Lissencephaly

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Lissencephaly Information Page.

Lissencephaly, which literally means "smooth brain," is a rare, gene-linked brain malformation characterized by the absence of normal convolutions (folds) in the cerebral cortex and an abnormally small head (microcephaly). In the usual condition of lissencephaly, children usually have a normal sized head at birth. In children with reduced head size at birth, the condition microlissencephaly is typically diagnosed. Lissencephaly is caused by defective neuronal migration during embryonic development, the process in which nerve cells move from their place of origin to their permanent location within the cerebral cortex gray matter. Symptoms of the disorder may include unusual facial appearance, difficulty swallowing, failure to thrive, muscle spasms, seizures, and severe psychomotor retardation. Hands, fingers, or toes may be deformed. Lissencephaly may be associated with other diseases including isolated lissencephaly sequence, Miller-Dieker syndrome, and Walker-Warburg syndrome. Sometimes it can be difficult to distinguish between these conditions clinically so consultation with national experts is recommended to help ensure correct diagnosis and possible molecular testing.