

Opis choroby *

Definicja

Otospondylomegaepiphyseal dysplasia (OSMED) is an inborn error of cartilage collagen formation characterized by sensorineural hearing loss, enlarged epiphyses, skeletal dysplasia with disproportionately short limbs, vertebral body anomalies and a characteristic facies.

Dane

Klasyfikacja

Choroba

Synonimy

OSMED

OSMED

Kod ORPHA

1427

Kod OMIM

215150

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet