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Adult-onset autosomal recessive sideroblastic anemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Adult-onset autosomal recessive sideroblastic anemia. ORPHA:255132

Adult onset autosomal recessive sideroblastic anemia or GLRX5-related sideroblastic anemia is a very rare non-syndromic autosomal recessive pyridoxine-refractory sideroblastic anemia due to a splice defect of glutaredoxin-5 (GLRX5) described in a single patient with adult onset microcytic hypochromic anemia with liver iron overload and type 2 diabetes.