Acromelic frontonasal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acromelic frontonasal dysplasia. ORPHA:1827

Acromelic frontonasal dysplasia (AFND) is a rare variant of 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.