Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation. ORPHA:300570

A rare, genetic, non-syndromic cerebral malformation due to abnormal neuronal migration disease characterized by the association of cortical dysplasia and pontocerebellar hypoplasia, manifesting with global developmental delay, mild to severe intellectual disability, axial hypotonia, strabismus, nystagmus and, occasionally, optic nerve hypoplasia. Brain imaging reveals variable malformations, including frontally predominant microgyria, gyral disorganization and simplification, dysmorphic and hypertrophic basal ganglia, cerebellar vermis dysplasia, brainstem/corpus callosum hypoplasia, and/or olfactory bulbs agenesis.