DISEASE:
Richieri Costa-Pereira syndrome

NAME:	Richieri Costa-Pereira syndrome
DESCRIPTION:	Richieri Costa-Pereira syndrome is characterized by short stature, Robin sequence, cleft mandible, pre/postaxial hand anomalies (including hypoplastic thumbs), and clubfoot. It has been described in 14 Brazilian families and in one unrelated French patient. Prominent low set ears and a highly arched palate were also observed. Transmission is autosomal recessive.
ORPHACODE:	3102
SYNONYMS:	Short stature-Pierre Robin sequence-cleft mandible-hand anomalies clubfoot syndrome Short stature-Pierre Robin syndrome-cleft mandible-hand anomalies clubfoot syndrome
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	EIF4A3
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