CASE REPORT

Mal de Meleda in an 18-year-old male from a non-consanguineous family: a case report

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Abstract

Mal de Meleda is a rare autosomal recessive palmoplantar keratoderma caused by biallelic mutations in the SLURP1 (Secreted Ly-6/Urokinase-type Plasminogen Activator Receptor Related) gene, which plays a critical role in keratinocyte differentiation and skin barrier integrity. The estimated prevalence is approximately 1 in 100,000, making it an exceedingly rare genodermatosis. The disease is typically observed in consanguineous families, and the clinical onset occurs shortly after birth, following a *transgrediens* pattern – extending from the palmoplantar surfaces to the dorsal aspects of the hands and feet – along with *progrediens* progression, with symptoms worsening over time. In addition to causing significant cutaneous disfigurement, the condition can severely impact hand and foot function, thereby compromising the patient's quality of life.

The authors present an atypical case of Mal de Meleda in an 18-year-old male patient from a non-consanguineous family, presenting with sharply demarcated hyperkeratotic plaques extending from the palms and soles to the dorsal hands and feet. This case highlights the clinical variability of Mal de Meleda and stresses the importance of considering this diagnosis even in the absence of a consanguineous background.

Keywords: Mal de Meleda; palmoplantar keratoderma; transgrediens; consanguinity; autosomal recessive

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Introduction

Mal de Meleda (MDM), also known as keratoderma palmoplantaris transgrediens, is a rare autosomal recessive palmoplantar keratoderma first identified on the Croatian island of Meleda (Mljet).[1] It is caused by biallelic mutations in the SLURP1 (Secreted Ly 6/Urokinase type Plasminogen Activator Receptor Related) gene, typically observed in consanguineous families.[2] Clinically, MDM presents as a progressive, sharply demarcated, waxy, ivory yellow hyperkeratosis extending from the palms and soles to the dorsal surfaces of the hands and feet. Lesions often arise in early childhood and evolve with associated features such as hyperhidrosis, fissuring, and malodor, leading to significant discomfort, social stigma, and restriction of daily activities.[3] We report a rare case of MDM presenting in an 18-year-old male patient from a non consanguineous family.

Case Report

An 18-year-old male presented to the dermatology outpatient department with thickening and peeling of the skin of the bilateral palms and soles (Figure 1A-1B), associated with mild pruritus and moderate pain. The patient stated that lesions appeared 2 years earlier and progressed slowly, extending from the palmar and plantar surfaces to the dorsal aspect of the hands and feet, with a sharp demarcation. The thickened, hyperkeratotic skin caused painful contractures and reduced range of movements. Cutaneous examination revealed diffuse, waxy palmoplantar hyperkeratosis, hyperpigmentation, and lichenification on the dorsal aspect of the hands and feet, and hyperkeratotic plaques on the proximal interphalangeal joints (Figure 1C). An ulcer was observed on the medial aspect of the right fourth toe, while the left fifth toe showed conical tapering (Figure 1D).

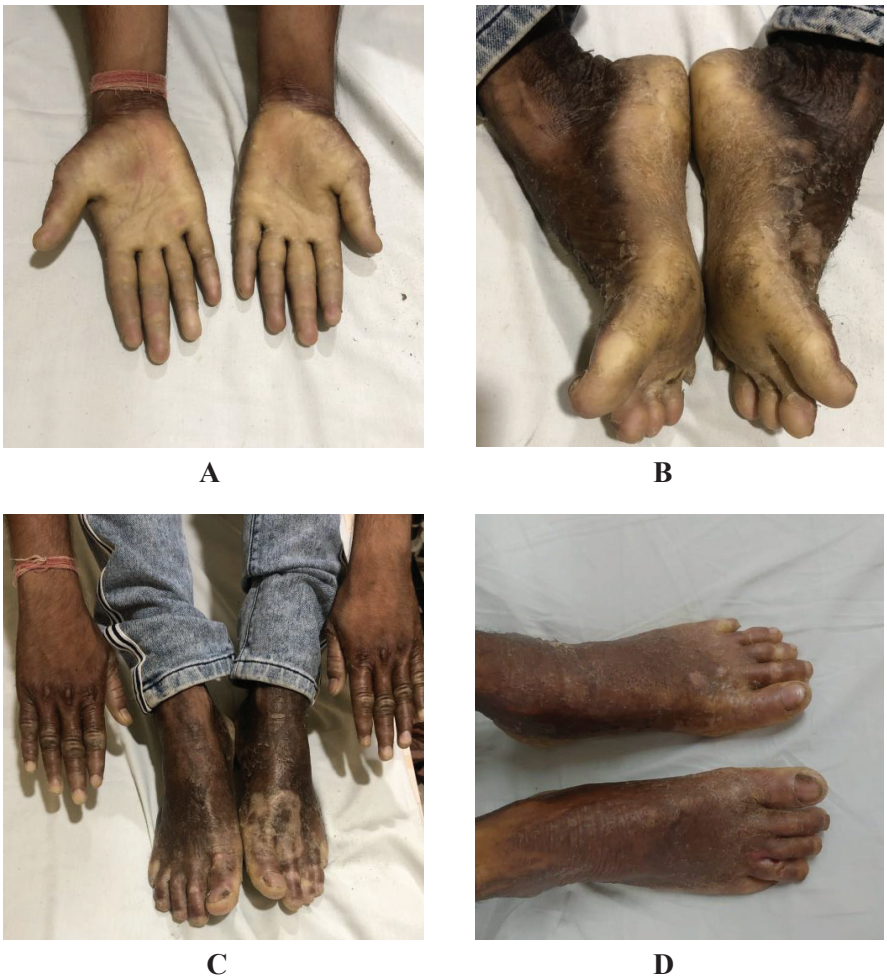

Figure 1. Clinical features of Mal de Meleda. (A) Diffuse waxy palmoplantar keratoderma on palms. (B) Diffuse, waxy hyperkeratosis extending to the dorsum of the feet. (C) Hyperkeratotic plaques on the proximal interphalangeal joints. (D) Conical tapering of the toe with ulcerations.

The patient also had hyperhidrosis, leading to fissuring and maceration of the interdigital spaces, with associated malodor. The nails were rough and exhibited onycholysis. No other systemic or mucosal abnormalities were noted, and there was no family history of a similar condition. Histopathology revealed hyperkeratosis, hypergranulosis, and acanthosis in the epidermis with perivascular lymphocytic infiltration in the dermis. For logistical reasons, genetic analysis for SLURP1 could not be done.

Based on clinical examination and disease progression, a diagnosis of Mal de Meleda was made. The patient was treated with oral acitretin (initial dose 20 mg/day) for six months, supplemented by topical antibacterial and antifungal treatments. The patient experienced significant clinical improvement within this period, with reduced hyperkeratosis, pain, and malodor, allowing a return to routine daily activities.

Discussion

Mal de Meleda is caused by mutations in the SLURP1 gene, which plays a crucial role in epidermal differentiation and keratinocyte adhesion. This rare disorder is characterized by sharply demarcated, transgrediens hyperkeratotic plaques (glove and stocking keratoderma), associated erythema, and scleroatrophy.[4] The clinical spectrum may also include hyperhidrosis, fissuring, lichenification, digital contractures (pseudoainhum), perioral erythema, high arched palate, and ocular complications.[5]

Although typically inherited in an autosomal recessive manner within consanguineous families, this case highlights that MDM can arise at a later age, in the absence of parental consanguinity, suggesting possible de novo or compound heterozygote mutations. Differential diagnoses for MDM include Greither disease, Olmsted syndrome, Vohwinkel syndrome, Huriez syndrome, Nagashima-type palmoplantar keratosis, Unna-Thost Syndrome, and Papillon Lefèvre syndrome, as mentioned in Table 1.[6,7]

Table 1. Clinical differentiation of Mal de Meleda from other hereditary palmoplantar keratodermas

Disorder	Inheritance	Cutaneous Features	Nail Features	Other Features
Mal de Meleda	Autosomal recessive *(SLURP1)*	Diffuse, sharply demarcated transgredient PPK extending to dorsal hands/feet; progressive; fissuring; hyperhidrosis; pseudoainhum tendency	Dystrophy and onycholysis may occur	Possible contractures, malodor, perioral erythema; no systemic involvement
Greither disease	Autosomal dominant *(KRT1)*	Transgredient PPK; may extend proximally; sometimes improves with age	Usually mild or absent	Generally non-mutilating; no systemic features
Vohwinkel syndrome	Autosomal dominant *(GJB2)*	Diffuse honeycomb PPK; constricting bands; starfish keratoses on knuckles	May show dystrophy	Sensorineural deafness common
Olmsted syndrome	AD or X-linked *(TRPV3, MBTPS2)*	Mutilating PPK; severe periorificial keratotic plaques; flexural involvement	May show dystrophy	Alopecia; severe pain; ocular and dental anomalies
Huriez syndrome	Autosomal dominant	Mild diffuse PPK with scleroatrophy	Nail hypoplasia	Increased risk of squamous cell carcinoma

(Continued)

Table 1 Continued

Disorder	Inheritance	Cutaneous Features	Nail Features	Other Features
Nagashima-type PPK	Autosomal recessive *(SERPINB7)*	Mild diffuse transgredient PPK; erythematous border; non-mutilating	Usually, absent	Generally mild; no contractures
Unna–Thost (non-epidermolytic PPK)	Autosomal dominant *(KRT1/KRT9)*	Diffuse non-gradient PPK	Minimal or absent	No systemic features
Papillon–Lefèvre syndrome	Autosomal recessive *(CTSC)*	Diffuse PPK	May show dystrophy	Severe early periodontitis and premature tooth loss

Although genetic confirmation was not possible due to financial constraints, the diagnosis of Mal de Meleda was made on strong clinical grounds. The patient demonstrated classical features of Mal de Meleda, including diffuse, sharply demarcated transgredient palmoplantar hyperkeratosis extending onto the dorsal surfaces of the hands and feet, progressive disease course, hyperhidrosis with fissuring and maceration, nail involvement, and early contracture tendency.

A thorough clinical evaluation was performed to exclude other hereditary palmoplantar keratodermas. There was no evidence of early-onset periodontitis or premature tooth loss to suggest Papillon–Lefèvre syndrome, no starfish keratoses, constriction bands, or sensorineural deafness to support Vohwinkel syndrome, no periorificial keratotic plaques or alopecia as seen in Olmsted syndrome, and no scleroatrophy or nail hypoplasia indicative of Huriez syndrome. The distinctly transgredient distribution excluded Unna–Thost PPK, while the progressive severity and mutilating tendency were inconsistent with the milder phenotype of Nagashima-type PPK or Greither disease. Therefore, despite the absence of genetic confirmation, the clinical findings in this case strongly support the diagnosis of Mal de Meleda.

Management involves systemic and topical retinoids combined with keratolytics, emollients, and antimicrobials, supplemented by patient education on skin care and prevention of hyperhidrosis and infection. Oral acitretin is highly effective in reducing hyperkeratosis, fissuring, and discomfort. In refractory cases, surgical debridement and full thickness grafting may be required.[8]

Conclusion

This case emphasizes the clinical heterogeneity and rare presentation of Mal de Meleda in a non consanguineous patient presenting in adulthood. The significant clinical improvement with oral acitretin highlights the role of early intervention in preserving mobility and quality of life. Awareness of this rare entity and its clinical manifestations can aid in early diagnosis and targeted therapy.

Ethics statement

Written informed consent was obtained from the patient for publication of this case report. Patient anonymity has been preserved.

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