## 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