Opis choroby *

Definicja

Deafness-ear malformation-facial palsy syndrome is characterized by profound conductive deafness due to stapedial abnormalities associated with variable malformations of the external ears and facial paralysis. It has been described in three sibs and their mother. Inheritance is autosomal dominant.

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Hearing loss-ear malformation-facial palsy

syndrome

Zespół Sellarsa i Beightona Sellars-Beighton syndrome

Kod ORPHA

Kod OMIM

Kod ICD10

3232

124490

Q87.0

Kod ICD11 LD2F.1Y

*Źródło

orphanet