

## A Different Presentation of Mal De Meleda: New Skin Lesions in a Residual Limb after Traumatic Amputation

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**ABSTRACT** Mal de Meleda is a rare autosomal recessive skin disease which is known as keratoderma palmoplantaris transgradiens. Here we report a case of Mal de Meleda who had skin lesions in the residual limb and pseudoainhum in the thigh after traumatic lower leg amputation. A 71-year-old female was admitted to our tertiary hospital for prosthetic rehabilitation. On the physical examination, thickening of the skin on palms, left sole and residual limb was present. The patient reported that she had these skin lesions since infancy and she realized new skin lesions after amputation in the residual limb. We requested dermatology consultation and she was diagnosed as Mal de Meleda. To our knowledge, this is the first Mal de Meleda case in the literature with new lesions at the residual limb. Although exact pathophysiological mechanisms are not well known in Mal de Meleda, prosthesis use might have accelerated disease process in our patient.

**KEY WORDS:** Mal de Meleda, residual limb, traumatic amputation

### INTRODUCTION

Mal de Meleda is a rare autosomal recessive skin disease which is known as keratoderma palmoplantaris transgradiens (1). Most cases of the disease are reported among inhabitants of island of Mljet (Meleda), Croatia (2), but recently other geographical case series and sporadic cases have been reported from countries such as Turkey, Japan, and Libya (3-5).

Mal de Meleda disease has obligatory and facultative clinical features (6). The obligatory features are [1] autosomal recessive inheritance, [2] transgressive and progressive nature of the keratoderma, and [3] onset of diffuse keratoderma palmoplantaris soon after birth. The facultative features are [1] palmoplantar hyperhidrosis, [2] lichenoid polycyclic plaques on the elbows, knees, and groin, [3] pitting in the keratoderma palmoplantaris, [4] subungual keratosis, koilonychia, and dystrophy of the great toenail, [5] progressive

conical tapering of the fingertips which may lead to contractures of the fingers, [6] perioral erythema, [7] high arched palate, and [8] corneal lesions.

To our knowledge, new skin lesions in the residual limb and pseudoainhum at the thigh due to prosthesis use have not yet been reported. Herein we report a case of Mal de Meleda with skin lesions in the residual limb and pseudoainhum in the thigh after traumatic lower leg amputation.

### CASE REPORT

A 71-year-old woman was admitted to our tertiary hospital for prosthetic rehabilitation. She had worn a below-knee prosthesis for 20 years because of traumatic amputation of the leg caused by a motor vehicle accident. On physical examination, thickening



**Figure 1.** Plantar keratoderma of the left foot.

of the skin on the palms, left sole, and residual limb was present. There was diffuse palmoplantar keratoderma involving bilateral palms, the left sole, and the residual limb. Keratoderma extended proximally up to the ankle, dorsum of the hands, and the left foot with irregular margins and prominent knuckle pads. There were multiple erosions on the sole as well as fissures and keratolysis. There was also conical tapering of the fingertips leading to contractures. Flexion deformities were noted in the fingers. Digital constriction was mild in the little fingers of the both hands. Subungual keratosis and dystrophy of the nails (including the great toenail) were noted (Figure 1, Figure 2, and Figure 3). In addition to these features, the thigh of the amputated leg was thinned (Figure 3). The patient also had pigmented, rough, raised group lesions on the elbows and knees. The patient reported that she had had these skin lesions since infancy and that one of her siblings suffered from the same skin lesions. She noticed new skin lesions on the residual limb after amputation, which occurred before wearing the first prosthesis. She also had a constricting band on her thigh after the first year of the prosthesis usage. We did not find perioral erythema, palmoplantar hyperhidrosis, or high arched palate. There was no abnormality on laboratory investigation. We requested dermatology consultation, and the patient was diagnosed with Mal de Meleda.

## DISCUSSION

Palmoplantar keratodermal diseases are a heterogeneous group which present with hyperkeratosis on the distal regions of the body such as the palms



**Figure 2.** Keratoderma of both hands, conical tapering of the distal digits, and thickening dystrophic changes.

and soles. The disease pattern may vary from barely detectable signs to high morbidity which impairs the quality of life (7,8). Mal de Meleda is a rare form of palmoplantar keratodermal diseases. It usually presents soon after birth (or up to 3 years of age), and the dorsal sides of the hands and feet are also affected in addition to palmoplantar surfaces. A sharp demarcation which is called “glove and stocking keratoderma”



**Figure 3.** Keratoderma in the residual limb and thinning of the right thigh.

may be seen in the extremities (9). Hyperkeratotic plaques like knuckle pads may be seen on the interphalangeal joints (1). Patients may also experience pain due to fissures, hyperhidrosis with macerated skin, malodor, keratotic plaques over the joints, and nail changes. Brachydactyly, perioral erythema, and pseudoainhum are other characteristics of the disease (10). Our patient had hyperkeratosis not only on the palmoplantar regions, but also on the residual limb. Acquired forms of palmoplantar keratodermas may come to mind because of prosthesis use in our patient. But the palmar lesions which occurred spontaneously and the family history of the case excluded acquired forms of keratoderma.

On the other hand, pseudoainhum is a rare disease entity. It usually begins with a circular groove, constricting band, or crease (11) and can progress to auto-amputation of the involved body segment. Pseudoainhum is due to three etiologies: developmental anomalies present at birth are primary pseudoainhum. Secondary pseudoainhum which develops in later stages of life is a result of an identifiable disease. Trauma and mechanical injury such as burns and scar formation from frostbite can cause the third variant (12-14). In this etiological view, our case is supposed to be the primary type, but the constricting effect of the socket might have contributed to pseudoainhum formation.

Although digital involvement is typical, the limbs, trunk or other body regions may also be affected in pseudoainhum (11). A stump is not a usual localization for the disease but our patient had pseudoainhum on the residual limb. She recognized the lesion after using the prosthetic device. This history supports the role of a socket in pseudoainhum formation.

## CONCLUSION

To our knowledge, this is the first Mal de Meleda case in the literature with new lesions on the residual limb. Although exact the pathophysiological mechanisms are not well known in Mal de Meleda, prosthesis use might have accelerated the disease process in our patient.

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