AN UNSUAL CASE OF SPLENOMEGALY WITH POLYARTHRITIS

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Abstract: Massive splenomegaly is defined as spleen palpable more than 8cm below the left costal margin or its drained Weight more than 1000g. Myelofibrosis is one of the rare causes of massive splenomegaly. Idiopathic myelofibrosis is being mentioned as a rare disease in the Office of Rare Diseases (ORD) by National Institute of Health (NIH). Even rarer is the presentation of myelofibrosis with polyarthritis. Extramedullary haematopoiesis involving the synovium is encountered in myelofibrosis patients with myeloid metaplasia. We encountered one such situation in a 42 year old male, who presented with abdominal fullness, polyarthritis, pruritic rash and weight loss, which later turned out to be a case of myelofibrosis with polyarthritis.

Keyword: MYELOFIBROSIS, POLYARTHRITIS, SPLENOMEGALY, BONE MARROW

42 yrs old male presented to our OPD with the complaints of pain and swelling affecting the both large and small Joints and Left upper quadrant abdominal fullness for the past 1yr. History of progressive weight loss for the past 6Months. History of generalized pruritus with reddish patches all over the upper trunk and flexor aspect of both Upper limbs were Present. Patient gave history of weight loss, about 25kg over the period of 6 months. History of Abdominal Fullness was present for the past 1 year, which was progressive in nature and associated with a dragging sensation, discomfort & early satiety.

On examination patient was thin built & poorly nourished. He had anemia, sparse scalp hair and healed pigmented macules over the flexor aspects of the upper limbs and upper back. An enlarged spleen which measured 18 cms below the left costal margin was present. Other systems were normal. Patient had arthritis affecting the PIP, MCP, Shoulder, Knee and Ankle joints. Joint movements were restricted.

INVESTIGATIONS:

- Complete blood count: Total count – 11,200 cells/cu.mm , Differential Count : polymorphs -70% , Lymphocytes 20%, Atypical lymphocytes-10%, Hemoglobin – 5 g/dl, PCV – 16%, ESR – 132 mm/hr, Platelet count – 1.26 lakhs/cu.mm.

- Renal function test and Liver function tests – within normal limits.

- Prothrombin Time and Activated partial thromboplastin time - within normal limits.

- HIV 1 & 2 –Negative.

- USG ABDOMEN AND PELVIS: Enlarged spleen of size 19.3 cm.

- CT ABDOMEN-MASSIVE SPLEEN WITH INFARCT

- HRCT LUNG: Normal.

- Venous Doppler: Prominent portal vein.

- Rheumatoid factor- negative

- Anti citrulinated cyclic peptide-negative

- Anti nuclear antibody-negative

- Antistreptolysin O titre -negative.

- Anti- DS DNA negative

- Peripheral Smear study: Microcytic Hypochromic RBC, with moderate anisopoikilocytosis including tear drop cells, atypical cells - 10%, shift to left in granulocytic series, circulating normoblast seen
Reticulocyte Count – 1%, Corrected Retic Count – 0.33%, Reticulocyte Production Index (RPI) – 0.13%.

15. Serum LDH-202 u/l

PERIPHERAL SMEAR OF THE PATIENT

16. BONE MARROW STUDY

a. TREPINE BIOPSY- Thickened Bony trabeculae seen with inter trabecular spaces showing extensive fibrosis with cellular areas showing moderate megakaryocytosis and granulocytic hyperplasia. Intra sinusoidal hematopoiesis seen. Myeloid metaplasia marked reticulin fibrosis

b. BONE MARROW ASPIRATE- Scanty aspirate, reduced hemopoietic elements. Erythroid component decreased.

BONE MARROW BIOPSY SPECIMEN

ABL BCR translocation study- no evidence of ABL/BCR translocation present.

Serum uric acid -5.1mg/dl

X RAY BOTH KNEE JOINT – joint space enlargement, synovial thickening. (Patient was not willing for synovial biopsy or knee joint aspiration)

DISCUSSION:
Myelofibrosis is a chronic bone marrow disorder in which the marrow is replaced by fibrous tissue. {1} Chronic Primary Myelofibrosis abbreviated as PMF. {2} is a clonal disorder of a multipotent Hematopoietic progenitor cell of unknown etiology characterized by marrow fibrosis, extramedullary hematopoiesis, and splenomegaly. Other designations for PMF include idiopathic myelofibrosis, agnogenic myeloid metaplasia or myelofibrosis with myeloid metaplasia. It is the least common form of chronic myeloproliferative disorder. Establishing this diagnosis in the absence of a specific clonal marker is difficult because myelofibrosis and splenomegaly may also occur in patients with Polycythemia Vera and Chronic myeloid leukemia. There is matrix overgrowth or underresorption or both. Age > 65years, Presence of Constitutional symptoms. Hemoglobin of <10gms/dl and the presence of circulating blast cells of <1% are the poor prognostic factors. Reticuloendothelial system hyperplasia, bleeding manifestations, pallor are the usual presentation. Differential diagnosis include Acute Myeloid Leukemia, Gaucher’s disease, Histiocytosis, Histoplasmosis and Myelodysplasia.

DIAGNOSTIC APPROACH:

Steroids, Interferon alpha, hydroxy urea, thalidomide, vitamin D are the drugs used in treatment. {5} Bone marrow transplant is the only potential cure.

CONCLUSION:

Even though presentation in this case initially pointed towards the diagnosis of connective tissue disorder, with systemic manifestation like symmetrical arthitis, high ESR & Mantoux positivity of 24mm. The final outcome of Bone marrow biopsy revealed IDIOPATHIC MYELOFIBROSIS. Possible cause for myelofibrosis with polyarthritis can be due to {3} myeloid metaplasia in the synovium. This case is reported for a rare cause of massive splenomegaly with thalidomide in the form of regression of size of spleen improvement in anemia which was previously refractory, arthralgia nonresponding to conventional treatment showed remarkable improvement to thalidomide thus giving us the indirect evidence of polyarthritis due to myeloid metaplasia. {3}

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