

## 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

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#### \*Źródło

orphanet