

## 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  
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#### Kod ORPHA

93282

#### Kod OMIM

612847

#### Kod ICD10

Q77.7

#### Kod ICD11

LD24.3

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#### \*Źródło

orphanet