Opis choroby *

Definicja

A rare disorder characterised by congenital nerve deafness and piebaldness with no ocular albinism. It has been described in one large pedigree. Transmission is X-linked with affected males presenting with profound sensorineural deafness and severe pigmentary abnormalities of the skin, and carrier females presenting with variable hearing impairment without any pigmentary changes. The causative gene has been mapped to Xq26.3-q27.1.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Albinism-hearing loss syndrome

Kod ORPHA
998

Kod OMIM
300700

Kod ICD10
H90.5

Kod ICD11
LD2H.Y

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*Źródło

orphanet