

## **Opis choroby \***

### Definicja

A rare, lethal, genetic, multiple congenital anomaly disorder characterized by the triad of brain malformation (mainly occipital encephalocele), large polycystic kidneys, and polydactyly, as well as associated abnormalities that may include cleft lip/palate, cardiac and genital anomalies, central nervous system (CNS) malformations, liver fibrosis, and bone dysplasia.

### Dane

#### **Klasyfikacja**

Zespół wad wrodzonych Dysencephalia splanchnocystica

Zespół Meckela i Grubera

Meckel-Gruber syndrome

#### **Kod ORPHA**

564

#### **Kod OMIM**

616258

#### **Kod ICD10**

Q61.9

#### **Kod ICD11**

LD2F.13

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#### \*Źródło

orphanet