Zespół Townesa i Brocksa
Kod Orpha: 857 Kod OMIM: 617466

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

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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