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Case Report

NODULAR CUTANEOUS MASTOCYTOSIS: A CASE REPORT

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ABSTRACT
Mastocytosis is a rare condition caused by excessive amounts of mast cells gathering in the skin and in some instances in other organs. It is traditionally divided into cutaneous mastocytosis and systemic mastocytosis. We present here a case of a one-year-old girl who presented with maculo-papular, hyperpigmented and nodular pruritic lesions, in which a dermatopathologic diagnosis of nodular cutaneous mastocytosis was made.

Key Words: *Mastocytosis, Nodular Cutaneous*

INTRODUCTION
Mastocytosis is a rare and heterogeneous group of disorder of mast cell proliferation that occurs in both cutaneous and systemic forms (Correia *et al.*, 2010). These cells release large amounts of histamine and other chemicals into blood, causing symptoms such as a skin rash, itching and hot flushes (Andersen *et al.*, 2012).
Skin is the most frequent site of organ involvement (Correia *et al.*, 2010). We presents here a case of nodular cutaneous mastocytosis in a one-year-old child.

CASES
A one-year-old girl presented with maculo-papular, hyperpigmented and nodular pruritic rash since 4 months. Symptoms like abdominal pain, diarrhea, palpitations, syncope, bone pain, or weight loss were absent. There was no family history of any significant skin disorder. Physical examination revealed multiple oval to round hyperpigmented papules and nodules distributed all over the body (face, neck, chest and trunk), sparing her palms and soles. Darier's sign was positive. There was no pallor, icterus, edema, lymphadenopathy or organomegaly. Investigations were done, revealed normal complete blood count, liver function test and blood coagulation profile. Her Serum total Tryptase and Plasma Histamine levels were 23.5 ng/mL (reference range 1 to 10 ng/mL) and 1.424 pg/mL (reference range 0.3 to 1.0 ng/mL) respectively. Bone marrow examination was not done. Chest x-ray was normal. With a provisional diagnosis of utricaria pigmentosa, biopsy was taken from one of the nodular lesions over trunk. The specimen showed a single skin covered tissue with an ill–defined grayish white area (m) 0.5x0.4cm. Micro sections showed tissue lined by thin keratinized stratified squamous epithelium, with a dense confluent cellular infiltrate in the entire dermis and around appendiceal structures (Figure 1). The cuboidal mast cells showed moderate cytoplasm with distinct cell border, round to indented nuclei, low N: C ratio, there were no nucleoli or mitosis seen. The cells exhibited cytoplasmic granules that stained metachromatically with Giemsa (Figure 2). Immunohistochemistry revealed CD117 positivity. These findings along with serum Tryptase and CD117 positivity a diagnosis of cutaneous mastocytosis - nodular type was made.

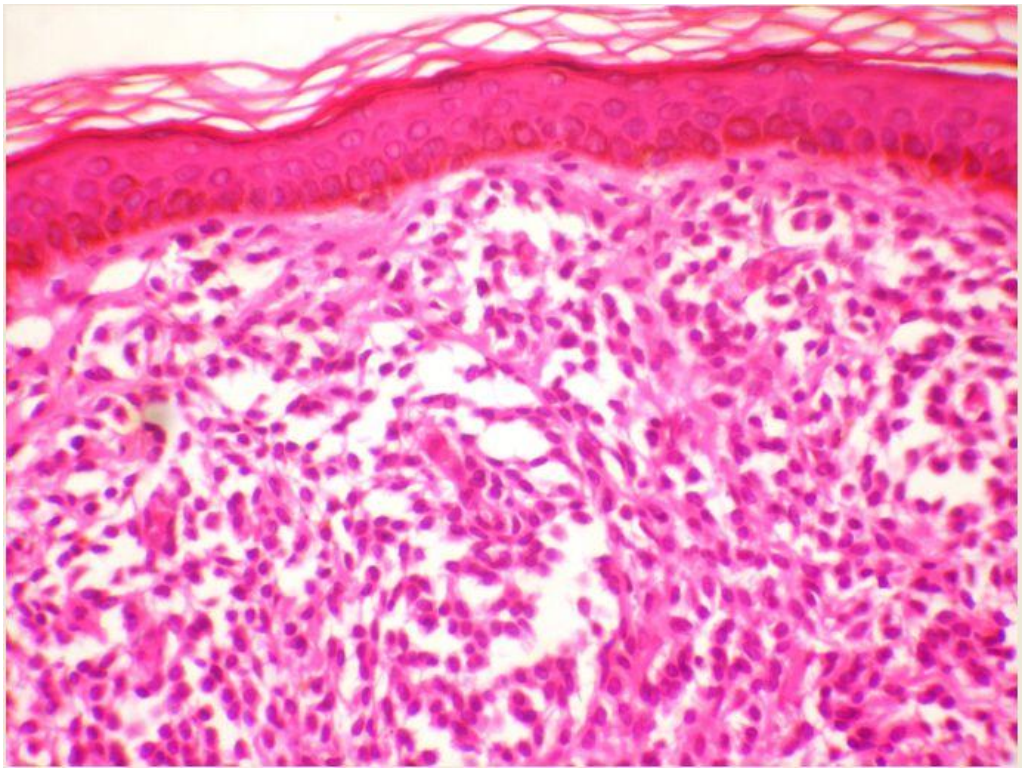

Figure 1: Diffuse infiltration of mast cells through the dermis (Hematoxylin & eosin, x400).

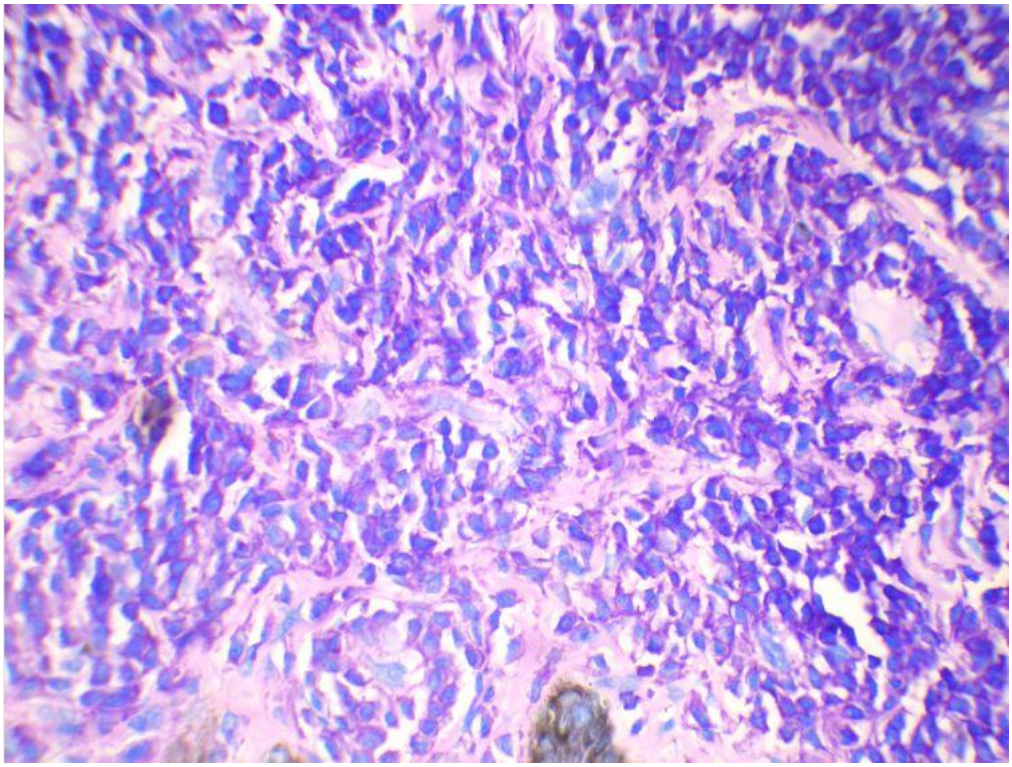

Figure 2: Giemsa staining reveals metachromatic granules within mast cells (Giemsa, x400).

DISCUSSION

Mastocytosis is a rare disorder characterized by abundance of mast cells in the tissues (Valent *et al.,* 2001). Its true incidence rates are unknown (Valent *et al.,* 2001). Skin is the most frequent site of organ involvement (Correia, 2010) . It is traditionally divided into cutaneous mastocytosis (CM) and systemic mastocytosis (SM). The cutaneous form include urticaria pigmentosa, solitary mastocytoma, diffuse cutaneous mastocytosis and telangiectasia macularis eruptive perstans. Systemic mastocytosis is further classified as: indolent systemic mastocytosis (ISM), SM with an associated clonal hematologic non-mast cell lineage disease (AHNMD), aggressive systemic mastocytosis (ASM), and mast cell leukemia (MCL) (Andersen *et al.,* 2012 and Valent *et al.,* 2001).

Cutaneous mastocytosis is a rare, mostly sporadic disorder (Asher *et al.,* 2012). The condition affects all races and does not exhibit any sex predilection or familial predisposition (Arun and Aparna, 2006). The commonest form of cutaneous mastocytosis is urticaria pigmentosa. Most often, these lesions begin during infancy and early childhood. The risk of developing mastocytosis also increases during middle age (Andersen *et al.,* 2012). Activating mutations of the c-Kit tyrosine kinase receptors have been implicated in the etiopathogenesis of mastocytosis (Andersen *et al.,* 2012). New tyrosine kinase inhibitors may be useful in targeting treatment for mastocytosis (Cancer, 2012). Skin is the most commonly affected site, but rarely bone marrow and gastrointestinal tract may be involved. The disease manifests as numerous reddish-brown, monomorphic maculopapules, nodules or plaques. Positive Darier's sign may be demonstrated in the lesions. Dermographism of normal skin is often present. Sometimes, it may present as blister in early infancy and childhood and heal without scarring (Asher *et al.,* 2012).

Mastocytosis can be easily diagnosed when it presents with the typical maculopapular rash and the characteristic Darier's sign, as was seen in our patient. In the absence of this presentation, the diagnosis can be missed. Usually a skin biopsy is necessary for confirmation of mastocytosis (Hogan and Elston (2012). Adult-onset mastocytosis, it has 8- fold increase incidence of systemic involvement. Approximately, 30% of patients with benign, systemic mastocytosis will eventually develop a malignant process. Patients with malignant mastocytosis significantly have lower erythrocyte sedimentation rate and platelet count while their leukocyte counts are significantly higher. These lesions are usually seen in an average age group of 66 years (Chow, 1999).

The diagnosis in this case was confirmed on skin biopsy, where it revealed abundance of mast cell infiltrate around the blood vessels, skin appendiceal structures and the entire dermis. The mast cells stained metachromatically with Giemsa and also showed positivity with CD117. The immunhistochemical staining with CD117 might be particularly helpful, especially in macular lesions when there is lower number of mast cells (Kiszewski, 2007).

Bone marrow examination is essential in the presence of other features, such as anaemia, unexplained eosinophilia, hepatosplenomegaly or lymphadenopathy. Serum tryptase of >20 ng/mL is considered to be a useful marker in adults, although its significance in young children is less certain (Asher *et al.,* 2012; and Arun *et al.,* 2006). Patients with mastocytosis have elevated plasma histamine levels but demonstrate a diurnal variation; hence plasma histamine levels alone are not useful to screen patients for mastocytosis (Friedman, 1989).

Cutaneous mastocytosis may mimic other dermatological conditions such as Juvenile xanthogranuloma, spitz nevus or Histiocytosis X, requiring a high degree of suspicion for clinical diagnosis followed by confirmation by histopathological examination. The condition is usually self-limiting in young children and of good prognosis. However because of extensive involvement, these patients should be followed closely. Our case is remarkable for its severe presentation in a child in whom a dermatopathological diagnosis of nodular cutaneous mastocytosis was made.

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