Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome. ORPHA:397709

Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome is a rare, genetic, central nervous system malformation syndrome characterized by early-onset, progressive, severe cerebellar ataxia associated with progressive, moderate to severe intellecutal disability, global developmental delay, progressively coarsening facial features, relative macrocephaly and absence of seizures. Sensorineural hearing loss may be associated. Neuroimaging reveals cerebellar atrophy/hypoplasia.