

Opis choroby *

Definicja

A rare lysosomal storage disease characterized by mild to severe spondylo-epiphyso-metaphyseal dysplasia, manifesting with disproportionate short stature (short neck and trunk), joint laxity, pectus carinatum, genu valgum, abnormal gait, tracheal narrowing, spinal abnormalities (kyphosis and scoliosis), respiratory impairment and valvular heart disease.

Dane

Klasyfikacja

Choroba

Synonimy

MPS4
Choroba Morquio
MPS4
MPSIV
Mukopolisacharydoza typu IV
MPSIV
Morquio disease
Mucopolysaccharidosis type IV

Kod ORPHA

582

Kod OMIM

253010

Kod ICD10

E76.2

Kod ICD11

5C56.32

[*Źródło](#)

orphanet