

# Zespół mnogie wady wrodzone - hipotonia - drgawki

Kod Orpha: 280633 Kod OMIM: 614080

## 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, 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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## **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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