

Opis choroby *

Definicja

Deafness-craniofacial syndrome is characterised by the association of congenital hearing loss and facial dysmorphism (facial asymmetry, a broad nasal root and small nasal alae). It has been described in two members (father and daughter) of one Jewish family. Temporal alopecia was also noted. Transmission appeared to be autosomal dominant.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Hearing loss-craniofacial syndrome

Kod ORPHA

3241

Kod OMIM

125230

Kod ICD10

Q87.0

Kod ICD11

LD2H.Y

*Źródło

orphanet