

**Case Report**

**PAEDIATRIC CUTANEOUS MASTOCYTOSIS : REPORT OF 3 CASES**

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**Abstract**

Mastocytosis occurs due to clonal, neoplastic proliferation of mast cells that accumulate in one or more organ systems. It is characterized by an abnormal mast cell infiltrates<sup>1</sup>. It can be divided into cutaneous and systemic Mastocytosis. Mast cell infiltrates remain confined to skin in cutaneous Mastocytosis. Urticaria pigmentosa / Maculopapular cutaneous Mastocytosis, Diffuse cutaneous Mastocytosis and mastocytoma of skin are the 3 variants of cutaneous Mastocytosis. Systemic mastocytosis is characterized by the involvement of atleast one extracutaneous organ with or without evidence of skin lesions. This paper describes 3 cases of paediatric cutaneous mastocytosis.

**Keywords:** Cutaneous mastocytosis, Mast cell, Pediatric

**INTRODUCTION**

Mastocytosis is characterized by an abnormal mast cell infiltrates. It can occur at any age. Severity of this disease ranges from fleetingly benign to aggressively malignant conditions. Cutaneous mastocytosis is most common in children. About 50% of affected children develop typical skin lesions before the age of 6 months. In cutaneous mastocytosis, mast cells infiltrates remain confined to skin, whereas systemic mastocytosis is characterized by involvement of at least one extra cutaneous organ with or without evidence of skin lesion.

**CASE REPORTS**

**CASE 1**

A 9 year old male child presented with generalized eruption of multiple hyperpigmented maculopapular lesions on face, trunk and extremities of 5 year duration. There was also history of generalized urticarial flushing. Growth and development status of the child is normal. On examination there were multiple sharply defined macules and papules over trunk, face and extremities. On stroking the individual lesions, there was formation of wheal and flare.

Darier's sign was positive, Systemic examinations were normal and routine blood examination, liver and renal function test were within normal limits. Peripheral blood smear was normal. No abnormality

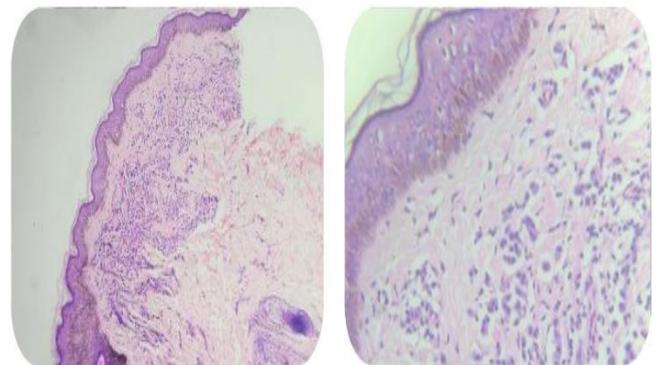
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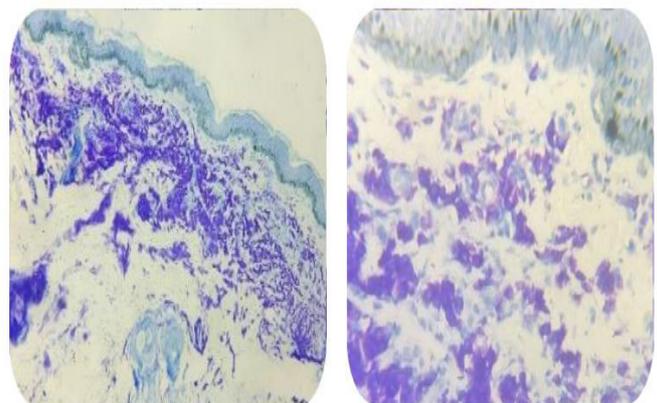
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**Figure 1 :** Hyperpigmented macules over chest and face



**Figure 2:** Upper dermis showing infiltration of mast cells. (H & E stain)



**Figure 3:** Mast cells in dermis showing metachromatic granules. (Toluidine blue stain.)

**Diagnosis :** Cutaneous Mastocytosis -Urticaria Pigmentosa

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detected in abdominal sonography. Histopathological examination showed skin with epidermis showing increased basal layer pigmentation, with mononuclear infiltrates in dermis. Toluidine blue stain showed multiple perivascular mast cells with metachromatic granules (purple colored granules ) in the dermis.

**Diagnosis :** *Cutaneous Mastocytosis -Urticaria Pigmentosa*

**CASE 2**

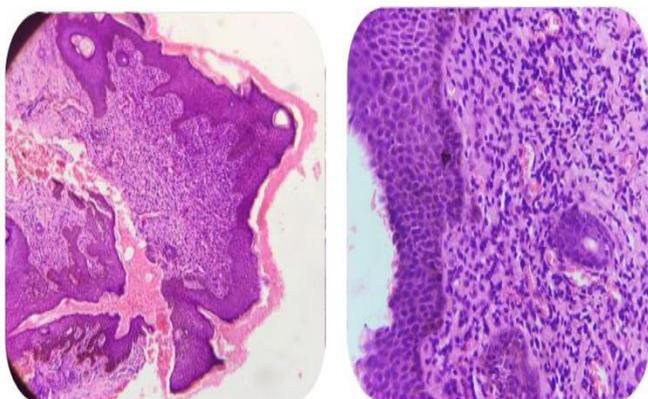
2 month old male infant presented with complaints of brownish to purplish macules and papules all over the body and excessive crying. Perinatal period was uneventful and exclusively breast fed with immunization up to date. No history of any drug intake. On examination



**Figure 4:** Diffuse hyper pigmented and hypopigmented macules over trunk

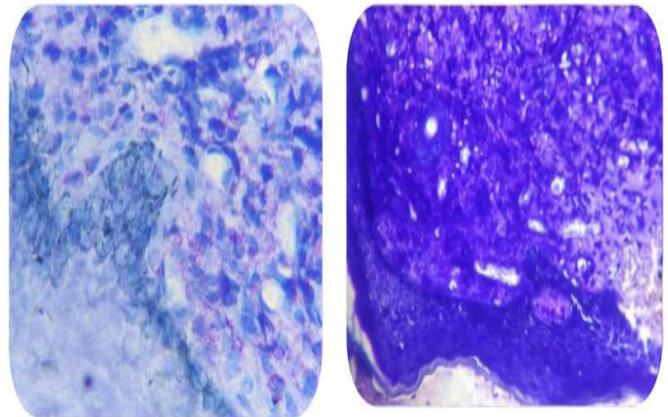
there is diffuse macules and papules all over the body, chiefly distributed over the trunk associated with hyper and hypopigmentation. No hepatosplenomegaly or lymphadenopathy.

Darier's sign was Positive, Systemic examination was normal. Routine blood examination and serum biochemistry are within normal limits. Peripheral smear showed mild thrombocytosis No abnormality detected in abdominal sonography. H&E –Diffuse infiltration of round to spindle shaped mononuclear cells in dermis. Toluidine blue and gi-

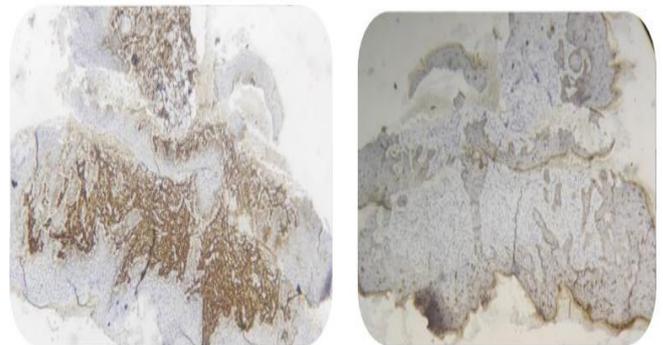


**Figure 5:** H & E section showing diffuse mast cell infiltrate in dermis

emsa stain showed metachromatic granules in mast cells. Immunohistochemistry – Mast cells showed strong positivity with CD117 . CD1a and CD 68 were negative.



**Figure 6** Toluidene blue and Giemsa stained sections showing mast cells with meta chromatic granules



CD117

CD1a

**Figure 7:** CD117 mast cells showing strong positivity, CD1a – Negative.

**Diagnosis :** *Diffuse Cutaneous mastocytosis*

**CASE3**

7 month old male baby presented with hyper pigmented raised lesion and a few discrete hyper pigmented and skin colored small papules on



**Figure 8:** Hyperpigmented raised crusted plaque with a few surrounding macules over ankle joint.

left ankle for 4 months. History of scratching and occasional oozing of pus from the lesion present . Child is developmentally normal. On examination, Single hyperkeratotic crusted plaque 2x1 cm over left ankle and a few small hyper pigmented and skin colored papules seen. No other similar lesions over the body.

Darier's sign – Positive. Systemic examination was normal. Routine blood investigations and serum biochemistry are within normal lim-

its. Peripheral blood smear within normal limits. Abdominal Sonography – No abnormality detected. Histopathology - Diffuse infiltration of round to oval shaped cells in superficial and deep dermis. Eosinophils seen interspersed between mononuclear cells. Metachromatic granules in Mast cells demonstrated with Toluidene blue stain Immunohistochemistry – CD117, CD1a and CD68 were negative

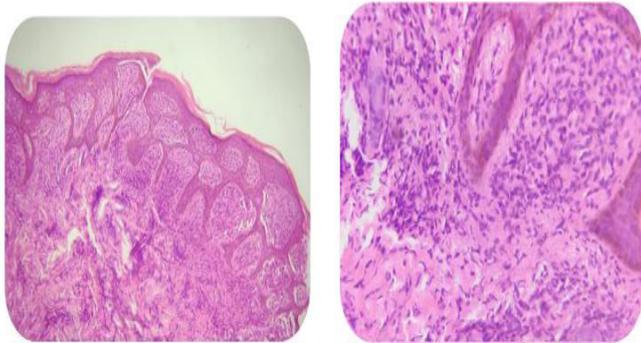


Figure 9: H&E stained sections showing infiltration of mast cells in dermis

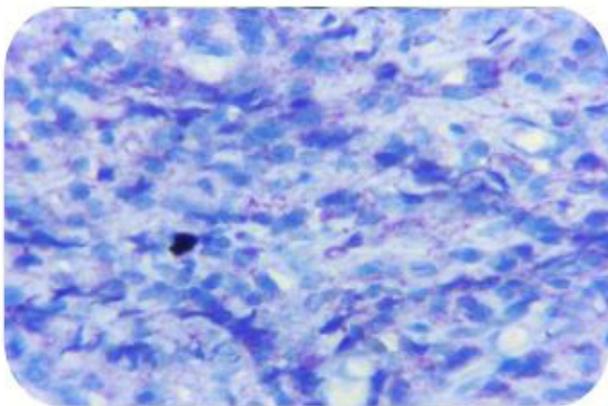


Figure 10: Toluidene Blue stain , demonstration of metachromatic granules.

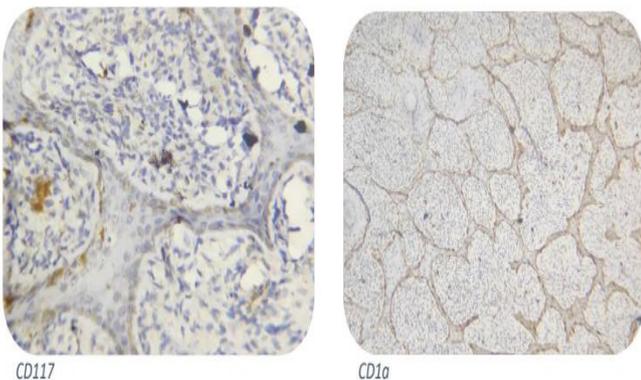


Figure 11 : CD117 and CD 1a Negative  
Diagnosis : Mastocytoma of skin

## DISCUSSION

Mastocytosis is first described by Nettleship and Tay in 1869 and later due to the appearance like urticarial, The term Urticaria pigmentosa was coined by Sangster in 1878. Classification of Mastocytosis variants<sup>2</sup>

(WHO classification of tumors of Hematopoietic and lymphoid tissue-2017)

### 1. Cutaneous Mastocytosis

- Urticaria Pigmentosa or Maculopapular cutaneous mastocytosis.

- Diffuse cutaneous mastocytosis
- Mastocytoma of skin

### 2. Systemic mastocytosis

- Indolent systemic mastocytosis
- Smouldering systemic mastocytosis
- Systemic mastocytosis with an associated hematological neoplasm.
- Aggressive systemic mastocytosis
- Mast cell leukemia

### 3. Mast cell sarcoma

#### Mastocytosis

Mastocytosis can occur at any age. There is no gender difference. Cutaneous mastocytosis is most common in children and can be present at birth. About 50% of affected children develop typical skin lesions before the age of 6 months. There is a tendency of regression with age. In cutaneous mastocytosis, mast cells infiltrates remain confined to skin, whereas systemic mastocytosis is characterized by involvement of at least one extra cutaneous organ with or without evidence of skin lesion. About 10% of patients with proven systemic mastocytosis do not have skin lesions<sup>3</sup>. The lesion of all forms may urticate when stroked (Darriers sign) and most show intra epidermal accumulation of melanin pigment. Despite variety of clinical presentation, The common picture is diffuse infiltration of mast cells in the dermis. Urticaria pigmentosa is the most common form of cutaneous mastocytosis, Lesions are larger, fewer and more papular. Diffuse cutaneous mastocytosis is less common than urticarial pigmentosa and present almost exclusively in childhood. Mastocytoma of skin, typically occurs as a single lesion, almost exclusively in children. In adult patients there is a genetic inactivation mutation of growth factor receptor C-kit and this mutation may be absent in pediatric cases and which is not associated with prognosis<sup>4</sup>. The diagnosis is mainly clinical. Serum tryptase levels correlates with severity. Histopathological examination is essential for a definite diagnosis, The mast cells are usually oval or spindle shaped with granules that stain metachromatically with toluidine blue. They are also well demonstrated by Giemsa, tryptase or chloroacetate esterase in formalin fixed biopsies. Distinguishing between cutaneous and systemic mastocytosis in patients presenting with skin lesions depends on further investigation, including bone marrow biopsy and blood tryptase measurements<sup>3</sup>. Prognosis depends on age, severity and clinical subtypes<sup>5</sup>. Treatment is symptomatic with antihistamines and avoidance of drugs which may stimulate the mast cell to degranulate<sup>6</sup>. Topical steroids also useful.

#### FOLLOW UP

All the 3 children showed good response to antihistamines and topical steroids, and currently doing well

#### CONCLUSION

Mastocytosis is characterized by an abnormal mast cell infiltrates. The diagnosis of cutaneous mastocytosis requires the demonstration of typical clinical findings and Histological proof of abnormal mast cell infiltration of dermis. In cutaneous mastocytosis. There is no evidence of systemic involvement in the bone marrow or any other organ.

Patients with cutaneous mastocytosis without systemic involvement have a good prognosis. Careful long term follow up is essential for the early detection of systemic involvement.

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