Opis choroby *

Definicja

A rare multiple congenital anomaly syndrome characterized by bilateral choanal atresia associated with characteristic cranio-facial dysmorphisms (hypertelorism with narrow palpebral fissures, coloboma of inferior eyelid with presence of eyelashes medial to the defect, prominent nasal bridge, thin lips, prominent ears), that can be accompanied by hearing loss, unilateral cleft lip, preauricular tags, cardiac septal defects and anomalies of the kidneys. Affected individuals have normal intelligence.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome
Zespół Burna i McKeowna

Kod ORPHA
1200

Kod OMIM
616462

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet