Opis choroby *

Definicja

A rare neurologic disease characterized by lethargy, hypotonia, poor feeding, opisthotonus, and a typical high-pitched cry due to bilirubin accumulation in the globus pallidus, sub-thalamic nuclei, and other brain regions, resulting from severe neonatal unconjugated hyperbilirubinemia. Onset of symptoms is typically within the first three to five days of life. Additional features include fever, apnea, seizures, and coma. Especially respiratory failure or refractory seizures may lead to a fatal outcome.

Dane

Klasyfikacja
Zespół kliniczny

Synonimy
ABE
Ostra żółtaczka jąder podstawy mózgu
Acute kernicterus

Kod ORPHA
529799

Kod OMIM
-

Kod ICD10
P57.8

Kod ICD11
-

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*Źródło

orphanet