

## Opis choroby \*

### Definicja

Brachyolmia, recessive type is a form of brachyolmia (see this term), a group of rare genetic skeletal disorders, characterized by short-trunked short stature with platyspondyly and scoliosis. Corneal opacities and precocious calcification of the costal cartilage are rare syndromic components. Premature pubarche may occur.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Brachyolmia, Hobaek/Toledo type  
Autosomalna recesywna brachyolmia, typ  
Hobaek/Toledo

#### Kod ORPHA

448242

#### Kod OMIM

271630

#### Kod ICD10

Q76.3

#### Kod ICD11

LD24.5Y

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#### \*Źródło

orphanet