Opis choroby *

Definicja

Porencephaly-microcephaly-bilateral congenital cataract syndrome is a rare, genetic, central nervous system malformation syndrome characterized by bilateral congenital cataracts and severe hemorrhagic destruction of the brain parenchyma with associated massive cystic degeneration, enlarged ventricles and subependymal calcification. Patients typically present generalized spasticity, increased deep tendon reflexes and seizures. Hepatomegaly and renal anomalies have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
306547

Kod OMIM
613730

Kod ICD10
Q07.8

Kod ICD11
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*Źródło

orphanet