

Opis choroby *

Definicja

Metaphyseal acroscyphodysplasia is an extremely rare form of metaphyseal dysplasia characterized by the distinctive radiological sign of cone-shaped upper tibial and lower femoral epiphyses embedded in large cup-shaped metaphyses, associated with short stature and micromelia. Upper limb involvement includes brachydactyly and phalangeal and metacarpal cone-shaped epiphyses. The association of metaphyseal acroscyphodysplasia with psychomotor delay and alopecia has also been reported in some cases.

Dane

Klasyfikacja

Choroba

Synonimy

Bellini syndrome

Niepełnosprawność intelektualna - niski wzrost -
klinowate nasady kości kolan

Zespół Belliniego

Intellectual disability-short stature-wedge-
shaped epiphyses of knees syndrome

Kod ORPHA

1240

Kod OMIM

250215

Kod ICD10

Q78.5

Kod ICD11

LD24.7

[*Źródło](#)

orphanet