

Opis choroby *

Definicja

An exceedingly rare form of brachyolmia, characterized by mild platyspondyly, broad ilia, elongated femoral necks with coxa valga, scoliosis, and short trunked short stature associated with amelogenesis imperfecta of both primary and permanent dentition.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Platyspondyly-amelogenesis imperfecta syndrome
platyspondylia - amelogenesis imperfecta
Zespół Verloesa i Bourguignona
Verloes-Bourguignon syndrome

Kod ORPHA

2899

Kod OMIM

601216

Kod ICD10

Q76.3

Kod ICD11

LD24.5Y

*Źródło

orphanet