

## 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

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#### [\\*Źródło](#)

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