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Sideroblastic anemia with marked dyserythropoiesis: A rare entity

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Abstract

Congenital Sideroblastic anemia is an uncommon variety. It is characterized by the presence of ringed sideroblasts in the bone marrow. Erythroid dysplasia is usually not the feature of this disease. Congenital Dyserythropoietic Anemia (CDA) on the other hand is found to have excessive Eythroid dysplasia. We came across a very rare case of a seven month old male child who was referred to us for bone marrow examination with clinical suspicion of hematological malignancy. Complete blood picture showed bicytopenia. Bone marrow aspirate from anterior tibia revealed a hyper cellular smear with marked eryhthroid dysplasia (14%) while Iron stain showed ring sideroblasts (16%). A very few case reports were found from the literature search and it is proposed that these cases should be classified as a variant form of the disease.

Keywords

sideroblastic anemia; congenital dyserythropoietic anemia; bone marrow; hematology

Introduction

Sideroblastic anemia is characterized by accumulation of iron in mitochondria of erythroblasts, which is visible as Prussian blue positive granules. These erythroblasts are termed as ringed sideroblasts when the granules form a ring around the nucleus, covering more than one third of the circumference [1]. Presence of 15% or greater number of such ringed sideroblasts is essential for the diagnosis.

There are two forms of sideroblastic anemia; congenital and acquired. The former being very uncommon and is further divided into X-linked or autosomal pattern of mutations. Among X-linked sideroblastic anemia the most common is caused by mutations of the erythroid-specific d-aminolevulinate synthase gene (ALAS2).

Acquired sideroblastic anemia is associated with Myelodysplastic Syndromes, alcoholism and certain drugs [2].

Case Report

A seven months old male child was referred to us for bone marrow biopsy with suspicion of malignancy. He had a history of progressive pallor for the last two months and fever for the last one month. Fever was high grade and intermittent. There was no history of loss of consciousness, seizures or

bleeding from any site. His past medical and surgical history was insignificant. His parents were third degree relative and he was their fourth child. There was no family history of any hematological disorder. His other siblings were alive and healthy.

On examination, the child had severe pallor. There was no jaundice, bruising, skeletal abnormality, lymphadenopathy or hepatosplenomegaly.

Lab Findings

The child had RCC transfused twice before Bone Marrow aspiration. Pretransfusion hemoglobin was 5.2 g/dL. His complete blood picture post- transfusion showed following:

Sr no	Parameters	Patient's value	Normal Range
1	Total leucocyte count	2.5 × $10^{3/}$ µL	3.5-12.0 × $10^{3/}$ µL
2	Red Blood cell count	3.38 × $10^{6/}$ µL	4.75-4.85 × $10^{6/}$ µL
3	Hemoglobin	9.8 g/dL	12.0-14.4 g/dL
4	Hematocrit	28.10%	30.9-37 %
5	Mean corpuscular volume	77 fL	80-100 fL
6	Mean corpuscular hemoglobin	26.0 pg	25-31 pg
7	Mean corpuscular hemoglobin concentration	34.0 g/dL	32-36 g/dL
8	Platelet count	194 × $10^{3/}$ µL	150-400 × $10^{3/}$ µL

Other laboratory investigations were as follows:

1	Serum Urea	13 mg/ dL	24-40 mg/ dL
2	Serum Creatinine	0.2 mg/ dL	0.6-1.2 mg/ dL
3	Blood sugar random	142 mg/ dL	70-110 mg/ dL
4	Total Bilirubin	0.22 mg/ dL	0.2-1.3 mg/ dL
5	Serum ALT	17 IU/L	4-42 IU/L
6	Serum Alkaline phosphatase	270 IU/L	40-130 IU/L
7	Serum Iron	118 µg/ dL	20-115 µg/ dL
8	Total iron binding capacity	188 µg/ dL	275-458 µg/ dL
9	ICT MP	Negative	Negative
10	HbsAg (kit)	Negative	Negative
11	Anti HCV (kit)	Negative	Negative

Peripheral film

Peripheral film showed a dimorphic picture as the child was recently transfused. Occasional target and pencil cells were also present. Platelets were adequate on smear (Figure 1) the differentiated leucocyte count showed 27% Neutrophils, 68% Lymphocytes, 3% Monocytes and 2% Eosinophils. There were no blasts or atypical cells seen.

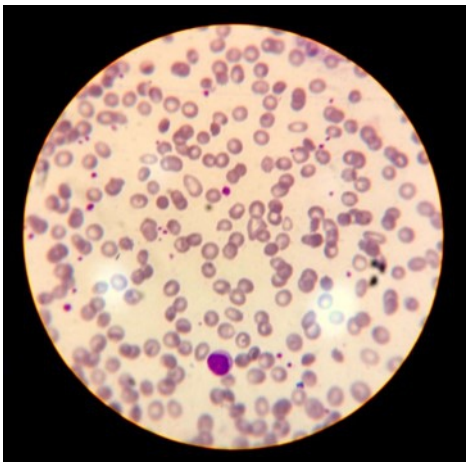

Figure 1: Peripheral film showing microcytic hypochromic anemia

Bone marrow aspirate

Bone marrow was aspirated from anterior tibial tuberosity. The smear was hypercellular as shown in Figure 2. Erythropoiesis was hypercellular with predominantly normoblasts along with marked dyserythropoiesis which was 14% (Figure 3). Dyserythropoiesis included bi and multinuclearity with some intercytoplasmic bridging and mitotic cells. Myelopoiesis was also hypercellular and all cell stages were seen. The dysplasia in the myeloid series was insignificant. Megakaryocytes were adequate with normal maturation. Lymphocytes and plasma cells were not increased nor any blasts or atypical cells were seen.

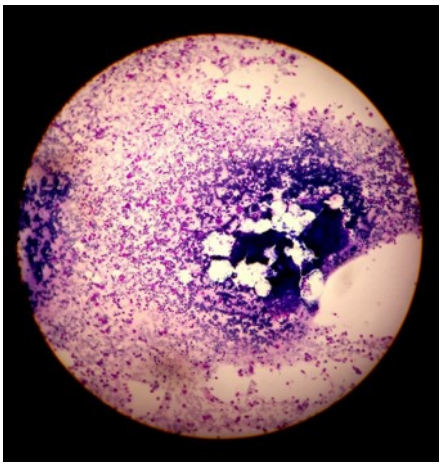

Figure 2: Bone marrow aspirate showing hypercellular fragments and trails.

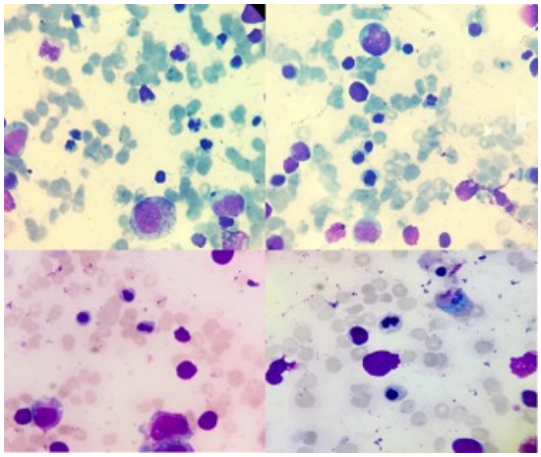

Figure 3: Bone marrow aspirate showing marked Erythroid dysplasia.

Iron stain was markedly increased with presence of ringed sideroblasts (16%), as shown in Figure 4.

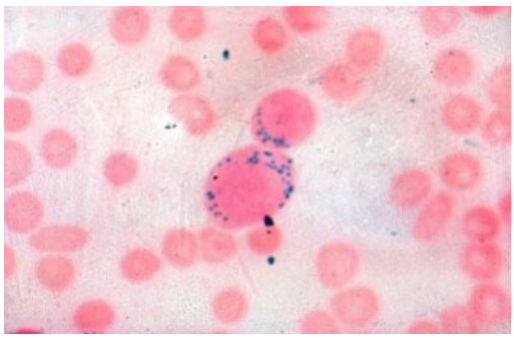

Figure 4: Ringed sideroblasts seen in Iron stain.

Discussion

We came across a very rare case of congenital sideroblastic anemia with marked dyserythropoiesis. Dyserythropoiesis is a feature of congenital dyserythropoeitic anemia, however in this case both features of dyserythropoiesis and ringed sideroblasts were present which made the diagnosis difficult. In the previous literature, only a few case reports have been found where both features were present.

Kasturi et al reported an autosomal pattern of inheritance of sideroblastic anemia in four siblings, two of them being male and the other two females. Peripheral blood showed microcytosis, hypochromia and poikilocytosis. Bone marrow examination was done in three of them which revealed erythroid hyperplasia with predominance of normoblasts. Nuclear polypoidy and binuclear cells were also present while iron stain showed numerous ringed sideroblasts [3].

Another case report by Jun et al described a case of 4 year old female child with microcytic hypochromic anemia. Bone marrow showed dyserythropoiesis along with ringed sideroblasts. A diagnosis of hereditary sideroblastic anemia was made [4].

Brien et al described a case of 19 years old female who had microcytic hypochromic anemia, not responding to treatment. Bone marrow biopsy showed Erythroid hyperplasia with marked dyserythropoiesis which included nuclear budding, karryorrhexis, multinucleation and defective haemoglobinization. Iron stores were increased with presence of ringed sideroblasts. Family study was done which revealed an autosomal recessive pattern of inheritance. Because X-linked mutation is more common in hereditary sideroblastic anemia, it was proposed by them to classify such cases as Variant Congenital Dyserythropoietic anemia with ringed sideroblast [5]. Similar views were shared by Soysal who described a case of 25 years old male with anemia jaundice and splenomegaly. Peripheral film showed anisocytosis with basophilic stippling while bone marrow examination showed erythroid hyperplasia with binuclearity and presence of ringed sideroblasts. It was proposed that such cases should be designated as variant of congenital dyserythropoietic anemia with ringed sideroblasts [6]. The case reported by Mohamed and Alfaraidy of a neonate had both findings however ringed sideroblasts were occasional [7].

Our findings of peripheral film and bone marrow examination were similar to those mentioned in the above case reports. Considering the gender of the child we labeled him as a case of sideroblastic anemia with marked Erythroid dysplasia. His mutational analysis was not done due to non-availability of the test. He was advised pyridoxine therapy and to monitor the response to the treatment.

Conclusion

Although this is a rare case presentation but requires classification as a variant form of Sideroblastic anemia. It is also necessary to follow up such cases to see the prognosis of the disease. Genetic and family studies is also required to see the pattern of inheritance.

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