

Opis choroby *

Definicja

Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome is a rare, genetic, orofacial clefting malformation syndrome characterized by severe frontonasal dysplasia with complete cleft palate, facial cleft, extreme microphthalmia and hypertelorism, frequently associated with eyelid colobomata, sparse or absent eyelashes/eyebrows, wide nasal bridge with hypoplastic alae nasi, low-set, posteriorly rotated ears and caudal appendage in the sacral region.

Dane

Klasyfikacja

Zespół wad wrodzonych ALX1-related frontonasal dysplasia

Synonimy

Dysplazja czołowo-nosowa związana z ALX1

Frontonasal dysplasia type 3

Kod ORPHA

306542

Kod OMIM

613456

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet