

# 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

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.