CASE REPORT

Pseudoxanthomatous diffuse cutaneous mastocytosis in a child

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Abstract

Abnormal accumulation of mast cells in the skin is called cutaneous mastocytosis. Diffuse cutaneous mastocytosishas two clinical variants, pseudoxanthomatous/xanthelasmoid and bullous type. Childhood disease is mostly self limiting. This may sometimes be associated with other organ involvement. We present a case of pseudoxanthomatous cutaneous mastocytosis in a child without systemic involvement.

Keywords: Cutaneous Mastocytosis, Skin, Pseudoxanthomatous

Introduction

Mast cells are naturally distributed in the dermis, respiratory, gastrointestinal, and genitourinary mucosa, adjacent to the blood vessels, lymphatics, and peripheral nerves. However, diffuse infiltration of one or more organs with mast cells leads to the clinical condition called mastocytosis. The most common site for an abnormal accumulation of mast cells is the skin, known as cutaneous mastocytosis [1]. Cutaneous mastocytosis is the most common form of mastocytosis in children. The 2016 revision of the World Health Organization classification of mastocytosis classifies it into urticaria pigmentosa, Diffuse Cutaneous Mastocytosis (DCM), masto-cytoma, and telangiectasia macularis eruptiva perstans [2-3].

DCM has two clinical variants, pseudoxantho-matous or xanthelasmoid and bullous type [1]. Patients with this type of mastocytosis are at risk of systemic disease and severe complications but it often resolves spontaneously as in other types of childhood disease [4]. We present a rare case of DCM with xanthomatous appearance.

Case Report

A 2-year-old male child presented to the outpatient department with severely itchy raised lesions and raw areas on the extremities for one year. There was history of recurrent episodes during upper respiratory tract infections in winters. The child was atopic and born out of non-consanguineous marriage. There were no similar complaints in the family. On cutaneous examination, the skin was dry, lusterless, rugose, yellowish, leathery with diffuse infiltration all over the body predominantly in the flexures (axilla, groin, neck) (Figures 1 and 2). Few nodules were present over them. There were a few tense blisters and erosions on the forearms, hands, feet and buttocks. Systemic examination was normal. Differential diagnoses included papular urticaria, bullous impetigo, chronic bullous disease of childhood, diffuse mastocytosis and lepromatous leprosy. There was no nerve thickening but Nikolsky's sign and bulla-spread sign were negative while Darier's sign was positive.

Investigations like complete blood count, peripheral smear for abnormal cells, renal and liver function tests, erythrocyte sedimentation rate and ultrasound scanning of abdomen were normal. Slit skin smear for acid fast bacilli was negative. Biopsy on Haematoxylin and Eosin (H&E) stain revealed structure of skin with epidermis showing parakeratosis, mild thinning of prickle cell layer with shortening of rete pegs. Papillary dermis showed diffuse infiltration by round to oval cells having granular cytoplasm and spindle cells of fibroblast type. Lower dermis was unremarkable (Figure 3). The cytoplasmic granules were positive with toluidine blue stain representing mast cells (Figure 4). The case was diagnosed as DCM, xanthomatous variant. Serum tryptase levels and immunohistochemistry were not done due to lack of facility.

Child was given first generation antihistaminics for four weeks along with emollients and topical steroid. Parents were counselled to avoid triggers like extreme heat, cold, cough syrups and insect bites by covering the body with clothing. There was good improvement in four weeks. However, the parents were explained that the xanthomatous appearance will take time to resolve. The child was kept under observation to watch for adverse anaphylactic reactions or systemic involvement.

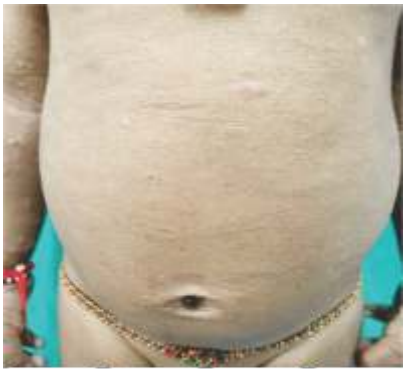

Figure 1: Pruritic papules, nodules, bullae, diffuse infiltration on the upper limbs and trunk

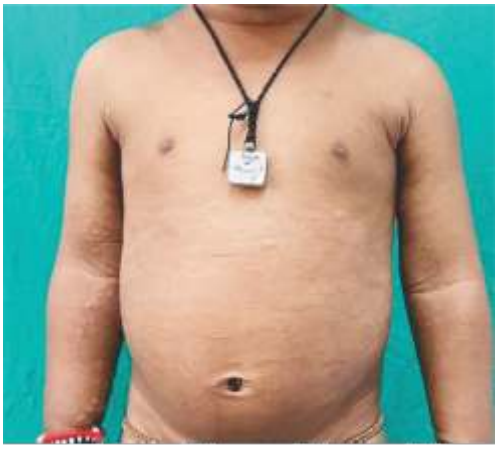

Figure 2: Yellowish, thick leathery skin and a few weals on the trunk

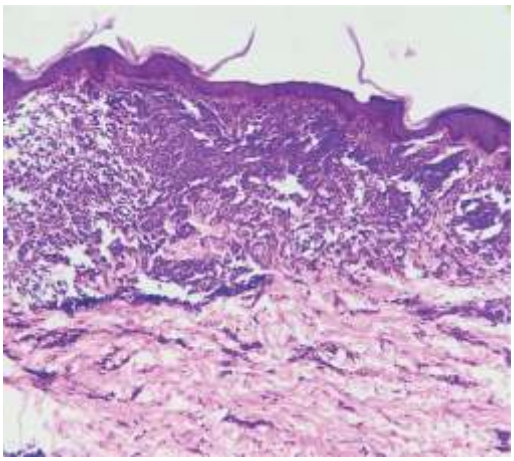

Figure 3: H and E, ×100, epidermis shows mild thinning of prickle cell layer. Papillary dermis shows diffuse infiltration by round to oval cells having granular cytoplasm and spindle shaped fibroblasts

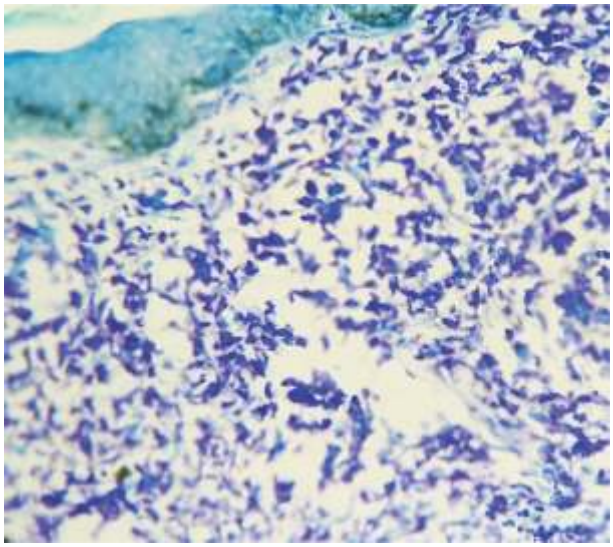

Figure 4: Toluidine blue, ×400, the cytoplasmic granules are positive with toluidine blue stain representing mast cells

Discussion

DCM is a rare form of pediatric mastocytosis, and can manifest as an eruption of papules, erythematous plaques, bullae, and erythroderma, skin thickening and pigmentation changes [2]. Approximately 65% of individuals with mastocytosis present with disease in childhood; nearly 55% of these patients have manifestations of disease by the age of two years. The remaining 35% of those that develop their disease after puberty are classified as adult onset [5]. Nabavi *et al.*, had reported cases of adult onset of xanthelasmoid mastocytosis [6]. Childhood-onset Mastocytosis (COM) is usually cutaneous and regresses spontaneously, while Adult-onset Mastocytosis (AOM) is often persistent with systemic involvement [7]. The symptoms of systemic disease vary from patient to patient and include pruritus, dyspnea, flushing, blistering, bone pain, and features of gastrointestinal involvement in the form of vomiting, diarrhea and epigastric pain [2]. The prognosis of childhood disease is good, but there is a risk of sudden mast cell degranulation due to any trigger and subsequent collapse [4].

Triggers for the release of mast cell mediators in mastocytosis are

1. Physical agents like temperature changes, rubbing, endoscopy, surgery
2. Hymenoptera stings
3. Emotional factors like stress, anxiety
4. Drugs like NSAIDS, opioids, anesthetics, intravenous iodinated radiocontrast agents, b-lactam antibiotics, aminoglycosides, streptomycin and phenylephrine [8].

This child had recurrent episodes of itchy lesions in winters and on taking cough syrups. However, there were no episodes of severe reactions or anaphylaxis.

A study by Kettelhut *et al*., revealed that most cases of cutaneous mastocytosis in children do not involve internal organs which precludes the need for routine prognostic bone marrow biopsies [9]. This child had diffuse infiltration of mast cells into the skin but fortunately no systemic symptoms. His developments were normal.

The treatment of the condition includes avoidance of triggers. Medications like antihistamines, topical or systemic corticosteroids, mast cell stabilizer, PUVA therapy, and interferons can be given [5].

This child showed improvement in the symptoms and clearance of lesion within four weeks of first-generation antihistamines and topical steroids. However, educating the patients about the triggers and avoiding them, explaining the need to observe for any adverse reactions will help.

Limitation

It is a single case report and advanced investigations like serum tryptase levels, immunohistochemistry could not be done.

Conclusion

We report a rare case of pseudoxanthomatous DCM in a child that presented with recurrent itchy papules, vesicles and weals who was systemically normal,similar tomost of the cases with bullous presentation. Such cases could be missed for simple papular urticaria, insect bite reactions, urticaria or prurigo, if the underlying yellow diffuse infiltration is missed.

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How to cite this article:
Vishwanath RG, Vishwanath R, Vishwanath V. Pseudoxanthomatous diffuse cutaneous mastocytosis in a child. *J Krishna Inst Med Sci Univ* 2023;12(3):136-140

Submitted: *06-Mar-2023* ***Accepted:*** *06-May-2023* ***Published:*** *01-July-2023*