Opis choroby *

Definicja

A rare syndromic frontonasal dysplasia characterized by distinctive facial dysmorphic features including hypertelorism, almond-shaped palpebral fissures, nasal deformity with creased ridge, depressed or absent tip, and asymmetry and partial absence of nasal bones, and downturned corners of the mouth. Additional reported manifestations are limb anomalies (e. g. Poland anomaly, transverse limb agenesis, and anomalies of the hands and feet, such as camptodactyly, oligodactyly, clinodactyly, and syndactyly), frontonasal encephalocele, choanal atresia, congenital renal/cardiac malformations, and corpus callosum agenesis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
521308

Kod OMIM
-

Kod ICD10
Q87.0

Kod ICD11
-

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*Źródło

orphanet