DISEASE:
Yunis-Varon syndrome

NAME:	Yunis-Varon syndrome
DESCRIPTION:	A rare, genetic, multiple congenital malformation syndrome, characterized by cleidocranial dysplasia (wide fontanelles, calvaria dysostosis, absent or hypoplastic clavicles), absent thumbs and halluces, hypoplastic distal and medial phalanges of fingers, pelvic dysplasia with hip dislocations. Dysmorphic features include sparse scalp hair, protruding eyes, low-set ears, anteverted nares, midfacial hypoplasia, tented upper lip, high arched palate, and micrognathia. Brain malformations are frequently associated. From birth, affected individuals tend to be significantly hypotonic and present with global developmental delay, and respiratory, feeding and swallowing difficulties.
ORPHACODE:	3472
SYNONYMS:	Cleidocranial dysplasia-micrognathia-absent thumbs syndrome
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	FIG4 VAC14
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Related Analytes

- FIG4 phosphoinositide 5-phosphatase
- VAC14 component of PIKFYVE complex