Intellectual disability, Birk-Barel type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Intellectual disability, Birk-Barel type. ORPHA:166108

Intellectual disability, Birk-Barel type is a rare, genetic, syndromic intellectual disability characterized by congenital central hypotonia, developmental delay, moderate to severe intellectual disability and subtle dysmorphic features which evolve over time (dolichocephaly, myopathic facies, ptosis, short and broad philtrum, tented upper lip vermillion, palatal anomalies, mild micro- and/or retrognathia). Patients present reduced facial movements, lethargy, weak cry, transient neonatal hypoglycemia, severe feeding difficulties and failure to thrive. Dysphagia, particularly of solid food, asthenic body build, joint contractures and scoliosis are additional features.