DISEASE:
Ablepharon macrostomia syndrome

NAME:	Ablepharon macrostomia syndrome
DESCRIPTION:	An extremely rare multiple congenital malformation syndrome characterized by the association of ablepharon, macrostomia, abnormal external ears, syndactyly of the hands and feet, skin findings (such as dry and coarse skin or redundant folds of skin), absent or sparse hair, genital malformations and developmental delay (in 2/3 of cases). Other reported manifestations include malar hypoplasia, absent or hypoplastic nipples, umbilical abnormalities and growth retardation. It is a mainly sporadic disorder, although a few familial cases having been reported, and it displays significant clinical overlap with Fraser syndrome.
ORPHACODE:	920
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	TWIST2
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