Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by facial dysmorphism (hypertelorism, broad and high nasal bridge, depressed nasal ridge, short columella, underdeveloped maxilla, and prominent cupid-bow upper lip vermillion), mild to severe congenital sensorineural hearing loss, and skeletal abnormalities consisting of brachytelephalangy and broad thumbs and halluces with large, rounded epiphyses. Additional manifestations that have been reported include pulmonary valve stenosis, voice hoarseness and renal agenesis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Nasodigitoacoustic syndrome
Zespół nosowo-palcowo-akustyczny

Kod ORPHA
2662

Kod OMIM
301026

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet