

Open Peer Review on Qeios

## Albinism-deafness syndrome

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Albinism-deafness syndrome</u>. ORPHA:998

Albinism-deafness syndrome (ADFN) is 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.

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