

Opis choroby *

Definicja

Metaphyseal dysplasia, Braun-Tischert type is characterised by metaphyseal undermodeling with broadening of the long bones and femora with an 'Erlenmeyer flask' appearance, expansion and bowing of the radii with severe varus deformity and flat exostoses of the long bones at the metadiaphyseal junctions.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85188

Kod OMIM

605946

Kod ICD10

Q78.5

Kod ICD11

LD24.1Y

*Źródło

orphanet