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Paediatrics

KEY WORDS: Mastocytosis, Langherhans cell histiocytosis, mast cells

CASE REPORT OF CUTANEOUS MASTOCYTOSIS

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ABSTRACT

Mastocytosis is a heterogeneous group of disorder characterized by abnormal growth and accumulation of mast cells in skin and other organs such as bones, gastrointestinal tract, liver, spleen and lymph node. A 3 months old male child, presented with multiple, discrete, reddish, papulo-nodular lesions all over body which became more prominent and itchy after a warm bath and crying. Darrier sign was positive and systemic examination was normal. SCORMA index was calculated. Skin biopsy was suggestive of Langherans cell histiocytosis. Immuno- histochemistry profile was positive for CD68, Vimentin, LCA and CD117. This condition needs high index of suspicion and should be differentiated from other causes of urticaria. It is a benign condition, but parental counselling and avoidance of trigger factors is essential. It is imperative to rule out malignancy and systemic complications in a case of cutaneous mastocytosis.

INTRODUCTION

Mastocytosis is a heterogeneous group of disorder characterized by abnormal growth and accumulation of mast cells in skin and other organs such as bones, gastrointestinal tract, liver, spleen and lymph node. The phenotypic expression of the disease is dependent on the pattern of localization of the mast cells to specific organs and the release of mast cell mediators. The skin is the most common organ involved in children and may be the only manifestation of the disease. Etiopathogenesis is multifactorial. Pediatric mastocytosis can be very easily be mistaken for a variety of common rashes that plague the pediatric population. Approximately 65% of individuals with mastocytosis present with disease in childhood; 55% of these patients have manifestations of disease by the age of 2 years

PATHOGENESIS

When mast cells degranulate, two of the main enzymes released are histamine and tryptase, which participate in the skin reaction that appears as a rash and is classified as mastocytosis.

Mast cells are most abundant in connective tissues, with a predilection for peripheral nerves, and blood and lymphatic vessels. It is at these sites, under the influence of cytokines (interleukins 3, 4, 5, 6, 9, 10, and 15) and the principal mast cell growth factor, stem cell factor (SCF), that mast cells differentiate from a CD34+ pluripotent hematopoietic stem cell

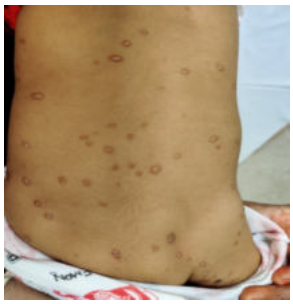
WHO Classification

1. Cutaneous Mastocytosis: Maculopapular type, Diffuse cutaneous Mastocytoma of skin
2. Systemic Mastocytosis: Indolent systemic mastocytosis, Smoldering systemic, Systemic mastocytosis with an associated haematological neoplasm mast cell leukemia
3. Mast cell sarcoma

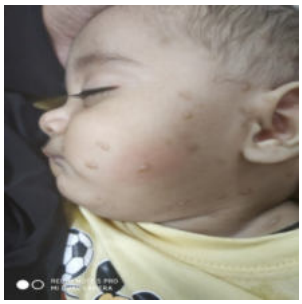
Children with cutaneous mastocytosis typically present with a spectrum of findings from solitary or multiple mastocytomas to urticaria pigmentosa (UP), or diffuse cutaneous mastocytosis (DCM). Pediatric mastocytosis is generally a benign disease that is transient in nature, as there is generally a spontaneous regression of the condition by puberty. SCORMA (SCORing MAstocytosis index) and serum triptase level should be evaluated in all paediatric patients with mastocytosis . The SCORMA index is a scoring system which is used in specifying the disease severity. The correlation between the SCORMA Index and serum tryptase levels underlines the benefit of the SCORMA Index as a clinical tool. Repeated SCORMA Index measurements can provide a rapid impression of changes in the clinical state of mastocytosis.The SCORMA Index consists of three parts and is based on the principles of the SCORAD (SCORing Atopic Dermatitis Index). In the SCORMA Index the extent of the skin abnormality is evaluated in the first part (A) The intensity of the disorder is dealt with in part (B) .Five subjective symptoms (triggering factors, flushing, diarrhea, itch and local bone pain) that may occur in mastocytosis are dealt with in part (C) .The formula: $A/5 + 5B + 2C/5$ is used to calculate the final SCORMA Index.The value of the SCORMA Index then lies between 5.2 and 100

CASE DETAILS:

A 3months old child, completely immunized, with no significant family history, developmentally appropriate for age came to tertiary care hospital with complain of multiple reddish raised lesion over body .No complain of diarrhoea, vomiting, Haemorrhage , symptoms suggestive of Malabsorption. O/E Patient had multiple discrete solid whitish yellow coloured papulo nodular lesions over trunk , upper limb , face, scalp, palms and multiple discrete brown coloured variously sized atrophied skin over trunk ,upper limb . Systemic examination was normal. his Hb- 7.6 mg/dl, Total WBC count-9200/cubic mm, Platelet count- 9.11 lakh, Darier's sign was positive. Skin biopsy was suggestive of Langherans cell Histiocytosis. There was infiltration of cells with vacuolated cytoplasm and oval to bean shaped nuclei and inconspicuous nuclei seen at dermo-epidermal junction and dermal pappilae. Scattered eosinophils were present. Immunohistochemistry profile was positive for CD 68, Vimentin , LCA , CD 117 markers suggestive of Cutaneous Mastocytosis.



A. DARIER'S SIGN POSITIVE



B. PAPULO-NODULAR LESION OVER FACE

DISCUSSION:

In our patient only cutaneous manifestations were present . There was no symptoms or signs of systemic involvement. This is case of utricaria pigmentosa which is most common type, seen in 70-90% of cases. SCORMA index was 38 suggestive of diffuse involvement.

Such lesions heal without scarring. It was confirmed by skin biopsy. Immunohistochemistry profile done to support the diagnosis. Symptomatic treatment was started . The patient was kept on regular follow up for monitoring of systemic manifestation and complication.

Education of parents about benign course spontaneous resolution before puberty is necessary.

Childhood mastocytosis is a rare genetic disorder. Cutaneous presentation has a good prognosis. 50–60% of children improve by adolescence and if it persists beyond , 10% of them go for systemic involvement with guarded prognosis. Cutaneous Mastocytosis should be followed once in 6 months to 1 year, and investigations must be repeated. Systemic Mastocytosis must be followed up to 5 years with regular monitoring. Counselling of parents and Information brochures regarding avoidance of trigger factors and life threatening complications should be given to prevent it. Clinical diagnosis is often delayed due to lack of awareness of disease and its consideration in differential diagnosis.

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