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Case Report

Mast cell leukemia- A rare case reported

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ABSTRACT

Background: Mast cell leukemia is a rare and aggressive clonal disorder of mast cells & their precursor cells. Systemic mastocytosis clinical course ranging from cutaneous mastocytosis, indolent disease, mast leukaemia and multisystem involvement.
Case Presentation: 14 Y/M presented with skin coloured plaque present over whole body with itching. On histopathological evaluation of skin lesion cutaneous mastocytosis was diagnosed. Further evaluation for systemic involvement by bone marrow aspiration was carried out, which shows hypercellular marrow for age, erythropoiesis and megakaryopoiesis within normal limits. The differential count revealed 70% of all nucleated cells were mast cells, with many large aggregates of >15% cells and 20% atypical mast cells seen as suggestive of MAST CELL LEUKEMIA (Aleukemic leukemia). S.trptase level >200ng/ml.
Results: The presence of the major criteria-bone marrow at least 20% atypical immature mast cell with least ≥15 multifocal dense Mast cells in bone marrow or extracutaneous organ, in addition to at least one minor criterion: 1) presence of atypical morphology in more than 25% Bone marrow or extracellular mast cells 2) Serum tryptase>20ng/ml. 3) Bone marrow, blood or extracutaneous organs: (a) CD2 and/or CD25 positive (b) Detection of KIT mutation at codon 816.
Conclusion: Considering characteristic bone marrow feature, and with biochemical and radiological investigation mast cell leukemia was diagnosed which helping clinicians to plan further management.

1. Introduction

Systemic mastocytosis is an uncommon heterogeneous disorder with manifestation ranging from cutaneous mastocytosis to systematic mastocytosis and may have indolent to aggressive clinical behaviour. Amongst this mast cell leukemia is exceedingly rare.[1] Cytoplasm of the mast cell contains numerous large granules that store predominantly histamine, heparin, eicosanoids, cytokines, chondroitin sulfate and neutral proteases.[2] Diagnosis is based on the presence of ≥ 20% atypical mast cells in the marrow or ≥ 10% in the blood; however, an aleukemic variant is frequently encountered in which the number of circulating mast cells is < 10%. The common phenotypic features of pathologic mast cells encountered in most forms of mastocytosis are unreliable in mast cell leukemia.[3] Clinical course of systemic mastocytosis due to mediator release from mast cells or infiltration of mast cells into tissues. They include signs, skin lesions, flushing, syncope, diarrhoea, hypotension, headache, abdominal pain and musculoskeletal disease.[4,5] Hepatomegaly, portal hypertension, splenomegaly, and ascites occur frequently in patients with systemic mastocytosis.[4]

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2. Case Report

A 14-year male presented with skin-coloured plaques over whole body with itching. Associated with nausea, vomiting, diarrhoea, and breathing difficulty. On examination multiple skin coloured plaque were present over face, trunk, upper limb, lower limb along with palpable bilateral inguinal and right cervical lymph node. Complete blood count showed all series within normal limit which was confirmed on peripheral smear. Diffential count revealed Neutrophils-56%, Lymphocytes-40%, Eeosinophil-01%, Monocytes-03%, Basophil-00%. On histopathological evaluation of skin shows dense diffuse infiltrate consisting predominantly mast cells which are round and spindle shaped with abundant eosinophilic cytoplasm, distinct cytoplasmic boundaries, large pale nuclei and spares eosinophils and lymphocytes, consistent with CUTANEOUS MASTOCYTOSIS was diagnosed. Further evaluation for systemic involvement by bone marrow aspiration was carried out.

2.1. Bone marrow aspiration findings

On bone marrow aspiration smears showed hypercellular marrow with reduced fat spaces. Erythropoiesis, Leukopoiesis and Megakaryopoiesis were within normal limits. 70% of all nucleated cells were mast cells, with many large aggregates of (>15 cells) seen, suggestive of Mast Cell Leukemia (aleukemic leukemia).

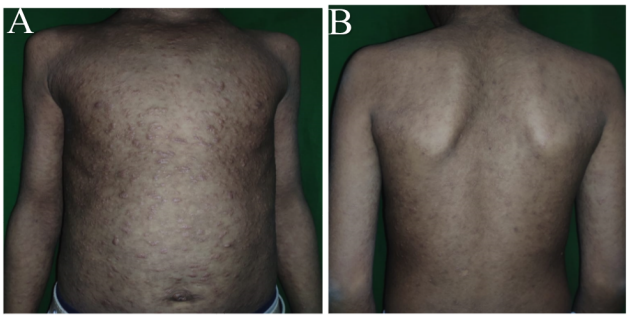

Figure 1: (A & B): Multiple skin-coloured plaques were present over trunk & back

2.2. Other investigation

Serum tryptase level >200ng/ml, ESR-18 mm/hr, ALT-17 IU/L, Serum Vitamin D- 15.5 ng/ml, Serum creatinine-0.6 mg/dl, Carpometacarpal bone X-ray findings showed Mildosteoporotic changes, USG Abdomen- Suggestive of minimal intraloop fluid.

3. Discussion

Mast cell leukemia is very rare form of systemic mastocytosis (<1%), which may appear de novo or

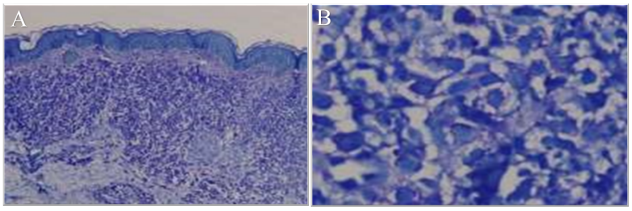

Figure 2: Photomicrograph (A & B): Low & High-power view of the biopsy shows mast cells which are round or spindle shaped with abundant eosinophilic cytoplasm, distinct cytoplasmic boundaries

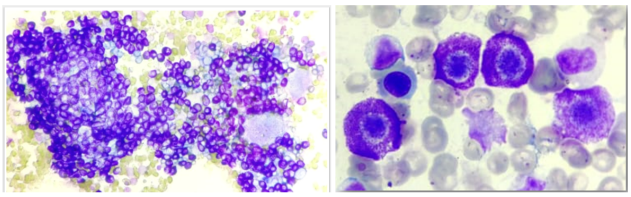

Figure 3: Photomicrograph: Low power view of bone marrow aspirate smears demonstrate dense infiltrates of mast cells. Large number of mast calls (>70%) have atypical morphology with many large aggregates of >15 cells seen

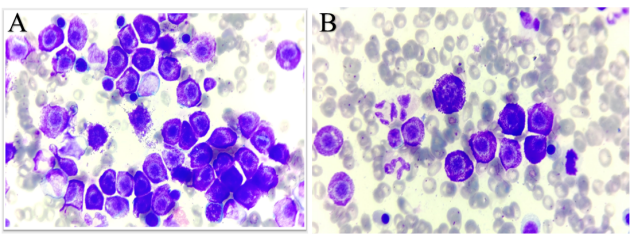

Figure 4: Photomicrograph (A & B): High power view of bone marrow aspirate smears demonstrate mast cells loaded with granules; A pinkish halo due to release of histamine is present around mast cells

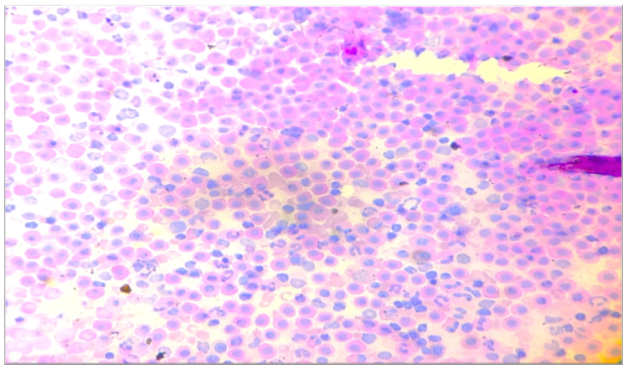

Figure 5: Photomicrograph (H): PAS stain the mast cells display magenta-coloured granules[6]

secondary to systemic mastocytosis.[7] Clinical presentations of mast cell leukemia are characterized by symptoms of mast cell activation, including fever, flushing, and tachycardia.[8] Organ involvement with signs of organ function impairment such as weight loss related to gastrointestinal involvement or progressive cytopenias related to bone marrow involvement are also frequently present. Mast cell leukemia is often "aleukemic", as in this case, where no mast cells found in the peripheral blood. The diagnosis of mast cell leukemia must meet the requirements Serum tryptase >20ng/ml, as in this case, Serum tryptase level >200ng/ml for systemic mastocytosis, demonstrate evidence organ function impairment, and have leukemic involvement of the bone marrow infiltrate is defined as ≥ 15 mast cells in aggregate, as in this case, 70% of all nucleated cells were mast cells, with many large aggregates of (>15 cells) seen, It can be promptly identified as is highly aggressive with median survival of < 6 months.

Table 1: The diagnostic criteria for mast cell leukemia: The presence of the major criteria & in addition to at least one minor criterion:[9]

Major criteria	**Minor criteria**
Multifocal dense mast cell infiltrates detected in sections of bone marrow or other extracutaneous organs; infiltrate is defined as ≥ 15 mast cells in aggregate.	1. Presence of atypical morphology in more than 25% of all mast cells infiltrates detected in sections of bone marrow or other extracutaneous organs. 2. Serum tryptase >20ng/ml. 3. Express 1 or more of CD2, CD30 or CD25. (Bone marrow, blood or extracutaneous organs). 4. Detection of KIT mutation at codon 816. (Bone marrow, blood or extracutaneous organs).

4. Conclusion

Considering characteristic bone marrow features and with supportive biochemical and radiological investigation mast cell leukemia was diagnosed, thus helping clinicians to plan further management.

5. Source of Funding

None.

6. Conflict of Interest

None.

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