Abstract: PB2029

Title: ACQUIRED ALPHA THALASSEMIA IN ASSOCIATION WITH MYELODYSPLASTIC SYNDROME- A CASE REPORT

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Session Title: Myelodysplastic syndromes - Clinical

Background:

Myelodysplastic syndrome (MDS) is a group of heterogeneous diseases derived from hematopoietic stem cells characterized by ineffective and dysplastic haematopoiesis. In HbH disease, there is deficient α-gene synthesis and the unbound β globin chain will form β-4 tetramers (HbH), which form a precipitate in red blood cells, leading to haemolytic anaemia.

Aims:

We present a case of acquired HbH in Myelodysplastic syndrome.

Methods:

A 60 years old diabetic male presented with generalized fatigue for 2 months. He had no lymphadenopathy or hepatosplenomegaly. The complete hemogram showed pancytopenia (Haemoglobin- 4.5 gm/dl, total leucocyte count - 3,670/ cu. mm and platelet count -22,000/cu.mm) and microcytic hypochromic indices. On peripheral blood smear examination, RBC showed marked anisopoikilocytosis with microcytic hypochromic RBCs, well-hemoglobinised RBCs, and few macrocytic red cells. Teardrop cells, elliptocytes, target cells, polychromatophils and few nucleated RBCs were also noted. WBCs showed dysgranulopoiesis in the form of Pelger Huet neutrophils. Platelets are mildly reduced with few giant forms. The marked anisopoikilocytosis in RBCs prompted us to do supravital staining which showed HbH inclusions in 10% of RBCs.

Results:

Bone marrow aspirate smears were hypercellular for age with marked erythroid hyperplasia with normoblastic to megaloblastic maturation. Myelopoiesis showed dysgranulopoiesis in the form of pseudo-pelgueroid neutrophils and hypogranulation. Megakaryocytes were mildly increased in number with dysmegakaryopoiesis in the form of megakaryocytes having lobe separation and micromegakaryocytes. No ring sideroblasts were noted on Perls staining

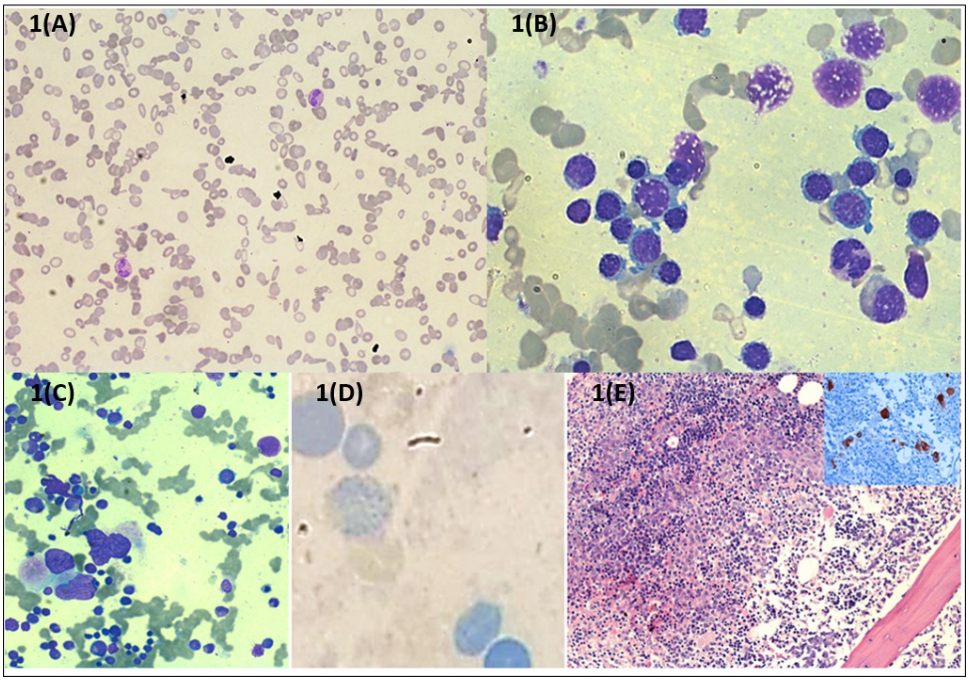
Summary/Conclusion: Acquired disorders of haemoglobin synthesis has been described in many clonal hematopoietic disorders. Acquired HbH disease is usually encountered in myelodysplastic syndromes with a strong predilection of males. Incidence of α-thalassemia myelodysplastic syndrome (ATMDS) is less than 0.5%. ATMDS is most frequently associated with somatic mutations in the *ATRX (α-thalassemia mental retardation X-linked)* gene, a trans-acting regulator of erythropoiesis.

With the absence of splenomegaly and the absence of the previous history of anaemia and blood transfusion a diagnosis of myelodysplastic syndrome with acquired α-thalassemia was considered.We present this case for its rarity, and also to emphasize the need for doing a simple stain like supravital staining for HbH inclusions so that such cases can be diagnosed.

NOTE: We have sent the sample for Whole exome sequencing to see for ATRX mutation and other mutations in this case. The results will be available soon and will be presented at the conference.

Fig 1: Peripheral smear show well-haemoglobins RBCs and microcytic hypochromic RBCs with marked anisopoikilocytosis, teardrop RBCs and elliptocytes(A). Bone marrow aspirate smear shows erythroid with megaloblastic maturation and a dyspoietic neutrophil (B). Bone marrow aspirate smear shows

dysmegakaryopoiesis with separated lobes and hypolobate megakaryocytes (C). Supravital stain on peripheral smear shows a golf ball inclusion in RBC(D). Trephine biopsy shows a hypercellular biopsy with predominantly erythroid precursors with normoblastic and megaloblastic maturation (CD61 IHC shows dysplastic megakaryocytes and micromegakaryocytes-inset) (E).



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