DISEASE:
Microcephaly-capillary malformation syndrome

NAME:	Microcephaly-capillary malformation syndrome
DESCRIPTION:	Microcephaly-capillary malformation syndrome is a rare, genetic vascular anomaly characterized by severe congenital microcephaly, poor somatic growth, diffuse multiple capillary malformations on the skin, intractable epilepsy, profound global developmental delay, spastic quadriparesis and hypoplastic distal phalanges.
ORPHACODE:	294016
SYNONYMS:	MIC-CAP syndrome MIC-CM syndrome Microcephaly-cutaneous capillary malformation syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	STAMBP
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RELATED CONTENT

Related Genetic Tests

- Arteriovenous malformation (gene panel)
- Capillary malformation - arteriovenous malformation (2 genes)
- Capillary malformation – microcephaly
- Cerebral cavernous malformation (gene panel)
- Primary lymphedema / fetal hydrops (gene panel)
- Rendu-Osler-Weber disease (4 genes)
- Venous malformation (3 genes)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- STAM binding protein

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Vascular malformations (germline) (38 genes) - UCL

Source URL: http://gentest.healthdata.be/disease/1927