Opis choroby *

Definicja

A rare genetic lethal multiple congenital anomalies/dysmorphic syndrome characterized by severe hydranencephaly and renal dysplasia or agenesis. Pregnancy is complicated by oligo- or anhydramnios, leading to features of Potter sequence (including typical facies and microretrognathia, limb contractures, talipes equinovarus, and pulmonary hypoplasia) in the fetus. Affected fetuses either die in utero or shortly after birth. Histology of the brain shows widespread presence of multinucleated neurons and glial cells.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MARCH syndrome
MARCH syndrome

Kod ORPHA
500135

Kod OMIM
236500

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet