Opis choroby *

Definicja

An extremely rare, major congenital malformation consisting of an absence of the nose ranging from hyporrhinia (absence of external nasal structures) to total arrhinia (absence of external nose, nasal airways, olfactory bulbs, or olfactory nerve) often causing respiratory distress and requiring surgical correction. Arrhinia can be bilateral or unilateral (hemiarrhinia). Associated anomalies include ocular features (hypertelorism, microphthalmia, eyelid coloboma), facial clefts, midline defects and microtia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Isolated nose agenesis
Agenezja nosa

Kod ORPHA
1134

Kod OMIM
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Kod ICD10
Q30.1

Kod ICD11
LA70.0

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*Źródło

orphanet