Olmsted syndrome: a rare cause of palmo-plantar keratoderma

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Olmsted syndrome is a rare disorder of unknown aetiology characterised by a dense palmo-plantar keratoderma with periorificial lesions. To date only around 32 cases have been reported in the literature. A case of the disease, with painful deep fissuring of the soles and palms, in a seven-year-old boy is presented here.

A seven-year-old boy presented to the podiatry clinic in Nicosia, Cyprus, with a history of marked hyperkeratosis with fissuring of the palms and soles that had been present since the age of three months. The boy was born prematurely to healthy parents, with no family history of skin problems, at seven months by caesarean section due to maternal pre-eclampsia.

At two days old, numerous blisters developed across his whole body, subsequently ulcerating and gradually healing over many weeks. The boy had normal nails at birth and he had hair, although at six months he developed alopecia and minor nail changes.

On examination, the patient was slightly underweight although his height was normal for his age. The nails of the fingers and toes were dystrophic, thickened with discolouration and hair was absent. No abnormalities were evident with his dentition. Circulation and sensation of the feet appeared normal.

His hands and feet exhibited gross hyperkeratosis accompanied by deep painful fissuring (Figures 1-3). The patient found it difficult to walk due to the pain, and fissured palms made writing difficult but generally he was able to play and interact with his friends. Contracture deformities of the digits were noted in both hands and deep fissures on wrists and the palmar area of the metacarpal-phalangeal joints reduced the patient's ability to fully extend his fingers due the pain he experienced. Overlapping and clawing of the lesser toes were also present, with inter-digital maceration.

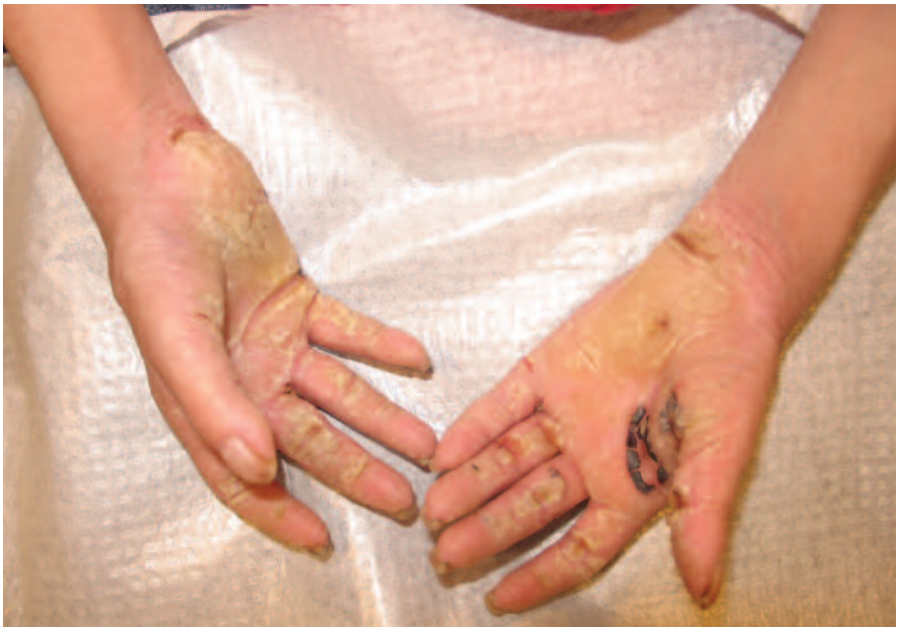


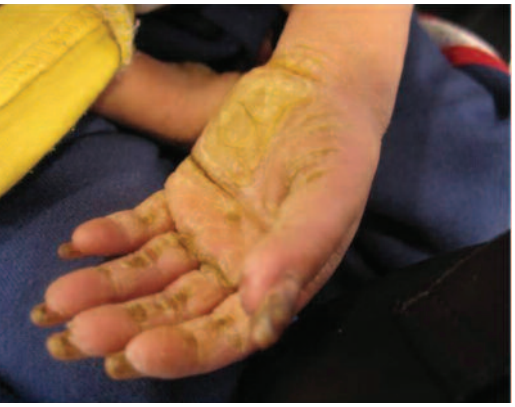
Figure 1. Hyperkeratosis and fissuring of the palms.

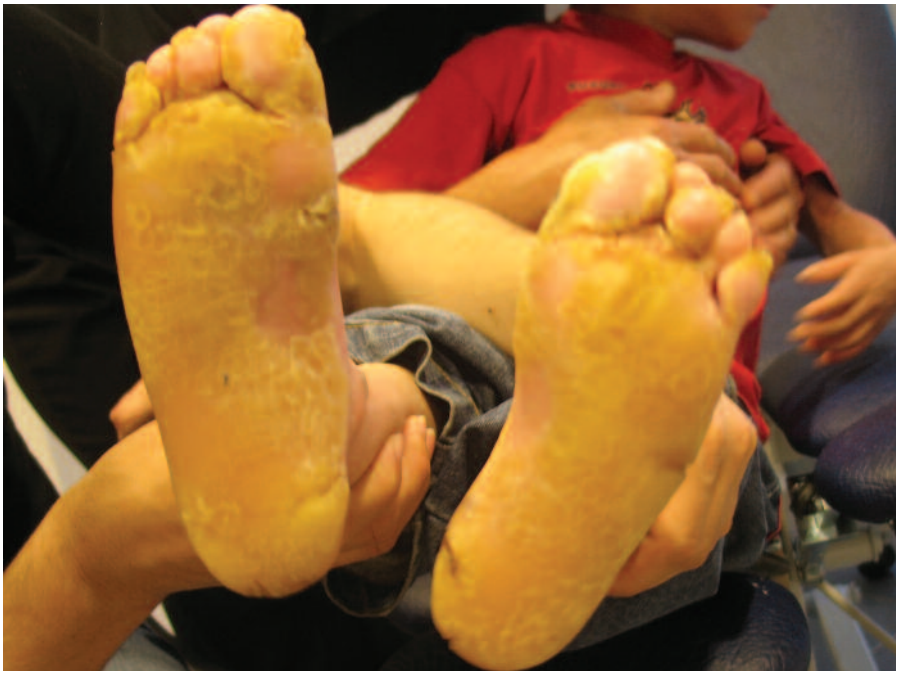
The patient reported frequent bouts of blistering on the soles of his feet. Otherwise, his growth and development were normal and he was able to attend mainstream education with some additional tutoring.

At the time of presentation, he was on no regular prescribed medications other than topical medicaments – emollients and softening agents for his skin condition.

MANAGEMENT

Management of the condition has proved to be a challenge. Before commencing treatment his hands and feet are soaked in water to soften the lesions and this subsequently makes debridement of the affected areas easier. Various creams and emollients have been used in the past. An





Figures 2 & 3. Hyperkeratosis and fissuring of the palms and soles.

improvement is recorded each time a new emollient is used; however, the effectiveness of the emollient decreases over time.

The most effective regime appeared to be mechanical debridement of the hyperkeratotic lesions and nail care three times a month. Urea-based creams, used twice daily, together with astringents have helped to reduce the maceration of the skin between the toes and reduce the occurrence of fissures at that area. Each treatment session can take up to 90 minutes. The condition of the skin improves during the summer months and it has been found that going to the beach helps greatly.

DISCUSSION

Olmsted syndrome was first mentioned in 1927 by H Olmsted[1] who described a case of an infant with a mutilating palmo-plantar keratoderma (PPK) accompanied by periorificial keratotic lesions of the mouth and anus. Later papers describing this condition labelled the disease as 'Olmsted syndrome' or 'PPK with periorificial keratosis'.

To date around 32 cases have been reported in the medical literature, making Olmsted syndrome a rare, congenital disorder demonstrated to be more common in males by a ratio of 2:1.[2]

Case reports have stated that the condition usually appears in the first few years of life[3] when a child begins to walk and grasp[4,5] as a mutilating palmo-plantar keratoderma with the development of sharply demarcated hyperkeratotic verrucous plaques around the mouth, nasal folds and other body orifices, although a few cases have been reported with later presentations.[6]

Typically the lesions are symmetrical and are frequently accompanied by a generalised hair thinning or complete loss (alopecia). Reported features are summarised in Table 1.

The course of the condition is gradual and progressive and is accompanied by major disability.[7] Extensive development of hyperkeratosis and fixed-flexion deformities of the digits are often accompanied by an Ainhum-like auto-amputation of the digits.[8]

Diagnosis

There are many types of PPK in existence. Diagnosis of these is usually based on a clinical assessment reinforced with family history and, where available, genetic analysis. Due to the specialist nature of these, only a few centres are able to offer this service.

Histological examination through skin biopsy is often general and non-specific for all but a few types of PPK. As no gene has been identified for Olmsted syndrome,[9] genetic testing is of little value.

Differential diagnoses may include other PPKs such as Mal de Maleda, Vohwinkels Syndrome and Zinc deficiency (acrodermatitis enteropathica). However, as periorificial keratoses are virtually unique to the condition, diagnosis is typically a straightforward process,[10] particularly in a patient with no family history as most cases are sporadic.

Aetiology

The exact aetiology of Olmsted syndrome remains a mystery, with suggestions made that the condition results as a defect of keratinsation.[11,12] Studies on skin samples carried out by Kress[13] have shown that immature keratins 5 and 14 are expressed in the upper layers of the epidermis when normally they would be restricted to the basal layer. Fonseca and colleagues[14] also noted a similar pattern but isolated abnormal keratins in unaffected as well as affected skin, concordant with the widespread nature of the disease, suggesting that external factors such as trauma may predict hyperkeratotic lesion location.

Although most cases of the disease appear to be sporadic, Yaghoobi *et al*[12] have reported two cases occurring within the same family. Moreover, data obtained by Requena and co-workers[5] also document four cases with a familial link. This potentially suggests genetic transmission in some cases, although the mode of inheritance is unknown, but possibly recessive in nature.

Associated malignancy

Within published literature, there is evidence of an association between palmo-plantar keratoderma and malignancy.[10,15] Nakajima and colleagues[16] reviewed 18 published cases of melanoma arising in the hyperkeratotic plaques of patients with PPK. Interestingly, as the authors report, the majority of cases (13 out of 18) arose in Japanese patients, regardless of the type of PPK exhibited, possibly correlating to

Consistent features

- Symmetrical mutilating palmo-plantar keratoderma
- Periorificial hyperkeratotic plaques (nasal fold, mouth, anus)

Inconsistent features

- Nail dystrophy
- Chronic paronychia
- Fixed flexural deformities of the digits
- Ainhum-like auto-amputation of digits
- Hair thinning, whitening (leucotrichosis) or total loss (alopecia)
- Hypohidrosis / Hyperhidrosis
- White plaques on oral mucosa (leukokeratosis)
- Verrucous plaques of the axillae and nuchal folds
- Hyperkeratotic linear streaks (elbows, knees, flexures)
- Joint laxity
- Short stature

Table 1. Features described in patients with Olmsted syndrome.

acral lentiginous melanoma, which has a high prevalence in the Japanese population compared with white populations.[17] The reason for the association remains unclear. To date, however, there has only been one recorded case of malignant melanoma in a patient with Olmsted syndrome.[7]

Treatment

In most cases, the literature documents topical treatments such as emollients, vitamins A & E, keratolytics and corticosteroids as only offering temporary relief of the symptoms by reducing or softening the keratotic skin lesions.[6,11] Requena *et al*[5] suggest that surgical removal of the lesions and subsequent skin grafting may offer improvement of movement; however this has been followed by the recurrence of hyperkeratosis in the skin graft as a result of the Koebner phenomenon.[7]

Etretinate, a retinoid, has proved to be effective, reducing lesion thickness in some cases, although the keratoderma recurred in fewer than five weeks after cessation of the drug.[7] Additionally, reported side-effects in children include growth retardation caused by premature epiphyseal closure.[18]

Improvement of the condition in the summer months was noted by the patient in this case study and has not been previously reported. It could be suggested that increased exposure to solar UVA/UVB radiation may possibly reduce cell turnover, as observed in psoriasis.

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