Open Peer Review on Qeios

Microcephaly-seizures-intellectual disability-heart disease syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-seizures-intellectual disability-heart disease syndrome. ORPHA:2519

A rare, multiple congenital anomalies/dysmorphic syndrome characterized by microcephaly, intellectual disability, seizures, and congenital heart defects (e.g. atrial/ventricular septal defect, hypoplastic aortic arch with persistent ductus arteriosus). Additional manifestations include mild hypothyroidism, skeletal abnormalities, micropenis, delayed psychomotor development, dysmorphic facial features (including epicanthus, depressed nasal bridge, prominent antitragus), and pulmonary vascular occlusive disease. There have been no further descriptions in the literature since 1989.