ABSTRACT

Sideroblastic anemia is a rare group of anemias characterized by a reduction in heme biosynthesis due to various acquired and genetic causes, leading to ineffective erythropoiesis. The hallmark of this condition is the presence of ring sideroblasts in the bone marrow. Sideroblastic anemia can present as either microcytic or macrocytic anemias, and iron studies may show features of iron overload. Misdiagnosis can occur when sideroblastic anemia presents as microcytic hypochromic anemia, potentially leading to incorrect or delayed treatment and poor outcomes.

CASE PRESENTATION

We present the case of a 61-year-old male who presented with exertional dyspnea, lethargy, tiredness, and fatigue for two weeks. His symptoms had been progressively worsening during this period. The patient denied any significant blood loss or bleeding symptoms. He was given a diagnosis of iron deficiency anemia in the past, but upon investigation, his iron studies revealed iron overload. Bone marrow aspiration confirmed the presence of ring sideroblasts, leading to the diagnosis of sideroblastic anemia. Genetic testingcould not be performed due to limited resources.

DISCUSSION

Sideroblastic anemia encompasses a heterogenous group of anemias, including both inherited and acquired forms. The mechanism involves impaired incorporation of iron into protoporphyrin IX in the mitochondria, leading to the accumulation of iron granules and ineffective erythropoiesis. Differential diagnosis from other anemic entities is crucial to avoid inappropriate treatment. Management includes addressing reversible causes, administering vitamin B6 for responsive subtypes or thiamine, iron chelation to prevent organ damage from iron overload, and red cell transfusions for symptomatic anemia. The prognosis varies depending on the etiological cause.

CONCLUSION

Sideroblastic anemia is a complex condition that requires careful investigation to reach an accurate diagnosis. Misdiagnosis can lead to detrimental consequences, making it essential for clinicians to be vigilant and consider this rare condition when presented with microcytic hypochromic anemia. Early diagnosis and appropriate management can significantly impact patient outcomes. Genetic testing is a valuable tool for further characterizing the condition and tailoring treatment strategies. Further research and increased awareness are necessary to enhance the understanding and management of sideroblastic anemia.