Microcephaly-capillary malformation syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-capillary malformation syndrome. ORPHA:294016

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.