DISEASE:
Acromelic frontonasal dysplasia

NAME:	Acromelic frontonasal dysplasia
DESCRIPTION:	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.
ORPHACODE:	1827
SYNONYMS:	AFND Acromelic frontonasal dysostosis Toriello syndrome
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	ZSWIM6
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RELATED CONTENT

Related Analytes

- zinc finger SWIM-type containing 6

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