

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	Synonimy
Zespół wad wrodzonych	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