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 Synonimy

Zespół wad wrodzonych Chitty-Hall-Baraitser syndrome

Zespół Chitty, Halla i Baraitsera

Hearing loss-epiphyseal dysplasia-short stature

syndrome

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 3218
 601351
 087.5

Kod ICD11 LD2H.Y

*Źródło

orphanet