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 Synonimy

Zespół wad wrodzonych Choanal atresia-hearing loss-cardiac defects-

craniofacial dysmorphism syndrome

Zespół Burna i McKeowna

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 1200
 616462
 Q87.8

Kod ICD11

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*Źródło

orphanet