DISEASE:
Megalencephaly-capillary malformation-polymicrogyria syndrome

NAME:	Megalencephaly-capillary malformation-polymicrogyria syndrome
DESCRIPTION:	A rare developmental defect during embryogenesis that is characterized by growth dysregulation with overgrowth of the brain and multiple somatic tissues, with capillary skin malformations, megalencephaly (MEG) or hemimegalencephaly (HMEG), cortical brain abnormalities (in particular polymicrogyria), typical facial dysmorphisms, abnormalities of somatic growth with asymmetry of the body and brain, developmental delay and digital anomalies.
ORPHACODE:	60040
SYNONYMS:	MCAP MCM MCMTC Macrocephaly-capillary malformation syndrome Macrocephaly-cutis marmorata telangiectatica congenita syndrome Megalencephaly-capillary malformation syndrome Megalencephaly-cutis marmorata telangiectatica congenita syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PIK3CA

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