Opis choroby *

Definicja

A rare, genetic, benign disorder of cobalamin transport, due to variable degrees of transcobalamin I deficiency, characterized by mildly low to almost undetectable plasma transcobalamin I levels and slighly low to absent serum cobalamin levels. Normal methylmalonic acid and homocysteine serum values and absence of megaloblastic anemia are reported. No specific clinical manifestations are associated and patients are typically asymptomatic.

Dane

Klasyfikacja
Choroba

Synonimy
Haptocorrin deficiency
Niedobór TCI
Niedobór haptokoryny
Niedobór transkobalaminy-1
TCI deficiency
Transcobalamin-1 deficiency

Kod ORPHA
2967

Kod OMIM
193090

Kod ICD10
E53.8

Kod ICD11
5C63.0

---

*Źródło

orphanet