Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome. ORPHA:329332

Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome is a rare, genetic congenital anomalies/dysmorphic syndrome characterized by growth failure, global developmental delay, profound intellectual disability, autistic behaviors, acquired second-degree heart block with bradycardia and vasomotor instability. Hands and feet present with long fusiform fingers, campto-clinodactyly and crowded toes while craniofacial dysmorphism includes microcephaly, broad forehead, thin eyebrows, upslanting palpebral fissures, large ears with prominent antihelix, prominent nose, long philtrum, thin upper lip vermillion and prominent lower lip. Neurological signs include hypotonia, brisk reflexes, dystonic-like movements and truncal ataxia and imaging shows cerebellar hypoplasia and simplified gyral pattern.