Case Report

Childhood Pseudoxanthomatous Mastocytosis; Report of Two Cases

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Abstract

Cutaneous mastocytosis (CM) is a mast cell proliferative disorder that commonly affects pediatric population. It has various clinical forms such as urticaria pigmentosa, solitary mastocytoma, telangiectasia macularis eruptiva perstans, diffuse CM, and erythrodermic mastocytosis. Pseudoxanthomatous mastocytosis is a type of diffuse CM, which is relatively rare. It can mimic xanthoma disseminatum, juvenile xanthogranuloma, and benign cephalic histiocytosis which are also seen commonly in children but have different prognostic behavior. Hence, high degree of clinical suspicion along with histopathology is required for the diagnosis. Herein, we report two cases of childhood pseudoxanthomatous mastocytosis.

Keywords: Mastocytosis, xanthelasmoid, xanthoma

Introduction

Mastocytosis refers to group of disorders characterized by accumulation of mast cells in various organs such as skin, liver, spleen, bone marrow, and lymph node. Skin is the most commonly affected organ in mastocytosis which can manifest in various forms such as urticaria pigmentosa, solitary mastocytoma, telangiectasia macularis eruptiva perstans, diffuse cutaneous mastocytosis (DCM), and erythrodermic mastocytosis. Pseudoxanthomatous or xanthelasmoid mastocytosis is a rare variant of DCM characterized by the presence of yellowish papules and nodules resembling xanthomatosis. On literature search, we were unable to find more than three cases of xanthelasmoid mastocytosis reported from India. Herein, we report two cases of pseudoxanthomatous mastocytosis.

Case Reports

Case 1
A 4-year-old male child presented to dermatology outpatient department with pruritic papules and nodules all over the body for 1 month. The lesions started initially as few itchy nodules over the back. Gradually, similar lesions appeared over rest of the body over a period of 1 month. He had no history of flushing, dizziness, palpitation, pain abdomen, diarrhea, fever, weight loss, or bone pain. Family and personal history were noncontributory. Some of lesions subsided partially with postinflammatory hyperpigmentation after taking antihistamines. On dermatological examination, there were multiple skin colored to yellowish papules and nodules over the face, trunk, and extremities. Subsiding lesions were present in the form of postinflammatory hyperpigmentation. Routine laboratory tests such as complete blood count, peripheral smear comment, liver function test and renal function tests, lipid profile were within normal limits. Chest X-ray and abdominal ultrasonography were normal. A differential diagnosis of xanthoma, xanthoma disseminatum, juvenile xanthogranuloma, mastocytosis, and benign cephalic histiocytosis was considered. Punch biopsy from a representative lesion was done, and histopathological examination revealed collection of mast cells in the form of uniform round cells with ample eosinophilic granular cytoplasm.

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cytoplasm filling the papillary dermis, which was consistent with mastocytosis [Figure 2a]. On immunohistochemistry, those cells showed membrane positivity for CD117 [Figure 2b]. On the basis of clinical picture and histopathological finding, a diagnosis of pseudoxanthomatous mastocytosis was made. He was prescribed with levocetirizine 2.5 mg daily orally along with proper counseling to the parents about avoidance of triggering factors. The lesions were well controlled with the treatment for around 3 months, and he is on regular follow-up.

**Case 2**

One-year-old male child presented with multiple itchy papules and plaques over the trunk and upper extremities. On examination, there were well-demarcated skin colored to yellowish plaques over the trunk and forearm [Figure 3]. On rubbing the lesion, there was no erythema or urtication (Darier’s sign negative). He had no other systemic symptoms. Family history was negative. General and systemic examination was within normal limits. Routine laboratory tests did not reveal any abnormality. Chest X-ray and abdominal ultrasonography were normal. Punch biopsy from the lesion was consistent with mastocytosis. The lesions improved and symptoms were controlled with daily 5 mg of hydroxyzine on regular follow-up for about 5 months till date.

**DISCUSSION**

Mastocytosis is a heterogeneous group of disorders with varied spectrum of manifestations ranging from the involvement of only skin to that of various internal organs. It can occur both in children and adults. In contrast to adult-onset mastocytosis, the disease in the pediatric population is usually benign and resolve spontaneously.[6] In most children, mastocytosis is usually confined to the skin, whereas in adults, it is invariably associated with systemic involvement. About two-thirds of all CMs occur in children.[1] Cutaneous manifestations, which are the most common presentation of mastocytosis, usually occur early in life. There is an increased number of mast cell both in lesional and nonlesional skin.[2] DCM is a rare form of CM accounts for 1%–3% of CM.[6] As compared to other forms of CM, DCM is more likely to be associated with more severe form of systemic symptoms such as hypotension, shock, and diarrhea due to higher concentration of mast cells although the cutaneous lesion may not be proportionate.[7] Fortunately, none of our patients had systemic manifestations. Darier’s sign was negative in both of our cases. Although Darier’s sign is highly specific of mastocytosis, it may not be positive in all cases, and positivity ranges from 88% to 92%.[8]

Xanthelasmoid mastocytosis was first described by Tilbury Fox. In 1975, Griffiths and Daneshbod described pseudoxanthomatous mastocytosis in the form of yellowish nodular lesion resembling xanthoma or pseudoxanthoma elasticum.[9] Some authors believe that the different nomenclatures such as nodular, xanthelasmoid, and pseudoxanthomatous mastocytosis can be used as synonyms as they represent similar clinical conditions with slight variations.[10]

We are reporting these cases because of their rarity of occurrence in India.[3-5] In both of our case, cutaneous lesions were resembling xanthoma, but histopathology reports were consistent with mastocytosis, so a diagnosis of pseudoxanthomatous mastocytosis was made. It is important to distinguish this entity from other closely mimicking

![Figure 1: Skin colored to yellowish papules and nodules over the trunk and extremities simulating xanthoma](image)

![Figure 2: (a) Mast cells as uniform round cells with ample eosinophilic granular cytoplasm filling the papillary dermis (H and E, ×40). (b) Immunohistochemistry showing cells with membrane positivity for CD117 (×10)](image)

![Figure 3: Well-demarcated yellowish plaque over the trunk](image)
dermatosis such as xanthoma disseminatum, juvenile xanthogranuloma, benign cephalic histiocytosis, and confirm the same by histopathological examination as the latter diseases have different prognostic values. As the majority of childhood CM remit spontaneously, it is important to counsel the parents regarding the benign and chronic course of the disease.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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