Multiple congenital anomalies-hypotonia-seizures syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Multiple congenital anomalies-hypotonia-seizures syndrome. ORPHA:280633

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by severe global developmental delay, hypotonia, and early-onset seizures, associated with multiple congenital anomalies, such as cardiac (e.g. patent foramen ovale, atrial septal defect, patent ductus arteriosus), genitourinary (i.e. hydrocele, renal collecting system dilatation, hydroureter, hydronephrosis, hypertrophic trabecular urinary bladder) and gastrointestinal (incl. gastroesophageal reflux, anal stenosis, imperforate anus, ano-vestibular fistula) abnormalities, as well as facial dysmorphism which includes coarse facies, a prominent occiput, bitemporal narrowing, epicanthal folds, hypertelorism, nystagmus/strabismus/wandering eyes, low-set, large ears with auricle abnormalities, depressed nasal bridge, upturned nose, long philtrum, large, open mouth with thin lips, high-arched palate, and micro/retrognathia.