Lissencephaly due to TUBA1A mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lissencephaly due to TUBA1A mutation. ORPHA:171680

Lissencephaly (LIS) due to TUBA1A mutation is a congenital cortical development anomaly due to abnormal neuronal migration involving neocortical and hippocampal lamination, corpus callosum, cerebellum and brainstem. A large clinical spectrum can be observed, from children with severe epilepsy and intellectual and motor deficit to cases with severe cerebral dysgenesis in the antenatal period leading to pregnancy termination due to the severity of the prognosis.