Sideroblastic anemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Sideroblastic anemia. ORPHA:1047

Sideroblastic anemias (SA) are a group of rare heterogeneous inherited or acquired bone marrow disorders, isolated or part of a syndrome, characterized by decreased hemoglobin synthesis, because of defective use of iron (although plasmatic iron levels may be normal or elevated) and the presence of ringed sideroblasts in the bone marrow due to the pathologic iron overload in mitochondria as visualized by Perls' staining. The group encompasses (idiopathic) acquired sideroblastic anemia and constitutional sideroblastic anemias (see these terms). The latter include syndromic sideroblastic anemias such as Pearson syndrome, mitochondrial mypathy and sideroblastic anemias, x-linked sideroblastic anemia-ataxia, thiamine responsive megaloblastic anemia syndrome and nonsyndromic sideroblastic anemias comprising x-linked and autosomal recessive sideroblastic anemias (see these terms).