DISEASE:
Miller-Dieker syndrome

NAME:	Miller-Dieker syndrome
DESCRIPTION:	Miller-Dieker Syndrome (MDS) is a contiguous gene deletion syndrome of chromosome 17p13.3, characterised by classical lissencephaly (lissencephaly type 1) and distinct facial features. Additional congenital malformations can be part of the condition.
ORPHACODE:	531
SYNONYMS:	Lissencephaly due to 17p13.3 deletion Monosomy 17p13.3 Telomeric deletion 17p
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10
ANALYTE(S):	HIC1 YWHAE PAFAH1B1
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