

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

*Źródło

orphanet