Yemen Journal of Medicine

https://yemenjmed.com

Article information

DOI: 10.63475/yjm.v5i2.0400

Article history:
Received: 09 May 2026
Accepted: 30 June 2026
Published: 22 July 2026

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How to cite this article
Ayub F, Tariq H, Yasmin W, Amin U, Batool S, Asad F. Mal de Meleda masquerading as Nagashima-type palmoplantar keratoderma: A genetically confirmed case from Pakistan. Yemen J Med. Published online July 22, 2026. doi: 10.63475/yjm.v5i2.0400:1-6.

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Case Report

Mal de Meleda Masquerading as Nagashima-Type Palmoplantar Keratoderma: A Genetically Confirmed Case from Pakistan

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ABSTRACT

Mal de Meleda (MDM) is a rare autosomal recessive palmoplantar keratoderma (PPK) caused by pathogenic variants in the *SLURP1* gene. Its early clinical manifestations can overlap considerably with Nagashima-type palmoplantar keratoderma (NPPK), which is caused by biallelic variants in *SERPINB7*, creating diagnostic uncertainty. We describe a 36-year-old Pakistani male born of a consanguineous union, presenting with congenital PPK initially resembling NPPK clinically. Molecular testing using clinical exome sequencing identified a homozygous pathogenic nonsense variant in *SLURP1* (c.286C>T; p.Arg96*), confirming autosomal recessive MDM. An additional heterozygous pathogenic missense variant in *WNT10A* (c.682T>A; p.Phe228Ile) was also identified. Progressive transgradient hyperkeratosis, hyperhidrosis, malodorous maceration, and recurrent dermatophyte superinfection supported the final diagnosis. This case underscores the importance of longitudinal clinical assessment combined with molecular genetic testing in hereditary PPKs with overlapping phenotypes, particularly in consanguineous populations. No pathogenic variants were identified in SERPINB7 on clinical exome sequencing, definitively excluding NPPK. It also highlights both the value of clinical exome sequencing for accurate diagnosis and genetic counselling, and the potential relevance of additional ectodermal variants such as WNT10A detected incidentally on comprehensive genomic panels.

Key words: Mal de Meleda, palmoplantar keratoderma, *SLURP1*, Nagashima-type keratoderma, *SERPINB7*, genetic diagnosis, consanguinity

INTRODUCTION

Hereditary palmoplantar keratodermas (PPKs) represent a clinically and genetically heterogeneous group of genodermatoses characterized by hyperkeratosis of the palms and soles. Among diffuse transgradient subtypes, Mal de Meleda (MDM; OMIM #248300) and Nagashima-type palmoplantar keratoderma (NPPK; OMIM #615598) share several overlapping features that can complicate early diagnosis. [1,2]

NPPK is an autosomal recessive PPK caused by biallelic loss-of-function variants in *SERPINB7*, a serine protease inhibitor expressed in the epidermis. It presents with well-

demarcated, diffuse palmoplantar erythema and mild-to-moderate hyperkeratosis that extends (transgresses) to the dorsal surfaces, wrists, and ankles. Clinically, it is regarded as generally non-progressive. [3,4] MDM, by contrast, is caused by pathogenic variants in *SLURP1*, which encodes secreted Ly-6/uPAR-related protein-1—a key regulator of keratinocyte proliferation, terminal differentiation, and epidermal homeostasis. MDM is characteristically progressive and more severe, with massive hyperkeratosis, hyperhidrosis, malodorous maceration, fissuring, and risk of pseudo-ainhum. [5,6]

Both conditions may present in infancy with spongy palmoplantar maceration after water exposure and recurrent fungal infections, rendering early clinical differentiation difficult. This phenotypic overlap has been well described, and molecular confirmation is considered the gold standard for accurate diagnosis. [7,8]

We present a genetically confirmed case of MDM in a Pakistani male in whom early clinical features closely resembled NPPK, emphasizing the diagnostic value of molecular testing and longitudinal assessment in consanguineous populations.

CASE REPORT

A 36-year-old male from Lahore, Pakistan, born to first-cousin parents, presented with a lifelong history of progressive palmoplantar hyperkeratosis (**Figure 1**). He was clinically normal at birth. Erythema and scaling of the palms and soles developed at 18 months of age and progressively worsened over subsequent years. By early adulthood, the patient had developed massive transgradient PPK with extension to the dorsa of the hands and feet, wrists, ankles, and elbows.

Disease severity progressed markedly in adult life, characterized by marked hyperhidrosis, malodorous maceration, painful fissuring, and recurrent dermatophyte superinfection—features classically associated with MDM. [5,6] A spongy white palmoplantar appearance was evident within minutes of water immersion. No abnormalities of hair, dentition, or mucosae were noted, which helped to narrow the differential diagnosis.

The patient's son displayed similar palmoplantar changes, although the remaining four children were clinically

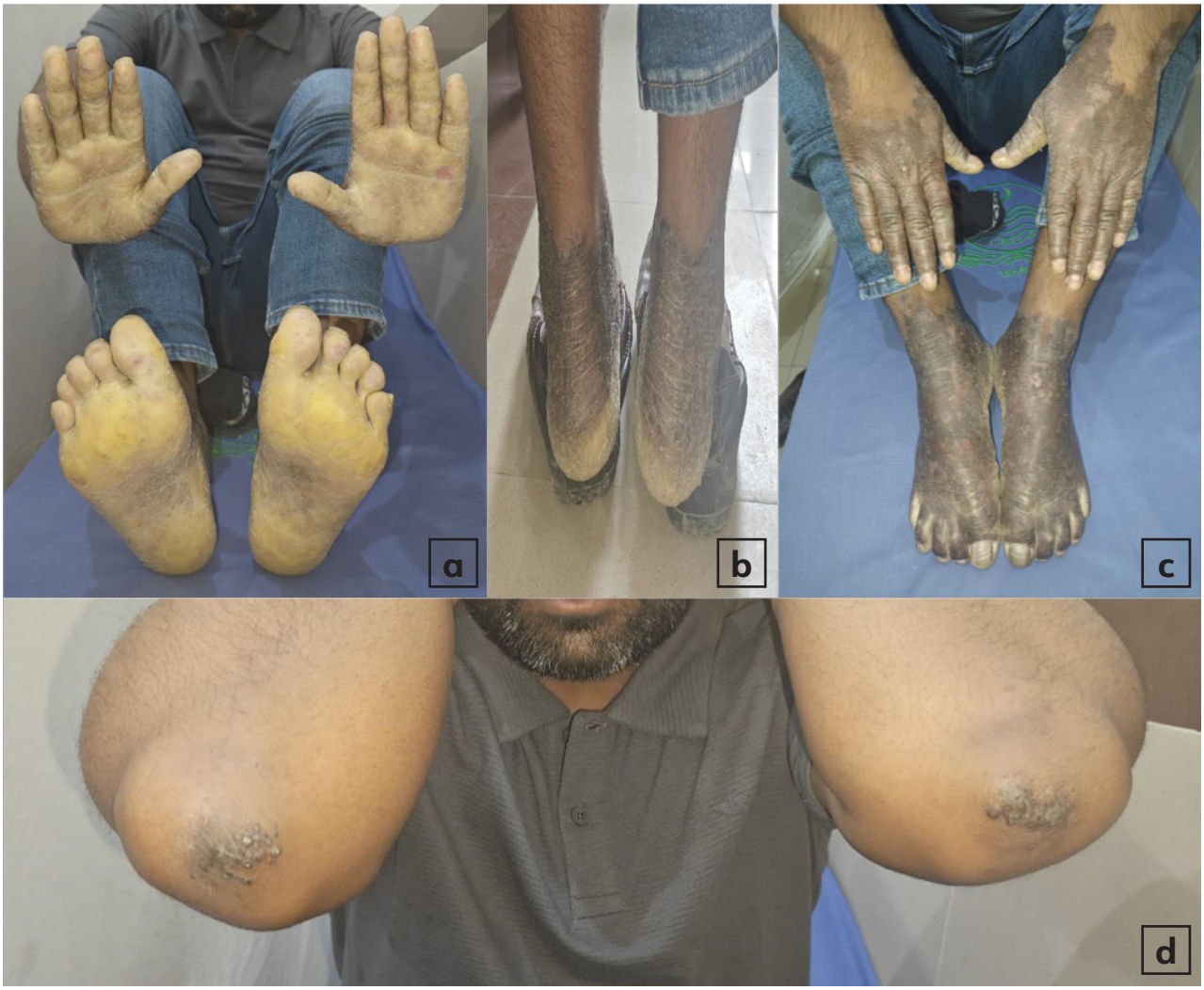

Figure 1: Clinical features of Mal de Meleda in a 36-year-old male. (a) Marked palmoplantar hyperkeratosis with well-demarcated thickening and scaling. (b) Extension of hyperkeratosis over Achilles tendon; panels (b) and (c) highlight transgradient extension beyond the wrists and ankles. (c) Diffuse, severe transgradient hyperkeratosis involving the dorsum of the hands and feet with extension beyond the wrists and ankles. (d) Extension of hyperkeratosis over elbows.

unaffected. The son was not genetically tested; his inclusion in the pedigree is based on clinical observation alone and should be interpreted as a limitation of the family evaluation. The patient's spouse was not available for carrier testing; whether she harbors a heterozygous SLURP1 variant (which would be required for autosomal recessive inheritance in the son) remains unconfirmed. Given the early clinical resemblance to NPPK and the subsequently progressive course, molecular genetic testing was pursued before a definitive diagnostic label was assigned. [7,8]

Histopathological Findings

Skin biopsy (**Figure 2**) demonstrated marked orthokeratotic hyperkeratosis with underlying psoriasiform epidermal hyperplasia and a prominent granular layer. The papillary dermis showed vertically oriented collagen bundles with a sparse perivascular lymphocytic infiltrate. No epidermolytic changes, Munro microabscesses, or other specific diagnostic features were identified. [5,9]

These histopathological findings are consistent with non-epidermolytic hereditary PPK but are not specific for either MDM or NPPK. These features are typical of non-epidermolytic diffuse PPK and have been reported in both MDM and NPPK, underlining their poor discriminatory value. No subtle inflammatory infiltrate, eccrine gland changes, or other histological features specifically reported to favor MDM over NPPK were identified, reinforcing that biopsy alone is insufficient for distinction. As reported in recent literature, biopsy findings alone are insufficient to differentiate these genodermatoses, and molecular testing remains essential for definitive classification. [8,9]

Genetic Analysis

Clinical exome sequencing (Illumina-based platform, minimum 100× mean coverage; variants confirmed by Sanger sequencing; classified per ACMG/AMP criteria) identified a homozygous pathogenic nonsense variant in *SLURP1* (c.286C>T; p.Arg96*), confirming the diagnosis of autosomal recessive MDM. The variant was classified as Pathogenic (ACMG criteria: PVS1, PM2, PP4; absent from gnomAD v4; reported in ClinVar as pathogenic). Critically, no pathogenic or likely pathogenic variants were identified in SERPINB7 on the same exome dataset, molecularly excluding NPPK. [5,6] In addition, a heterozygous pathogenic missense variant in *WNT10A* (c.682T>A; p.Phe228Ile) was detected. This specific *WNT10A* variant—one of the most frequently reported pathogenic variants in *WNT10A*-related ectodermal dysplasia—is classified as Pathogenic (ACMG: PS1, PM2, PP3; present in gnomAD v4 at low heterozygous frequency consistent with incomplete penetrance) and is known to demonstrate incomplete penetrance and variable expressivity, particularly in heterozygous carriers. [10,11]

Monoallelic *WNT10A* variants may contribute to minor ectodermal features, including subtle dental anomalies, nail changes, or palmoplantar skin alterations; however, they are insufficient on their own to explain the severe palmoplantar phenotype observed in this patient. [10,11] Familial segregation analysis was consistent with autosomal recessive inheritance of the *SLURP1* variant as the primary pathogenic driver. Notably, no targeted dental examination or panoramic radiograph was performed following the genetic result to assess for subtle ectodermal changes attributable to the WNT10A variant; this represents a limitation of the current evaluation. Although no comparable dual SLURP1–WNT10A finding has been described in the MDM literature to date, any potential gene–gene interaction remains speculative, and this dual finding should be interpreted with caution.

DISCUSSION

This case illustrates the diagnostic challenge posed by early-onset MDM, which can closely mimic NPPK in infancy and early childhood. The initial spongy maceration after water exposure, transgradient hyperkeratosis, and recurrent fungal superinfection are shared features of both conditions. [3,4,7]

The distinguishing progression to severe hyperhidrosis, malodor, massive transgradient hyperkeratosis, and painful fissuring in our patient is characteristic of MDM rather than NPPK. NPPK, caused by *SERPINB7* variants, is generally a non-progressive or mildly progressive condition, whereas MDM, driven by *SLURP1* loss of function, is characteristically progressive and more severe. [3,5,6]

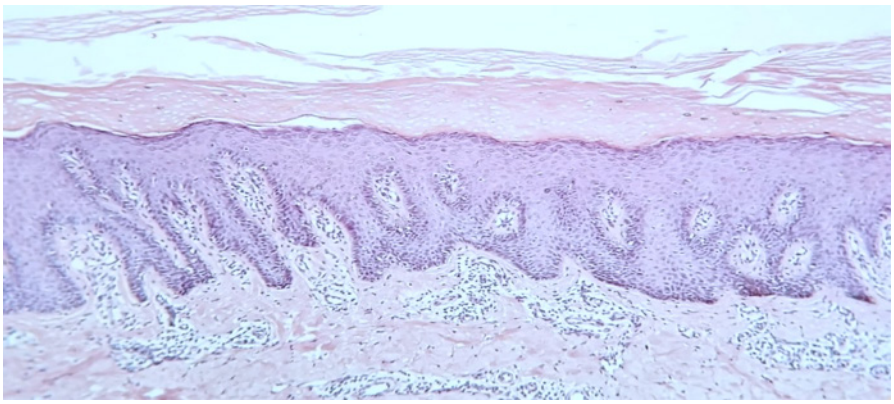

Figure 2: Histopathological features. Hematoxylin and eosin-stained section showing marked orthokeratotic hyperkeratosis with psoriasiform epidermal hyperplasia and a prominent granular layer. The papillary dermis demonstrates vertical collagen bundles with sparse perivascular lymphocytic infiltrate. No epidermolytic changes or Munro microabscesses are identified. Original magnification ×100 (overview) and ×200 (detail). No eccrine gland changes or specific inflammatory patterns favoring Mal de Meleda over Nagashima-type palmoplantar keratoderma were identified.

Recent work has expanded understanding of both conditions. A 2025 comprehensive review of NPPK history and pathogenesis confirmed that founder *SERPINB7* variants predominate in East Asian populations but have also been reported in Arab-Muslim individuals—highlighting the geographic spread of these founder mutations beyond their original populations. [4] Meanwhile, multiple recent case reports and mutation analyses have expanded the spectrum of *SLURP1* variants in MDM, including those from Asian and South Asian populations. [6,8,12]

Histopathological analysis, while demonstrating orthokeratotic hyperkeratosis consistent with non-epidermolytic PPK, was nondiagnostic for MDM versus NPPK. This limitation has been consistently documented in the literature and supports the current consensus that molecular testing is the gold standard for differentiating hereditary PPKs with overlapping phenotypes. [2,9]

The detection of a heterozygous *WNT10A* c.682T>A (p.Phe228Ile) variant in this patient is noteworthy. This is among the most prevalent pathogenic *WNT10A* variants reported in ectodermal dysplasia cohorts, and heterozygous carriers can exhibit variable expressivity affecting skin, hair, nails, and dentition. [10,11] The absence of clinically evident dental, hair, or nail abnormalities in this patient may reflect the known incomplete penetrance of monoallelic *WNT10A* pathogenic variants. Nonetheless, the phenotypic contribution of the *WNT10A* variant cannot be entirely excluded and may have contributed to phenotypic severity or heterogeneity.

Reports of MDM from Pakistan are few, and genetically confirmed cases are rarer still. In 2025, Saaqib et al. published the first familial case series of MDM from Pakistan, describing three female siblings aged 9 to 11 years born of consanguineous parents. [13] All three demonstrated transgradient hyperkeratosis, nail changes, and hyperhidrosis, with a similarly affected paternal aunt supporting autosomal recessive inheritance. However, unlike our case, molecular testing was not performed, and the diagnosis rested on clinical and family history criteria alone. To our knowledge, based on a systematic search of PubMed and EMBASE, this represents among the earliest exome-confirmed reports of MDM from Pakistan carrying a defined SLURP1 pathogenic variant (p.Arg96*), providing molecular validation that complements the clinical literature emerging from the region. In brief, prior South Asian reports have relied on clinical and histological criteria alone, whereas our case adds a molecularly validated diagnosis alongside a dual-gene finding.

Within the broader South Asian context, Sethy et al. (India, 2022) reported an atypical case of MDM in a 28-year-old female with flexural—rather than purely extensor—involvement, a finding not previously documented in the literature. [14] Genetic testing for *SLURP1* could not be performed owing to resource constraints, and clinical diagnosis was based on characteristic histopathology showing orthokeratotic hyperkeratosis with psoriasiform hyperplasia and a prominent granular layer—findings strikingly similar to those in our patient's biopsy. This case highlights a recurring limitation in South Asian MDM reporting: the absence of molecular confirmation. In contrast, our case demonstrates that clinical exome sequencing is feasible in a Pakistani clinical setting and should be pursued to achieve definitive diagnostic accuracy.

Somasundaram et al. (India, 2025) reported a novel *SLURP1* splice-site variant (c.58+1G>T) in a 27-year-old South Indian woman born to third-degree consanguineous parents, who also presented with reticulate hyperpigmentation on the dorsum of the feet—an atypical feature not previously reported in MDM. [15] This case reinforces the phenotypic breadth of MDM across South Asia and underscores that consanguinity remains a consistent epidemiological driver of the condition in this region. Our patient similarly arose from a consanguineous background, and the identification of a distinct nonsense variant (p.Arg96*) further expands the mutation spectrum of *SLURP1* reported from South Asia.

Kalyan et al. (India, 2021) described two South Indian siblings, aged 6 and 9 years, with MDM presenting with congenital-onset PPK and second-degree family consanguinity. [16] Histopathology in their report also showed hyperkeratosis, hypergranulosis, and acanthosis—consistent with our findings—but genetic testing was not performed. Taken together, the South Asian literature on MDM is characterized by clinical heterogeneity, consistent consanguinity as a risk factor, variable severity, and a near-universal absence of molecular confirmation, a gap our case begins to address.

Wang et al. (China, 2023) identified novel compound heterozygous *SLURP1* variants (c.243C>A and c.256G>A) in a Chinese patient, alongside a homozygous variant (c.211C>T) in a second unrelated family. [6] Both patients exhibited typical progressive PPK with transgradient extension. The c.256G>A variant (p.Gly86Arg) has been the predominant mutation detected in Chinese MDM patients and has also been identified in a 2022 case confirmed by whole-exome and whole-genome sequencing from China (Dai et al.), in which the patient additionally underwent therapeutic trials with ixekizumab and adalimumab with mild improvement of inflammatory erythema. [8] The variant identified in our patient (p.Arg96*) is a distinct nonsense mutation that has also been reported in a Turkish patient (p.Arg96Pro, a different substitution at the same codon), highlighting this residue as a mutational hotspot within exon 3 of *SLURP1*. [17]

Khan et al. (India, 2024) described an 18-year-old male with the transgradient variant of MDM presenting with yellowish thickened skin of the palms and soles managed with acitretin and topical emollients. [12] Although genetic testing was not performed, the clinical description closely aligns with our case, particularly the transgradient extension to the dorsa of the hands and feet and the characteristic yellowish discoloration. The response to systemic retinoids in this and other cases provides a useful therapeutic benchmark.

Guevara et al. (2023) described an atypical 42-year-old Hispanic male with asymmetric MDM managed with isotretinoin in a setting where acitretin and genetic testing were inaccessible. [18] The asymmetric and initially fingertip-predominant presentation in their patient contrasts with the symmetric, palm- and sole-predominant onset in our case, illustrating phenotypic variability across different ethnic and geographical backgrounds. The lack of molecular confirmation in the Hispanic case, due to healthcare resource

limitations, again underscores the global diagnostic gap that next-generation sequencing can address.

Pospischil et al. (Austria, 2022) reported a European MDM case with progressive palmoplantar hyperkeratosis, confirming the persistence of MDM case reports from European centers decades after the original Croatian island (Mljet/Meleda) description. [19] A 2026 Romanian case report described MDM in a 75-year-old patient who had lived undiagnosed for over seven decades, illustrating how MDM may go unrecognized even in European healthcare systems. [19] These cases collectively reinforce that MDM diagnosis is delayed worldwide—not just in low-resource settings—and that clinical awareness and molecular confirmation are needed globally.

The detection of a heterozygous *WNT10A* c.682T>A (p.Phe228Ile) variant in our patient is noteworthy, as discussed in the Genetic Analysis section. The clinical significance of this coexisting heterozygous variant remains uncertain and may represent an incidental finding; its potential contribution to phenotypic severity in the context of a co-existing primary *SLURP1* pathogenic variant cannot be entirely excluded but remains speculative. Although no comparable dual-gene finding has been described in the MDM literature, this observation should be interpreted cautiously without further functional or familial evidence.

Management of MDM globally has relied primarily on systemic retinoids (acitretin or isotretinoin), topical keratolytics, and infection control. Oral acitretin is regarded as the most effective agent in reducing hyperkeratosis, though its teratogenicity and hepatotoxicity limit long-term use. [18,19] The 2022 Chinese case by Dai et al. explored novel therapeutic avenues, with ixekizumab (an IL-17A inhibitor) providing mild improvement of inflammatory erythema—suggesting that the inflammatory component of MDM may be partially amenable to biologic therapy. [8] Our patient was managed conservatively with topical keratolytics (20% urea cream) and antifungal therapy during the diagnostic phase; systemic retinoid (acitretin) was recommended following molecular confirmation. However, the patient declined systemic treatment. The ongoing management plan includes long-term topical keratolytics, antifungal therapy as needed for recurrent dermatophyte infections, patient and family education regarding the genetic basis and inheritance pattern of the condition, and regular dermatological follow-up. The patient was counselled regarding footwear modification to reduce friction-related fissuring, and psychosocial support was discussed given the impact of chronic skin disease on quality of life. At last follow-up (approximately three months after genetic diagnosis), disease activity had remained broadly stable on topical therapy alone, though the patient reported continued functional limitation due to painful fissuring and malodor. Reconsidering systemic retinoid therapy remains an open discussion point for future consultations.

This case contributes to the molecular epidemiology of MDM and reinforces the role of next-generation sequencing—particularly clinical exome sequencing—as an efficient and comprehensive first-line diagnostic tool in hereditary skin disorders with overlapping phenotypes, especially in consanguineous populations.

CONCLUSIONS

MDM should be considered in patients presenting with early-onset diffuse transgradient PPK, even when the initial clinical picture resembles NPPK. Longitudinal follow-up combined with molecular genetic testing is essential for accurate diagnosis, prognostication, and genetic counselling. This genetically confirmed case from South Asia adds to the limited literature on MDM in the region and highlights the indispensable role of molecular diagnostics in hereditary genodermatoses with overlapping phenotypes.

CONSENT

Written informed consent was obtained from the patient for publication of this case report.

AUTHORS' CONTRIBUTION

All authors have significantly contributed to the work, whether by following the case at the bedside, conducting literature searches, drafting, revising, or critically reviewing the article. They have given their final approval of the version to be published, have agreed with the journal to which the article has been submitted, and agree to be accountable for all aspects of the work.

SOURCE OF FUNDING

None.

CONFLICT OF INTEREST

None.

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