

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

Choroba

Synonimy

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