

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

Kod ORPHA	Kod OMIM	Kod ICD10
2967	193090	E53.8

Kod ICD11
5C63.0

*Źródło

orphanet