

Opis choroby *

Definicja

Ulna metaphyseal dysplasia syndrome is a rare primary bone dysplasia characterized by dysplasia of the distal ulnar metaphyses, as well as metacarpal/metatarsal dysplasia and metaphyseal changes resembling enchondromata. Patients usually present bony swelling of the wrists with or without pain (knees and ankles may also be affected). Other variably associated features include platyspondyly, skeletal development delay, short stature and coxa valga.

Dane

Klasyfikacja

Choroba

Synonimy

Rosenberg-Lohr syndrome

Zespół Rosenberga i Lohra

Kod ORPHA

1837

Kod OMIM

191420

Kod ICD10

Q78.5

Kod ICD11

LD24.7

*Źródło

orphanet