

Opis choroby *

Definicja

Transcobalamin deficiency (TC) is a disorder of cobalamin transport that usually presents during the first few months of life and is characterized by megaloblastic anemia, failure to thrive, vomiting, weakness and pancytopenia.

Dane

Klasyfikacja

Choroba

Synonimy

Inherited deficiency of transcobalamin

Niedobór transkobalaminy II

Wrodzony Niedobór transkobalaminy

Transcobalamin II deficiency

Kod ORPHA

859

Kod OMIM

275350

Kod ICD10

D51.2

Kod ICD11

3A01.0

*Źródło

orphanet