

Opis choroby *

Definicja

Spondyloepiphyseal dysplasia, Cantu type is an extremely rare type of spondyloepiphyseal dysplasia (see this term) described in about 5 patients to date and characterized by clinical signs including short stature, peculiar facies with blepharophimosis, upward slanted eyes, abundant eyebrows and eyelashes, coarse voice, and short hands and feet (brachymetacarpalia, brachymetatarsalia and brachyphalangia).

Dane

Klasyfikacja

Choroba

Synonimy

SED-BDS

Dysplazja kręgowo-nasadowa - brachydaktylia -
zaburzenia mowy

Dysplazja Tattoo

SED-BDS

Zespół Fantasy Island

Tattoo dysplasia

Kod ORPHA

163654

Kod OMIM

611717

Kod ICD10

Q77.7

Kod ICD11

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[*Źródło](#)

orphanet