Deafness-craniofacial syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Deafness-craniofacial syndrome. ORPHA:3241

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.