DISEASE:
Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome

NAME:	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome
DESCRIPTION:	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome is characterised by the association of proximal fusion of the radius and ulna with congenital amegakaryocytic thrombocytopaenia. Less than 10 cases have been reported in the literature so far. The syndrome is transmitted as an autosomal dominant trait and is caused by mutations in the HOXA11 gene (7p15).
ORPHACODE:	71289
SYNONYMS:	ATRUS syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	MECOM HOXA11
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