

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	Synonimy
Zespół wad wrodzonych	MARCH syndrome
	MARCH syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
500135	236500	Q87.8

Kod ICD11

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*Źródło

orphanet