

Opis choroby *

Definicja

Frontorhiny is a distinct syndromic type of frontonasal malformation characterized by hypertelorism, wide nasal bridge, broad columella, widened philtrum, widely separated narrow nares, poor development of nasal tip, midline notch of the upper alveolus, columella base swellings and a low hairline. Additional features reported in some include upper eyelid ptosis and midline dermoid cysts of craniofacial structures and philtral pits or rugose folding behind the ears. An autosomal recessive inheritance has been proposed.

Dane

Klasyfikacja

Zespół wad wrodzonych ALX3-related frontonasal dysplasia

Synonimy

Dysplazja czołowo-nosowa zależna od ALX3
Izolowany zespół rozszczepu pośrodkowego
Frontonasal dysplasia type 1
Isolated median cleft face syndrome

Kod ORPHA

391474

Kod OMIM

136760

Kod ICD10

Q75.8

Kod ICD11

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*Źródło

orphanet