Lissencephaly due to LIS1 mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lissencephaly due to LIS1 mutation. ORPHA:95232

Lissencephaly due to LIS1 mutation is a cerebral malformation with epilepsy characterized predominantly by posterior isolated lissencephaly with developmental delay, intellectual disability and epilepsy that usually evolves from West syndrome to Lennox-Gastaut syndrome. Additional features include muscular hypotonia, acquired microcephaly, failure to thrive and poor control of airways leading to aspiration pneumonia.