

Mal De Meleda with Flexural Involvement

Abstract

Mal de Meleda is a rare variety of palmoplantar keratoderma with an autosomal recessive mode of inheritance and an estimated prevalence of 1 in 100,000 in general population. The disease is associated with consanguinity, starts in infancy, and characterized by progressive palmoplantar keratoderma spreading to dorsum of hands and feet. Involvement of extensors of knee and elbow were well described in literature. We report a rare case of Mal de Meleda with flexural involvement, which has not been reported in literature.

Keywords: Dermatoglyphics, Mal de Meleda, MDM, palmoplantar keratoderma, PPK

Introduction

Mal de Meleda (MDM, MIjet disease) is a very rare autosomal recessive form of hereditary palmoplantar keratoderma (PPK) with an estimated prevalence of 1 in 100,000 in general population.^[1] It is named after the Croatian island of Meleda (MIjet), where its prevalence is high due to inbreeding.^[2]

Case Report

A 28-year-old Hindu unmarried woman presented to us with thickening of palms and soles since infancy. The lesions started initially as small erythematous pinpoint papules with asymptomatic exfoliation on both palms when she was 3 months of age. Simultaneously, erythematous scaly plaques on both cubital fossae, and dorsal aspect of both hands and feet appeared. Gradually, the skin of palms and soles became thick, waxy, and yellow with loss of dermatoglyphics. There was also gradual tightening of skin over fingers and toes. On examination, along with the above findings, the additional findings were hyperhidrosis of palms and soles along with macerated interdigital spaces with erosions emanating a foul smell, fissures in palms and soles [Figures 1-3], conical tapering of the distal digits, minimal digital constriction in the little finger of the left hand, and mild flexion deformity of fingers [Figure 4]. Erythematous scaly plaques with sharp

margins were present over bilateral cubital fossa [Figure 5]. Oral mucosa and perioral skin were apparently normal. There was no history of ocular involvement, hearing impairment, dental abnormalities, or atopy. Hair and systemic examination did not reveal any abnormality. There was history of consanguinity, and the father and mother were first-degree relatives with no similar complaints. None of the siblings and relatives had similar complaints.

Histopathology of palmar skin showed compact hyperkeratosis, sparse superficial perivascular lymphocytic infiltrate with epidermal hyperplasia. The granular layer was thickened. The papillary dermis was thickened and showed thickened bundles of collagen in vertical arrays [Figure 6]. A diagnosis of MDM was made based on the above-mentioned history, clinical features, and histopathology findings. Genetic analysis for SLURP1 could not be done.

To decrease the foul odor, chance of infection and to improve the hygiene, she was advised to use condy's solution to cleanse her hands and feet, topical antibacterial and topical antifungal formulations for local application. She was advised oral vitamin A palmitate 25,000 IU twice daily. She sought advice regarding marriage and the possibility of the children developing the similar condition. She was counseled that she had an autosomal recessive condition, and there is a 25% chance of the child developing this during each pregnancy. She is under follow-up at present.

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Figure 1: Hyperkeratosis of both palms with sharp margin with maceration and erosion over interdigital spaces



Figure 2: Hyperkeratosis of both soles with maceration



Figure 3: Hyperkeratosis of both dorsae of feet

Discussion

Mal de Meleda PPK or keratosis palmoplantaris transgrediens of Siemens is an inherited autosomal recessive variety of hereditary PPK, which is associated with high degree of consanguinity and inbreeding. Usually, the disease starts at birth as palmoplantar erythema, which progresses to develop thick lesions during the first 3 to 4 years of life and manifests clinically as bilateral thick yellow to brown PPK with prominent erythematous border. The hyperkeratosis spreads to the dorsal aspects of hands and feet, like gloves and socks, which is referred to as transgrediens. Hyperkeratosis progresses without a tendency of spontaneous resolution. Keratoderma is most often complicated by hyperhidrosis, secondary bacterial and fungal infections, and is associated with foul smell. Oral manifestations in the form of perioral erythema, high-arched palate, angular cheilitis, and lip involvement have been reported.^[3-5] There may be nail involvement manifesting as Beau's lines, koilonychia, subungual hyperkeratosis, onycholysis, and onychogryphosis.^[3] Psoriasiform lesions occur occasionally over elbows and knees. The progressive



Figure 4: Hyperkeratosis extending to dorsum of hands and flexion deformity of fingers

form of disease presents with severe mutilation, constricting bands around digits, pseudoainhum, amputation of digits, and contractures.^[4] Moreover, MDM can lead to serious severe flexion contractures of hand leading to impairment of patient's daily activity.^[6] Very rare features include



Figure 5: Erythematous scaly plaques with well-defined margins over bilateral cubital fossa

syndactyly, lingua plicata, left-handedness, and hair on the palms and soles.^[7]

The absence of symptoms in biological parents is suggestive of either an autosomal recessive inheritance or a spontaneous mutation. The mutations in the gene encoding secreted Ly-6/uPAR-related protein 1 (SLURP-1) located on the chromosome 8q24.3 found to be the cause.^[3]

There are several inter-individual variations in the clinical diagnosis of MDM, which should be differentiated from other PPK syndromes like Greither's disease, Olmsted's syndrome, Vohwinkel's syndrome, Huriez syndrome, Papillon Lefevre syndrome, and so on.^[5]

MDM persists lifelong. Management mainly includes symptomatic treatment and genetic counseling. The patient is to be educated for selfcare like taking care of hyperkeratotic hands and feet, prevention of hyperhidrosis, and secondary bacterial and fungal infections. Treatment includes topical corticosteroids, emollients, keratolytics like topical urea, lactic acid, retinoid acid, and topical antimicrobials. Oral 13-cis retinoic acid,^[4] acitretin 20 mg/day has shown some improvement.^[8] The hyperkeratosis should be debrided surgically. Surgical treatment of hyperkeratosis can be an option with excision and subsequent placement of a full-thickness skin graft.^[9]

This case is reported for its rarity, and uniqueness in conformity. Although this disease has been reported in several countries or regions around the world, few such cases have been reported in India. To our knowledge, no case of MDM with flexural involvement has been reported till now. Therefore, in this case report, we represent a patient of MDM with flexural involvement from eastern India to strengthen and extend the understanding of MDM.

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

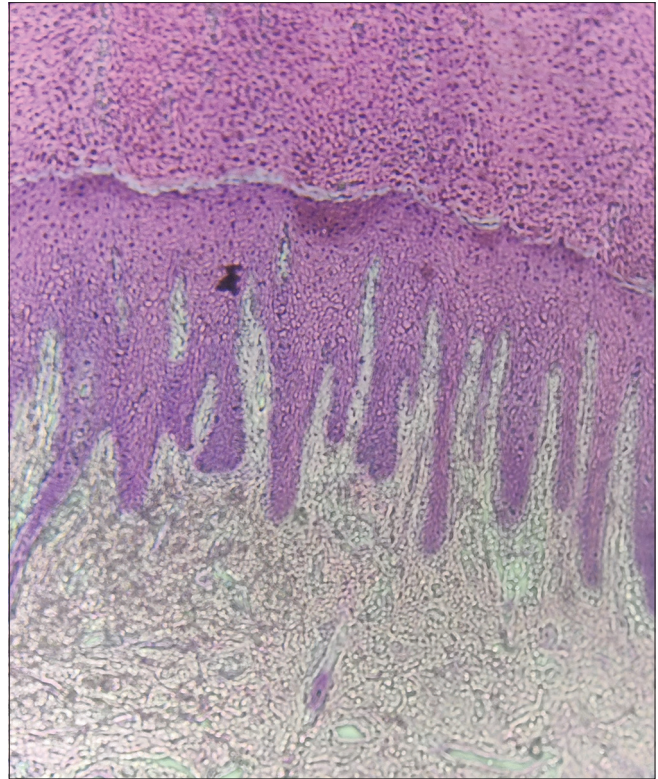


Figure 6: Hyperkeratosis, sparse superficial perivascular lymphocytic infiltrate with epidermal hyperplasia, thick granular layer, and the papillary dermis thickened with bundles of collagen in vertical arrays (hematoxylin and eosin, 10×)

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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