Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Congenital disorder of glycosylation due to PIGN deficiency
PIGN-CDG
Congenital disorder of glycosylation due to PIGN deficiency
PIGN-CDG

Kod ORPHA
280633

Kod OMIM
614080

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet