

CASE OF THE MONTH

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MASTOCYTOMA IN A 3-MONTH-OLD INFANT

Mastocytosis is a heterogeneous group of diseases characterised by the abnormal infiltration of mast cells (MC) in the skin and, sometimes, other organs. Symptoms in mastocytosis are caused by biological mediators released from MC and/or the infiltration of neoplastic MC in various organs, the skin and the bone marrow being predominantly involved. A WHO consensus classification for mastocytosis¹ exists, which is widely accepted and includes three major categories:

1. Cutaneous mastocytosis (CM), a benign disease in which MC infiltration is confined to the skin, is preferentially seen in young children and exhibits a marked tendency to regress spontaneously.
2. Systemic mastocytosis (SM), a rather rare disorder, it is commonly diagnosed in adults and includes four major subtypes:
 - (i) indolent SM (ISM, the most common form involving mainly skin and bone marrow);
 - (ii) a unique subcategory termed SM with an associated non-mast-cell clonal haematological disease (SM-AHNMD);
 - (iii) aggressive SM, usually presenting without skin lesions, and
 - (iv) MC leukaemia (MCL), probably representing the rarest variant of human leukaemias.

The diagnosis of SM, SM-AHNMD, and MCL might be confused with a variety of endocrinological, vascular, or immunological disorders. It is therefore of particular importance to be aware of the possibility of an underlying (malignant) MC disease in patients with unexplained vascular instability, unexplained (anaphylactoid) shock, idiopathic flushing, diarrhoea, headache, and other symptoms that might be mediator related. An important diagnostic clue in such cases is an increased serum tryptase level.

Systemic manifestations dependent on whether there is multiorgan/specific organ involvement, e.g. recurrent flushing with multiorgan involvement; gastrointestinal tract involvement: increase in gastric and colonic

mucosal mast cells, gastroduodenitis, bonemarrow: anaemia, thrombocytopenia, hepatosplenomegaly.²

3. The extremely rare localised extracutaneous MC neoplasms, either presenting as malignancy (MC sarcoma) or as benign tumour termed extracutaneous mastocytoma.

Diagnostic criteria for mastocytosis¹ are available and are widely accepted. SM criteria include one major criterion (multifocal compact tissue infiltration by MC) and four minor criteria:

- (i) prominent spindling of MC;
- (ii) atypical immunophenotype of MC with co-expression of CD2 and/or CD25 (antigens which have not been found to be expressed on normal/reactive MC);
- (iii) activating (somatic) point mutations of the c-kit proto-oncogene usually involving exon 17, with the imatinib-resistant type D816V being most frequent, and
- (iv) persistently elevated serum tryptase level (>20 ng/ml).

To establish the diagnosis of SM, at least one major and one minor criterion, or at least three minor criteria, have to be fulfilled.

Case report

A 3-month-old child presented with a solitary, progressively enlarging lesion on the abdomen. There were no associated systemic manifestations. There was no family history of mastocytosis. On examination the child was well-looking and had a brownish-red lesion, 5 cm in diameter on the abdominal wall. It was firm, shiny, nodular, non-tender with a typical *peau d'orange* appearance. (Fig 1). Stroking of the lesion with a tongue blade elicited a typical urticarial lesion (Darier's sign). Abdominal examination was otherwise normal. There were no other cutaneous lesions. A punch biopsy was performed after application of an emla patch. Histology revealed dense infiltration of mast cells, typical features of a mastocytoma (Figs 2-4). The child was not given any treatment. The follow-up visit a month later



Fig. 1. Mastocytoma on abdomen

This is a new feature aimed at highlighting instructive case studies that clinicians encounter. If you have an interesting case that you would like to share with our readers, please submit your case report to the Section Editor, Prof Cas Motala, e-mail cassim.motala@uct.ac.za

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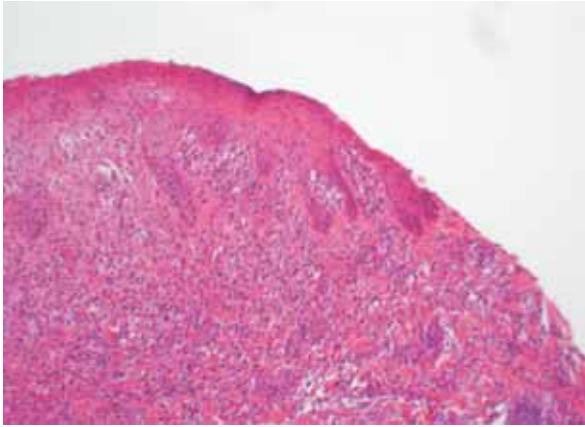


Fig. 2. Surface epidermis with underlying infiltrate of small mononuclear cells

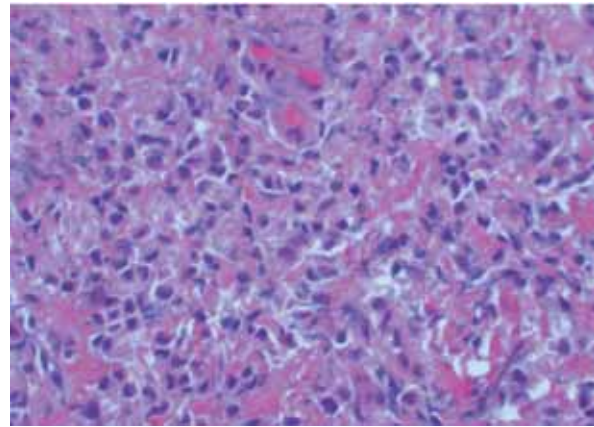


Fig. 3. High power view of infiltrate in dermis showing eosinophilic granules in the cytoplasm of the cells as well as interspersed eosinophils.

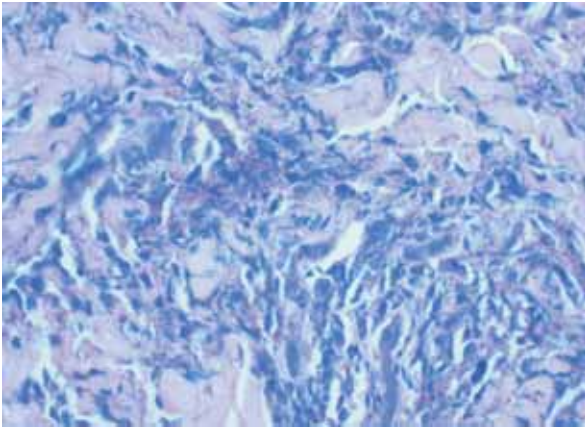


Fig. 4. Giemsa stain confirming granules in the cytoplasm typical of mast cells.

revealed that the lesion had almost completely resolved. The patient has remained well and asymptomatic since.

Discussion

Several recent studies³⁻⁷ have reported distinct epidemiological, clinical and prognostic differences in paediatric mastocytosis as compared with the adult form. The most common cutaneous lesions in children include urticaria pigmentosa, mastocytoma (one-third of cases) and diffuse cutaneous mastocytosis.^{3,4} Mastocytomas are present at birth in over 40% of cases. The disease occurs before the age of 2 years in 50% of children, with a peak incidence of 60% in the first year of life.⁵⁻⁷ Most cases of paediatric mastocytosis are sporadic and the majority of lesions are distrib-

uted over the trunk and limbs. Treatment of mastocytosis is usually symptomatic (H1 and H2 antihistamines to control itching). A particularly important aspect of management is the avoidance of triggering factors such as hot weather/bath. In contrast to adults, mastocytosis in children usually has a benign course. Systemic, mast cell disease in adults is a long-lasting disorder with recurrent and varying symptoms with the potential for malignant transformation, albeit rare.

Acknowledgement

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Declaration of conflict of interest

The author declares no conflict of interest.

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