Opis choroby *

Definicja

A relatively severe form of brachyolmia, a group of rare genetic skeletal disorders, characterized by short-trunked short stature, platyspondyly and kyphoscoliosis. Degenerative joint disease (osteoarthropathy) in the spine, large joints and interphalangeal joints becomes manifest in adulthood.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Brachyolmia type 3
Brachyolmia typu 3

Kod ORPHA
93304

Kod OMIM
113500

Kod ICD10
Q76.3

Kod ICD11
LD24.5Y

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*Źródło

orphanet