Microcephalic primordial dwarfism due to RTTN deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephalic primordial dwarfism due to RTTN deficiency. ORPHA:468631

Microcephalic primordial dwarfism due to RTTN deficiency is a rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by primary microcephaly, profound short stature, moderate to severe intellectual disability, global developmental delay, craniofacial dysmorphism (e.g. sloping forehead, high and broad nasal bridge) and variable brain malformations, including simplified gyration, pachygyria, polymicrogyria, reduced sulcation, dysgenesis of corpus callosum and deformed ventricles. Renal anomalies, bilateral hearing loss, multiple joint contractures, severe failure to thrive and a sacral lesion cephalad to the gluteal crease have also been reported.