DISEASE:
Auriculocondylar syndrome

NAME:	Auriculocondylar syndrome
DESCRIPTION:	A rare, genetic dysostosis with predominant craniofacial involvement characterized by bilateral external ear malformations, mandibular condyle hypoplasia, microstomia, micrognathia, microglossia and facial asymmetry. Additional manifestations include hypotonia, ptosis, cleft palate, full cheeks, developmental delay, hearing impairment and respiratory distress. Significant intra- and interfamilial phenotypic variation has been reported.
ORPHACODE:	137888
SYNONYMS:	Question mark ear syndrome
XREF(S):	Orphanet OMIM OMIM OMIM OMIM ICD-10 MeSH
ANALYTE(S):	GNAI3 PLCB4 EDN1

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