

## 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	Synonimy
Zespół wad wrodzonych	Brachyolmia, Hobaek/Toledo type Autosomalna recesywna brachyolmia, typ Hobaek/Toledo

Kod ORPHA	Kod OMIM	Kod ICD10
448242	271630	Q76.3

**Kod ICD11**  
LD24.5Y

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### \*Źródło

orphanet