Microcephaly-brain defect-spasticity-hypernatremia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-brain defect-spasticity-hypernatremia syndrome. ORPHA:2523

Microcephaly-brain defect-spasticity-hypernatremia syndrome is a rare congenital genetic syndrome with a central nervous system malformation as a major feature characterized by microcephaly, hypertonia, developmental delay and cognitive impairment, swallowing difficulty, hypernatremia, and hypoplasia of the frontal parts and fusion of the lateral ventricles on brain MRI. There have been no further descriptions in the literature since 1986.