

Dysplazja czołowo-nosowa

Kod Orpha: 250 Kod OMIM:

Opis choroby *

Definicja

A group of rare bone development disorders characterized by an array of abnormalities affecting the eyes, forehead, and nose, and linked to midfacial dysraphia. The clinical picture is highly variable, but the major findings include hypertelorism, a broad nasal root, a large and bifid nasal tip, and widow's peak. Occasionally, abnormalities can include accessory nasal tags, cleft lip, ocular abnormalities (coloboma, cataract, microphthalmia), conductive hearing loss, basal encephalocele and/or agenesis of the corpus callosum. Intellectual deficit is rare and more likely to occur in cases where hypertelorism is severe or where there is extra-cranial involvement.

Dane

Klasyfikacja

Grupa fenomenów

Synonimy

Median cleft face syndrome
Zespół rozszczepu pośrodkowego

Kod ORPHA

250

Kod OMIM

-

Kod ICD10

Q75.8

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl