Opis choroby *

Definicja

A rare frontonasal dysplasia characterized by distinct craniofacial (large fontanelle, hypertelorism, bifid nasal tip, nasal clefting, brachycephaly, median cleft face, carp-shaped mouth), brain (interhemispheric lipoma, agenesis of the corpus callosum), and limb (tibial hypoplasia/aplasia, club foot, symmetric preaxial polydactyly of the feet and bilateral clubbed and thickened nails of halluces) malformations as well as intellectual disability. Other manifestations sometimes reported include absent olfactory bulbs, hypopituitarism and cryptorchidism.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
AFND
Zespół Toriello
AFND
Kończynowa dyzostoza czołowo-nosowa
Acromelic frontonasal dysostosis
Toriello syndrome

Kod ORPHA
1827

Kod OMIM
603671

Kod ICD10
Q75.8

Kod ICD11
LD25.3

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*Źródło

orphanet