Microcephaly-thin corpus callosum-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-thin corpus callosum-intellectual disability syndrome. ORPHA:397951

Microcephaly-thin corpus callosum-intellectual disability syndrome is a rare, genetic, syndromic intellectual disability disease characterized by progressive postnatal microcephaly and global developmental delay, as well as moderate to profound intellectual disability, difficulty or inability to walk, pyramidal signs (including spasticity, hyperreflexia and extensor plantar response) and thin corpus callosum revealed by brain imaging. Ophthalmologic signs (including nystagmus, strabismus and abnormal retinal pigmentation), foot deformity and genital anomalies may also be associated.