

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 Synonimy

Zespół wad wrodzonych AFND

Zespół Toriello

AFND

Kończynowa dystroza czołowo-nosowa

Acromelic frontonasal dysostosis

Toriello syndrome

Kod ORPHA

1827

Kod OMIM

603671

Kod ICD10

Q75.8

Kod ICD11

LD25.3

*Źródło

orphanet