

## Opis choroby \*

### Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by developmental delay, neuropathic visceral dysmotility (resulting in neurogenic megacystis and sometimes chronic intestinal pseudo-obstruction syndrome), intracerebral calcifications, and dysmorphic facial features (including broad forehead, downslanted palpebral fissures, strabismus, protruding and low-set ears, and retrognathia). Microcephaly and renal abnormalities have also been reported.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

73246

#### Kod OMIM

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#### Kod ICD10

Q87.8

#### Kod ICD11

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### \*Źródło

orphanet