

Opis choroby *

Definicja

Spondylometaphyseal dysplasia, Kozlowski type is characterized by short stature (short-trunk dwarfism), scoliosis, metaphyseal abnormalities in the femur (prominent in the femoral neck and trochanteric area), coxa vara and generalized platyspondyly.

Dane

Klasyfikacja

Choroba

Kod ORPHA

93314

Kod OMIM

184252

Kod ICD10

Q77.8

Kod ICD11

LD24.4

*Źródło

orphanet