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Severe congenital hypochromic anemia with ringed sideroblasts

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe congenital hypochromic anemia with ringed sideroblasts. ORPHA:300298*

ST EAP3/T SAP6-related sideroblastic anemia is a very rare severe non-syndromic hypochromic anemia, which is characterized by transfusion-dependent hypochromic, poorly regenerative anemia, iron overload, resembling non-syndromic sideroblastic anemia (see this term) except for increased erythrocyte protoporphyrin levels.