DISEASE:
17q11 microdeletion syndrome

NAME:	17q11 microdeletion syndrome
DESCRIPTION:	17q11 microdeletion syndrome is a rare severe form of neurofibromatosis type 1 (NF1; see this term) characterized by mild facial dysmorphism, developmental delay, intellectual disability, increased risk of malignancies, and a large number of neurofibromas.
ORPHACODE:	97685
SYNONYMS:	Del(17)(q11) Monosomy 17q11 NF1 microdeletion syndrome Neurofibromatosis type 1 microdeletion syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NF1
CREATED:	13 May 2019 - 01:02
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