

Opis choroby *

Definicja

Spondyloepiphyseal dysplasia, Maroteaux type is a very rare type of spondyloepiphyseal dysplasia (see this term) described in fewer than 10 patients to date and characterized clinically by dysplastic epiphyses, short stature appearing in infancy, short neck, short and stubby hands and feet, scoliosis, genu valgum, abnormal pelvis, osteoporosis and osteoarthritis.

Dane

Klasyfikacja

Choroba

Synonimy

Pseudo-Morquio syndrome type 2

Zespół pseudo-Morquio typu 2

Kod ORPHA

263482

Kod OMIM

184095

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet