Opis choroby *

Definicja

Smith-McCort dysplasia (SMC) is a rare spondylo-epi-metaphyseal dysplasia characterized by the clinical manifestations of coarse facies, short neck, short trunk dwarfism with barrel-shaped chest and rhizomelic limb shortening, as well as specific radiological features (i.e. generalized platyspondyly with double-humped vertebral end plates and iliac crests with a lace-like appearance) and normal intelligence. The clinical and skeletal features are similar to those seen in the allelic disorder Dyggve-Melchior-Clausen syndrome (DMC; see this term), but can be distinguished from this syndrome by the absence of intellectual deficiency and microcephaly in SMC.

Dane

Klasyfikacja
Choroba

Kod ORPHA
178355

Kod OMIM
615222

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet