

Opis choroby *

Definicja

Conductive deafness-ptosis-skeletal anomalies syndrome is a rare, genetic ectodermal dysplasia syndrome characterized by conductive hearing loss due to atresia of the external auditory canal and the middle ear complicated by chronic infection, ptosis and skeletal anomalies (internal rotation of hips, dislocation of the radial heads and fifth finger clinodactyly). In addition, a thin, pinched nose, delayed hair growth and dysplastic teeth are associated. There have been no further descriptions in the literature since 1978.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Conductive hearing loss-ptosis-skeletal anomalies syndrome Zespół Jacksona i Barra Jackson-Barr syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
3236	221320	Q82.4

Kod ICD11
LD27.0Y

*Źródło

orphanet