Opis choroby *

Definicja

Spondylo-megaepiphyseal-metaphyseal dysplasia is a rare, genetic primary bone displasia characterized by disproportionate short stature with short, stiff neck and trunk and relatively long limbs, fingers and toes (which may present flexion contractures), severe vertebral body ossification delay (with frequent kyknodysostosis), markedly enlarged round epiphyses of the long bones, absent ossification of pubic bones and multiple pseudoepiphyses of the short tubular bones in hands and feet. Neurological manifestations resulting from cervical spine instability may be observed.

Dane

Klasyfikacja
Choroba

Kod ORPHA
228387

Kod OMIM
613330

Kod ICD10
Q77.7

Kod ICD11
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*Źródło

orphanet