

Opis choroby *

Definicja

A disorder of carnitine cycle and carnitine transport that is characterized classically by early childhood onset cardiomyopathy often with weakness and hypotonia, failure to thrive and recurrent hypoglycemic hypoketotic seizures and/or coma.

Dane

Klasyfikacja	Synonimy
Choroba	CDSP CDSP CUD Defekt transportera karnityny Deficyt wychwytu karnityny Niedobór osoczowo-błonowego transportera karnityny SPCD CUD Carnitine transporter defect Carnitine uptake deficiency Deficiency of plasma-membrane carnitine transporter SPCD

Kod ORPHA
158

Kod OMIM
212140

Kod ICD10
E71.3

Kod ICD11
5C52.00

*Źródło

orphanet