Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome. ORPHA:306558

Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome is a rare, genetic, neurologic disease characterized by congenital microcephaly, severe, early-onset epileptic encephalopathy (manifesting as intractable, myoclonic and/or tonic-clonic seizures), permanent, neonatal, insulin-dependent diabetes mellitus, and severe global developmental delay. Muscular hypotonia, skeletal abnormalities, feeding difficulties, and dysmorphic facial features (including narrow forehead, anteverted nares, small mouth with deep philtrum, tented upper lip vermilion) are frequently associated. Brain MRI reveals cerebral atrophy with cortical gyral simplification and aplasia/hypoplasia of the corpus callosum.