DISEASE:
Polymicrogyria due to TUBB2B mutation

NAME:	Polymicrogyria due to TUBB2B mutation
DESCRIPTION:	A rare, genetic, complex cerebral cortical malformation characterized by generalized or focal dysgyria (also named polymicrogyria-like cortical dysplasia) or alternatively by microlissencephaly with dysmorphic basal ganglia and dysgenesis of the corpus callosum. Clinical manifestations are variable and include microcephaly, seizures, hypotonia, developmental delay, severe psychomotor delay, ataxia, spastic diplegia or tetraplegia, and ocular abnormalities (strabismus, ptosis or optic atrophy).
ORPHACODE:	300573
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	TUBB2B
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Malformations of cortical development (235 genes)
- Polymicrogyria, asymmetric

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- tubulin beta 2B class IIb