

Opis choroby *

Definicja

Spondyloepiphyseal 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 Spondyloepiphyseal dysplasia, Pakistani type Spondyloepiphyseal dysplasia, Pakistani type
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Kod ORPHA

93282

Kod OMIM

612847

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet