

Opis choroby *

Definicja

Familial benign copper deficiency is a rare disorder of mineral absorption and transport characterized by hypocupremia that manifests as failure to thrive, mild anemia, repeated seizures, hypotonia, and seborrheic skin. Spurring of the femur and tibia are also noted on radiographic imaging. Symptoms are reversible or improve with supplements of oral copper. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja Choroba	Synonimy Familial benign hypocupremia	
Kod ORPHA 1551	Kod OMIM 121270	Kod ICD10 E83.0
Kod ICD11 -		

*Źródło

orphanet