

Opis choroby *

Definicja

Spondyloepimetaphyseal dysplasia (SEMD), Pakistani type is characterized by short stature, short and bowed lower limbs, mild brachydactyly, kyphoscoliosis, abnormal gait, enlarged knee joints, precocious osteoarthropathy, and normal intelligence.

Dane

Klasyfikacja

Choroba

Synonimy

Spondyloepimetaphyseal dysplasia, Pakistani type
Spondyloepimetaphyseal dysplasia, Pakistani type

Kod ORPHA

93282

Kod OMIM

612847

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet