Polymicrogyria due to TUBB2B mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Polymicrogyria due to TUBB2B mutation. ORPHA:300573

A rare, genetic, central nervous system malformation characterized by generalized or focal polmicrogryia-like cortical dysplasia and simplified gyral pattern, or alternatively by microlissencephaly and agenesis of the corpus callosum. Clinical manifestations are variable and include microcephaly, seizures, hypotonia, developmental delay, severe psychomotor delay, ataxia, spastic diplegia or tetraplegia, and ocular abnormalities (strabismus, ptosis or optic atrophy).