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Congenital Sideroblastic Anaemia-Classic Presentation

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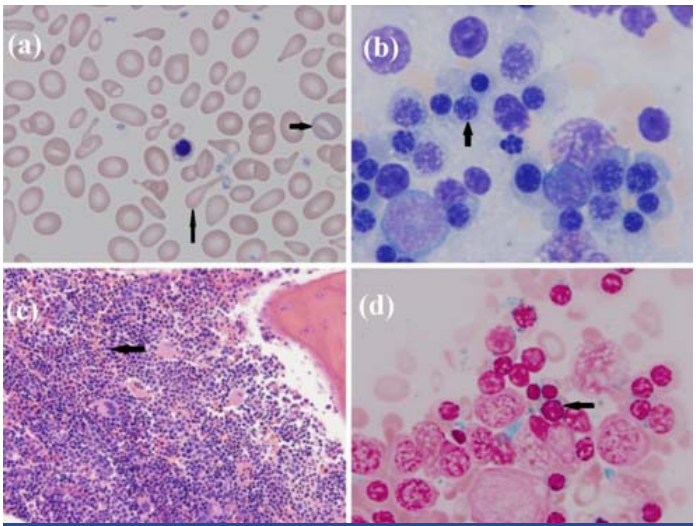
An eight-year-old girl child came to our hospital with high grade fever since four days associated with generalised weakness and lethargy. Past history was suggestive of multiple similar episodes each of 4 to 5 days duration since two years of age often accompanied with severe anaemia requiring packed cell transfusion. Symptoms used to subside with antipyretics and broad spectrum antibiotics within seven days. She was started on colchicine one year back with no significant improvement based on the diagnosis of periodic fever syndrome. Then her frequency of febrile attacks increased up to once every week. Differential diagnosis like haemolytic anaemia due to various aetiologies including autoimmune were considered. Also, anaemia due to bone marrow failure was considered as differential. Subsequently she was started on prednisolone 5mg daily after which frequency of attacks were reduced to once monthly. Child was born out of 3rd degree consanguineous marriage. Multiple male relatives including elder and younger sister had died at an early age with similar symptoms. Antenatal history of mother was suggestive of oligohydramnios. Child was born of full term normal delivery with a birth weight of 1 kg. She required packed cell transfusion immediately after birth due to severe anaemia. On examination vitals were normal but there was pallor with multiple dental caries and mild prominence of jaw. Haematological examination revealed haemoglobin of 7.9 with haematocrit 25.5, White Blood Corpuscle (WBC) 5260, platelets 269, Mean Corpuscular Volume (MCV) 57.3 and Red Blood Cell Distribution Width (RDW) 34.1. Her peripheral smear examination revealed Anisopoikilocytosis, Tear Drop cells, Basophilic stippling, Microcytosis, Hypochromatic red cells and stomatocytes [Table/Fig-1a]. Serum Lactate dehydrogenase (LDH) was 216, serum ferritin was 634, Total iron-binding capacity (TIBC) 47.8 to 51.6, serum haptoglobin 58, Erythrocyte Sedimentation Rate (ESR) 6, serum fibrinogen 248, C-reactive protein (CRP) 1.5, serum folate and vitamin B12 were 4.645 and 392 respectively. Serum copper was 27.4 and serum lead was 0.07. Ultrasonography of abdomen showed splenomegaly. Patient was evaluated for haemolytic disorder. Glucose-6-phosphate dehydrogenase (G6PD), haemoglobin electrophoresis and osmotic fragility tests were normal. Bone marrow examination revealed a hypercellular marrow with erythroid majority with maturing myelopoiesis and some evidence of dysplasia [Table/Fig-1b,c]. When stained with Prussian blue, bone marrow showed ring sideroblasts [Table/Fig-1d]. Enzyme levels of aminolevulinic acid synthase could not be estimated. A diagnosis of congenital sideroblastic anaemia was made. The patient was started on 40 mg oral pyridoxine per day. There was a rise in haemoglobin level from 7.9 to 9 in first three weeks of treatment and the patient is still under follow-up and is 10 years old now.



[Table/Fig-1]: (a) Peripheral smear evident of anisopoikilocytosis, tear drop cells (long black arrow), basophilic stippling, microcytosis, hypochromatic red cells and stomatocytes (short black arrow). **(b)** Bone marrow aspirate shows maturing myelopoiesis and may be some evidence of dysplasia (black arrow). **(c)** Bone marrow cellularity shows a packed marrow which is hypercellular with erythroid majority (black arrow). **(d)** Prussian blue stain of the bone marrow with ring sideroblasts (black arrow). Blue-stained ferritin iron deposits in the mitochondria of erythroid precursors form an apparent ring around the nucleus.

Sideroblastic anaemias constitute a group of disorders having features of microcytic hypochromic anaemia with ringed sideroblasts in mitochondria of erythroid precursors in bone marrow, increased in levels of iron in the tissues along with ineffective erythropoiesis [1]. Clinical features include extreme pallor, malaise, fatigue and dyspnea. Prior history of antibiotics, antituberculous agents and ingestion of zinc and exposure to lead pipes in houses is significant [2].

Differential diagnosis includes drug toxicity, acute myelogenous leukaemia, myelodysplastic syndrome, porphyrias and haemolytic anaemia [1,2]. Peripheral Smear examination is evident of Anisopoikilocytosis, Tear Drop cells, Basophilic stippling, Microcytosis, Hypochromatic red cells and stomatocytes. Siderocytes with Pappenheimer bodies (hypochromic erythrocytes with iron deposits which are basophilic) are mature sideroblasts in the peripheral blood [2]. Responders respond to treatment of pyridoxine supplementation. Non responders require regular blood transfusion [1,2].

A diagnosis of sideroblastic anaemia should be considered in all with unexplained anaemia from history, clinical examination, and laboratory data. A finding of microcytic hypochromic picture on peripheral smear along with the pathognomonic ring sideroblasts evident on a Prussian blue stain of the bone marrow aspirate smear helps in narrowing the differential diagnosis.

REFERENCES

[1] Ponka P, Prchal JT. Hereditary and acquired sideroblastic anaemias. In: Kaushansky K, Beutler E, Seligsohn U, Lichtman MA, Kipps TJ, Prchal JT (eds). Williams Haematology, 8th edn. McGraw Hill, New York, 2010; 865–881.
[2] Stavem P, Rørvik TO, Rootwelt K, Josefsen JO. Severe iron deficiency causing loss of ring sideroblasts. *Scand J Haematol*. 1983;31(4):389-91.

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