DISEASE:
Acrofacial dysostosis, Rodríguez type

NAME:	Acrofacial dysostosis, Rodríguez type
DESCRIPTION:	A rare, severe, multiple congenital anomalies syndrome characterized by severe mandibular hypoplasia, upper limb phocomelia with olygodactyly, absent fibula, and a number of additional skeletal (hypoplastic scapula and ischii, 11 ribs, clubfeet), facial (hypertelorism, hypoplastic supraorbital ridges, wide nasal bridge, microtia with low-set ears) and variable internal organ abnormalities (including arhinencephaly, hypolobulated lungs, and congenital cardiac defects), which usually lead to perinatal death. Surviving patients show features similar to Nagel syndrome.
ORPHACODE:	1788
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	SF3B4
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