

DISEASE:**Multiple congenital anomalies-hypotonia-seizures syndrome**

NAME:	Multiple congenital anomalies-hypotonia-seizures syndrome
DESCRIPTION:	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 abnormalities (including gastroesophageal reflux, anal stenosis, imperforate anus, ano-vestibular fistula), 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.
ORPHACODE:	280633
SYNONYMS:	Congenital disorder of glycosylation due to PIGN deficiency PIGN-CDG
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PIGN
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