

Opis choroby *

Definicja

A rare, early-onset and life-threatening, multiple carboxylase deficiency that when left untreated, is characterized by vomiting, tachypnea, irritability, lethargy, exfoliative dermatitis, and seizures that can worsen to coma and death.

Dane

Klasyfikacja

Choroba

Synonimy

Early-onset multiple carboxylase deficiency
Złożony Niedobór karboksylazy noworodków
Złożony Niedobór karboksylazy o wczesnym
początku
Neonatal multiple carboxylase deficiency

Kod ORPHA

79242

Kod OMIM

253270

Kod ICD10

E53.8

Kod ICD11

5C50.E0

*Źródło

orphanet