

Zespół Burna i McKeowna

Kod Orpha: 1200 Kod OMIM: 616462

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.