Opis choroby *

Definicja

Carnitine-acylcarnitine translocase (CACT) deficiency is a life-threatening, inherited disorder of fatty acid oxidation which usually presents in the neonatal period with severe hypoketotic hypoglycemia, hyperammonemia, cardiomyopathy and/or arrhythmia, hepatic dysfunction, skeletal muscle weakness, and encephalopathy.

Dane

Klasyfikacja
Choroba

Synonimy
CACT deficiency
Niedobór CACT

Kod ORPHA
159

Kod OMIM
212138

Kod ICD10
E71.3

Kod ICD11
5C52.00

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*Źródło

orphanet