

Zespół głuchoty i albinizmu

Kod Orpha: 998 Kod OMIM: 300700

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.