

Opis choroby *

Definicja

Thickened earlobes-conductive deafness syndrome is characterized by microtia with thickened ear lobes, micrognathia and conductive hearing loss due to congenital ossicular anomalies. It has been described in two families. The mode of inheritance is autosomal dominant.

Dane

Klasyfikacja

Zespół wad wrodzonych Escher-Hirt syndrome

Synonimy

Zespół Eschera i Hirta

Thickened earlobes-conductive hearing loss syndrome

Kod ORPHA

2405

Kod OMIM

128980

Kod ICD10

H90.0

Kod ICD11

LD2H.Y

*Źródło

orphanet