DISEASE:
12q14 microdeletion syndrome

NAME:	12q14 microdeletion syndrome
DESCRIPTION:	12q14 microdeletion syndrome is characterised by mild intellectual deficit, failure to thrive, short stature and osteopoikilosis. It has been described in four unrelated patients. The syndrome appears to be caused by a heterozygous deletion at chromosome region 12q14, which was detected in three of the four patients. The deleted region contains the LEMD3 gene: mutations in this gene have already been implicated in osteopoikilosis.
ORPHACODE:	94063
SYNONYMS:	Del(12)(q14) Deletion 12q14 Monosomy 12q14 Osteopoikilosis-short stature-intellectual disability syndrome
XREF(S):	Orphanet ICD-10
ANALYTE(S):	LEMD3 HMGA2
CREATED:	13 May 2019 - 01:02
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