Opis choroby *

Definicja

Progressive encephalopathy with leukodystrophy due to DECR deficiency is a rare mitochondrial disease, which presents with neonatal hypotonia, central nervous system abnormalities (ventriculomegaly, corpus callosum hypoplasia, cerebellar atrophy), acquired microcephaly, failure to thrive, developmental delay and intermittent lactic acidosis provoked by catabolic stress (e.g. infection). Hyperlysinemia and elevated C10:2 carnitine can be detected in plasma. Later on, epilepsy, cerebellar ataxia, renal tubular acidosis, severe encephalopathy, dystonia, spastic quadriplegia and other complications may develop.

Dane

Klasyfikacja
Choroba

Synonimy
2,4-dienoyl-CoA reductase deficiency
Niedobór DECR z hiperlizynemią
Niedobór reduktazy 2,4-dienoilo-CoA
DECR deficiency with hyperlysinemia

Kod ORPHA
431361

Kod OMIM
616034

Kod ICD10
G31.8

Kod ICD11
-

---

*Źródło

orphanet