Opis choroby *

Definicja

A rare, genetic neurological disorder characterized by hydranencephaly, distinctive glomeruloid vasculopathy in the central nervous system and retina, polyhydramnios and fetal akinesia with arthrogryposis. The disorder is usually prenatally lethal. In rare reported cases that survived beyond infancy, severe intellectual and neurologic disability with seizures, microcephaly and absence of functional movements were reported.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Cerebral proliferative glomeruloid vasculopathy
	Encefaloklastyczna waskulopatia proliferacyjna
	Proliferacyjna waskulopatia kłębuszkowa mózgu
	Waskulopatia proliferacyjna i hydrocefalia/hydranencefalia
	Wodogłowie/hydranencefalia z powodu waskulopatii mózgowej
	Encephaloclastic proliferative vasculopathy
	Hydrocephaly/hydranencephaly due to cerebral vasculopathy
	Proliferative vasculopathy and hydranencephaly/hydrocephaly

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
221126	225790	Q04.8

Kod ICD11
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*Źródło

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