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

Synonimy
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