DISEASE:
Distal 17p13.3 microdeletion syndrome

NAME:	Distal 17p13.3 microdeletion syndrome
DESCRIPTION:	Distal 17p13.3 microdeletion syndrome is a rare partial monosomy of the short arm of chromosome 17 with a variable phenotype characterized by prenatal and postnatal growth retardation, developmental delay, mild intellectual disability, macrocephaly, mild facial dysmorphisms including prominent forehead, hypertelorism, thick upper and/or lower lip vermillion, and structural abnormalities of the brain variably including white matter abnormalities, prominent Virchow-Robin spaces, Chiari I malformation, corpus callosum hypoplasia, but no lissencephaly.
ORPHACODE:	261257
SYNONYMS:	Distal del(17)(p13.3) Distal monosomy 17p13.3
XREF(S):	Orphanet
ANALYTE(S):	YWHAE
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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- tyrosine 3-monooxygenase/tryptophan 5-monooxygenase activation protein epsilon

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