara, short dental roots, hypogammaglobulinemia and cataracts may be occasionally associated.

Dane

Klasyfikacja
Choroba

Synonimy
Spondylar and nasal changes with striations of the metaphyses (SPONASTRIME) dysplasia
Dysplazja kręgowo-nasadowo-przynasadowa, typ Sponastrime
Spondyloepimetaphyseal dysplasia, Sponastrime type

Kod ORPHA
93357

Kod OMIM
271510

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet