DISEASE:
Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome

NAME:	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome
DESCRIPTION:	A rare syndrome with a central nervous system malformation as a major feature characterized by macrocephaly, megalencephaly, bilateral perisylvian polymicrogyria, variable degrees of ventriculomegaly/hydrocephalus, developmental delay and intellectual disability, oromotor dysfunction, hypotonia, seizures, and dysmorphic facial features (such as frontal bossing, low-set ears, a flat nasal bridge, and high-arched palate). Postaxial polydactyly of one or more extremities is also common.
ORPHACODE:	83473
SYNONYMS:	MPPH syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM
ANALYTE(S):	PIK3R2 PIK3R2 CCND2 AKT3
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- Overgrowth (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- AKT serine/threonine kinase 3
- cyclin D2
- phosphoinositide-3-kinase regulatory subunit 2

Related Gene Panels

- Overgrowth (24 genes) - IPG