

Opis choroby *

Definicja

Spondyloepimetaphyseal dysplasia, Irapa type is characterized by disproportionate short-trunked short stature, pectus carinatum, short arms, short and broad hands, short metatarsals, flat and broad feet, coxa vara, genu valgum, osteoarthritis, arthrosis and moderate-to-serious gait impairment.

Dane

Klasyfikacja

Choroba

Synonimy

SEMD, Irapa type

SEMD typu Irapa

SEMD, typ Irapa

Kod ORPHA

93351

Kod OMIM

271650

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet