Opis choroby *

Definicja

A rare deafness characterized by the association of bilateral sensorineural hearing loss and white hair with scattered black tufts, as well as skin areas of hyper- and hypopigmentation. Additional reported features include global developmental delay and moderate intellectual disability, growth retardation, microcephaly, hypotonia, mild dysmorphic facial features (deeply set eyes, broad nasal bridge, slight bowing of the upper lip), retinal depigmentation, anomalies of the fingers and toes, and white matter abnormalities on brain imaging.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
O'Doherty syndrome
Zaburzenia pigmentacji z utratą słuchu
Zespół O'Doherty
Pigmentary disorder with deafness
Pigmentary disorder with hearing loss

Kod ORPHA
999

Kod OMIM
227010

Kod ICD10
E70.3

Kod ICD11
LD2H.Y

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*Źródło

orphanet