Opis choroby *

Definicja

A rare pyruvate metabolism disorder characterized by neonatal onset of a mitochondrial encephalopathy with global developmental delay and the biochemical characteristics of lactic acidosis and increased serum pyruvate with normal lactate/pyruvate ratio. Additional reported manifestations include epilepsy, peripheral neuropathy, hypotonia, nystagmus, extensor plantar responses, hepatomegaly, and craniofacial dysmorphism (such as progressive microcephaly, epicanthus, long philtrum, and thin upper lip).

Dane

Klasyfikacja
Choroba

Kod ORPHA
447784

Kod OMIM
614741

Kod ICD10
E74.4

Kod ICD11
5C53.0Y

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*Źródło

orphanet