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
Zespół wad wrodzonych

Synonimy
Hearing loss-craniofacial syndrome

Kod ORPHA
3241

Kod OMIM
125230

Kod ICD10
Q87.0

Kod ICD11
LD2H.Y

---

*Źródło

orphanet