

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	Synonimy
Zespół wad wrodzonych	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