A Case of Pediatric Cutaneous Mastocytosis Successfully Treated with Imatinib Mesylate

Sir,
Cutaneous mastocytosis (CM) in children is a rare benign cutaneous proliferative disorder caused by mast cell proliferation in the dermis. It presents with highly itchy skin lesions on body which may show blistering at times. The condition is known to resolve slowly, but it may progress to systemic dissemination in some cases if not treated. Traditional treatments aim at only controlling itching and are partially effective. We report an uneventful therapy of diffuse CM with imatinib mesylate in a 14-month-old boy.

A 14-month-old boy was brought to our dermatology outpatient department with multiple, severely itchy, skin-colored–to-erythematous lesions all over his body [Figure 1]. The child was irritable and was unable to have a peaceful sleep due to severe itching. The patient was treated with multiple courses of oral antihistamines and steroids with partial improvement.

Cutaneous examination showed multiple skin-colored-to-yellowish xanthomatous papules and plaques over entire skin surface. After informed consent, a lesional skin biopsy was done. Histology showed diffuse dermal infiltrate of mast cells occupying the entire papillary and reticular dermis. On clinicopathological correlation, a diagnosis of CM was made and was confirmed by metachromatic special stain (Toluidine blue) showing purple staining granules [Figure 2a and b]. Systemic involvement was ruled out by clinical, laboratory, and radiological investigations. Final diagnosis of diffuse CM was made. Genetic analysis was done from skin biopsy for KIT mutation, which turned out to be negative.

Figure 1: Multiple skin-colored papulonodules over back with positive Darier’s sign

Figure 2: (a) Typical “fried egg” appearance of mast cells (H and E, ×10). (b) Toluidine blue stain showing pink cytoplasmic granules with degranulation (Toluidine blue under oil immersion)
The child was prescribed oral imatinib 100 mg once a day in consultation with a pediatric oncologist. The treatment was tolerated well. There was a significant reduction in itching and lesions started reducing within a month. Treatment was continued for 3 months with regular monitoring of blood counts, and liver and kidney function tests. At the end of 3 months, there was near complete clearance of lesions [Figure 3].

Imatinib is the only systemic mastocytosis treatment currently approved by the Food and Drug Administration (specific indication is treatment of adult patients with aggressive systemic mastocytosis without the KIT D816V mutation or with unknown KIT mutational status). However, there is paucity of literature documenting the therapeutic potential of oral imatinib in CM. Thorough search of literature showed only two reports of use of imatinib mesylate in CM without systemic involvement.[2,3] There is a single report supporting the use of oral imatinib in CM in a case of chronic myelomonocytic leukemia.[4] In our case, mutational analysis for KIT gene (CD117) did not reveal any aberrations (deletion and missense). Successful response to imatinib in our case could be attributed to hitherto undiscovered mutations in KIT gene. Imatinib may be useful in cases of systemic mastocytosis with cutaneous involvement, who do not show KIT mutation or have mutation located outside KIT activation loop. Imatinib as treatment for CM has not been tried in India till date. To conclude, we successfully treated disabling form of diffuse CM with a novel therapeutic approach without any major adverse effect.

**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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