## Opis choroby *

Definicja

A rare syndromic genetic deafness characterized by profound congenital bilateral sensorineural deafness, developmental delay, moderate intellectual disability, generalized delay in bone maturation, short stature, epiphyseal dysplasia particularly of the capital femoral epiphyses, and mild dysmorphic facial features such as prominent forehead and small, pointed chin. Bilateral obstruction of lacrimal ducts and inguinal and umbilical hernias have also been described.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Chitty-Hall-Baraitser syndrome
Zespół Chitty, Halla i Baraitsera
Hearing loss-epiphyseal dysplasia-short stature syndrome

**Kod ORPHA**
3218

**Kod OMIM**
601351

**Kod ICD10**
Q87.5

**Kod ICD11**
LD2H.Y

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*Źródło

orphanet