

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

Kod ORPHA	Kod OMIM	Kod ICD10
582	253010	E76.2

Kod ICD11
5C56.32

*Źródło

orphanet