DISEASE:
Acromegaloid facial appearance syndrome

NAME:	Acromegaloid facial appearance syndrome
DESCRIPTION:	A rare multiple congenital anomalies/dysmorphic syndrome with a probable autosomal dominant inheritance, characterized by a progressively coarse acromegaloid-like facial appearance with thickening of the lips and intraoral mucosa, large and doughy hands and, in some cases, developmental delay. AFA syndrome appears to be part of a phenotypic spectrum that includes hypertrichotic osteochondrodysplasia, Cantu type and hypertrichosis-acromegaloid facial appearance syndrome.
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XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	ABCC9
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