Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by congenital sensorineural hearing loss, varying degrees of intellectual disability, short stature, and dysmorphic facial features (such as telecanthus, epicanthic folds, broad nasal root, malar hypoplasia, low-set ears, dental anomalies, and micrognathia). Additional reported manifestations include microcephaly, renal and genitourinary abnormalities, widely spaced, hypoplastic nipples, and adult onset of progressive pancytopenia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hearing loss-intellectual disability syndrome, Martin-Probst type
Zespół głuchota sprzężona z chromosomem X - niepełnosprawność intelektualna
Zespół Martina i Probsta
Martin-Probst syndrome
X-linked deafness-intellectual disability syndrome syndrome
X-linked hearing loss-intellectual disability syndrome syndrome

Kod ORPHA
85321

Kod OMIM
300519

Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

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*Źródło

orphanet