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 Synonimy

Zespół wad wrodzonych O'Doherty syndrome

Zaburzenia pigmentacji z utratą słuchu

Zespół O'Doherty

Pigmentary disorder with deafness Pigmentary disorder with hearing loss

Kod ORPHA

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Kod OMIM

Kod ICD10 E70.3

227010

Kod ICD11 LD2H.Y

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