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# Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome. ORPHA:314575*

Intellectual disability-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome is a rare multiple congenital anomalies/dysmorphic syndrome characterized by craniofacial dysmorphism (brachycephaly resulting from craniosynostosis, frontal bossing, downslanting palpebral fissures, large and low-set ears, depressed nasal bridge, high-arched, wide palate, thin upper lip), impaired neurological development with intellectual disability, hypotonia, pyloric stenosis, pectus excavatum, bilateral cryptorchidism and short stature.