

Systemic Mastocytosis- A Case Report

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Abstract

Mastocytosis is a rare heterogenous disease characterized by the presence of excessive number of mast cells in various organs mainly in skin and bone marrow. The incidence was 1 in 1,000 to 8,000 patients. It can follow a benign or indolent course, or it may be associated with life threatening symptoms. We report a rare case of 2 month old female child presented with hyper pigmented macular rash all over the body and few nodular lesions in popliteal and inguinal regions. The skin biopsy and bone marrow examination revealed numerous mast cells which stained metachromatically with giemsa and toluidine blue stain and showed positivity with cd 117. Based on these findings a diagnosis of systemic mastocytosis was made.

Keywords: mast cells, mastocytosis, metachromatic granules

1. Introduction

Mastocytosis is a rare heterogenous disease characterized by the presence of excessive number of mast cells in various organs mainly in skin and bone marrow. The incidence was 1 in 1,000 to 8,000 patients. It can follow a benign or indolent course, or it may be associated with life threatening symptoms.

2. Case Report

A 2 month old female child brought by mother with multiple hyper pigmented macules all over the body since 20 days of age. Also gave the history of swellings in the popliteal and inguinal regions on the left side. The family history and the personal history was nil remarkable. On examination child was alert and afebrile. Besides skin lesions and nodules rest of the physical examination was normal. Lab investigation revealed leucocytosis mainly due to rise in neutrophil count with normal platelet and eosinophil count. No organomegaly was noted. Serum tryptase levels were elevated upto 250microgm/L. Other investigations were within normal limits. Skin biopsy was done with the clinical diagnosis of urticariapigmentosa.



Figure 1: Clinical photograph of child showing hyper pigmented macules.

The Histopathological examination of skin biopsy of the lesion showed diffuse infiltration of the dermis with mast cells, therefore diagnosis of cutaneous mastocytosis was made. She was subsequently advised for BM examination to rule out its involvement. BM aspirate was normocellular with normal hematopoietic cells and prominent mast cells. Subsequent staining with Giemsa and Toluidine blue, stained mast cells metachromatically. Based on these findings a final diagnosis of systemic mastocytosis was made.

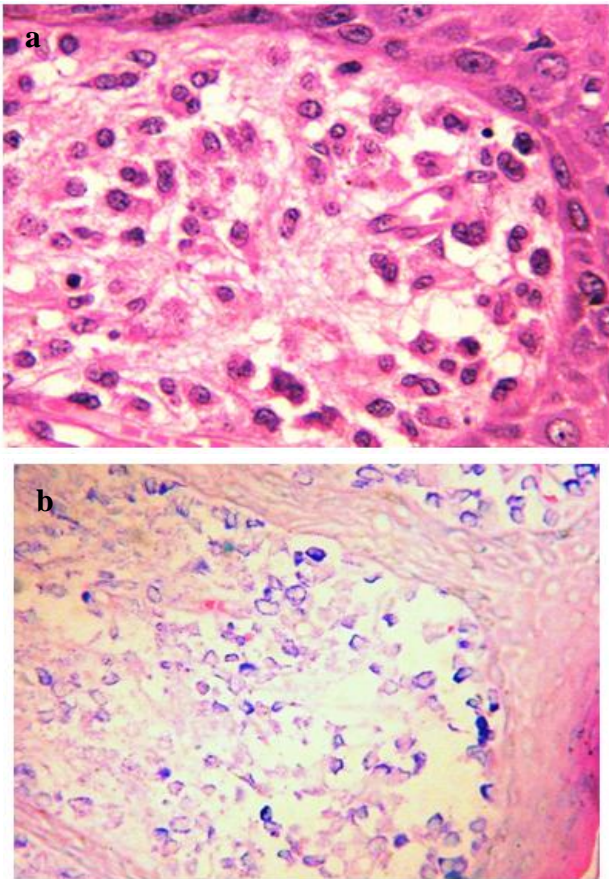


Figure 2a & b: Skin biopsy showing mast cells in H&E, GIEMSA

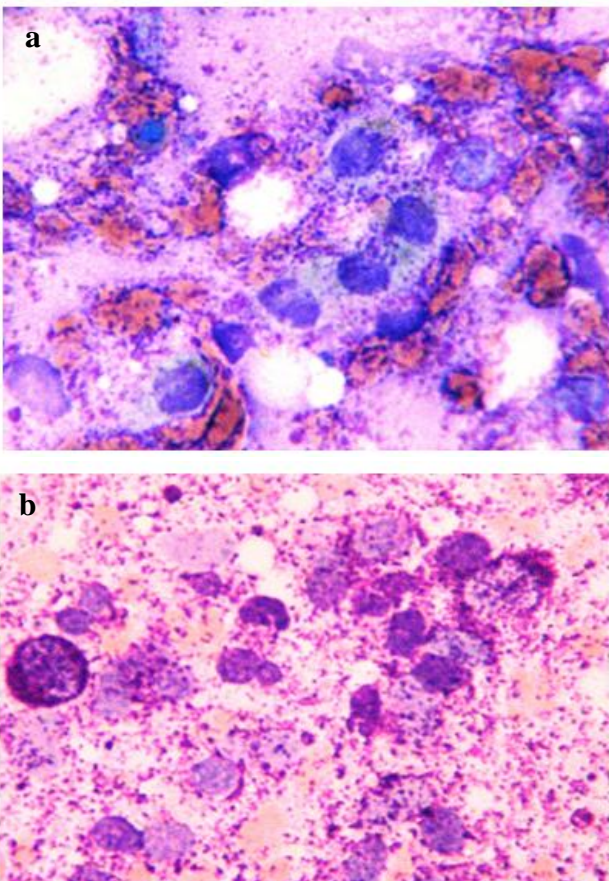


Figure 3a & b: Bone marrow aspiration with increased mast cells with Giemsa and toluidine blue stain.

3. Discussion

Human mast cells originate from pluripotential cells (CD34+) in the bone marrow and through the bloodstream migrate to a specific tissue where they undergo full maturation by acquiring a large amount of granules within the cell and assuming the definitive morphology. In tissues, mast cells differentiate into two subgroups: mucosal mast cells (MMC) and connective tissue mast cells (CTMC) based on their structures, biochemistry and functions. Mastocytosis is a rare disease characterised by abnormal growth and accumulation of mast cells in various organs [1].

This condition can be subdivided into: cutaneous (CM) and systemic mastocytosis (SM). CM is more frequent during childhood and it usually presents an early onset (up to the first year of age). The most frequently involved tissue is the skin, (cutaneous mastocytosis), bone marrow, gastro intestinal system, liver, spleen lymph node may be implicated to varying extent depending on the variant of disease (systemic mastocytosis) [2]. Systemic mastocytosis may follow a benign or indolent course or it may be associated with invalidating or life threatening symptoms. In indolent systemic mastocytosis patients present with maculopapular skin lesions and have a good prognosis [3].

The diagnosis of mastocytosis is established by demonstrating mast cell infiltration in an individual tissue, particularly the bone marrow and the measurement of serum tryptase is a good screening test since almost all patients with systemic mastocytosis have serum tryptase levels exceeding 20 microgm/ml [4].

Mastocytosis is an infrequent condition and although a number of classification schemes have been given for mast cell disease, the WHO classification of mast cell disease seems to be more practical and useful for characterizing the disorder [5]. Cutaneous mastocytosis has a relatively benign course and is seen more commonly in children. Our patient had maculopapular rash since 20 days. In indolent systemic mastocytosis the mast cells may form loose aggregates in bone marrow and become difficult to recognise without IHC [6]. They may be paratrabeular, perivascular or randomly distributed [7]. Our case showed scattered mast cells which are highlighted by special stains and final confirmed diagnosis of benign systemic mastocytosis was made.

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