

Opis choroby *

Definicja

A rare genetic disorder characterized by the triad of imperforate anus, dysplastic ears often associated with sensorineural and/or conductive hearing impairment, and thumb malformations. These features are often associated with other signs mainly affecting the kidneys and heart.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Imperforate anus-hand, foot and ear anomalies syndrome
REAR syndrome
Renal-ear-anal-radial syndrome
Sensorineural deafness with imperforate anus and hypoplastic thumbs
Sensorineural hearing loss with imperforate anus and hypoplastic thumbs
TBS
Townes syndrome

Kod ORPHA

857

Kod OMIM

617466

Kod ICD10

Q87.8

Kod ICD11

LD2F.1Y

*Źródło

orphanet