Journal of the Turkish Academy of Dermatology
eISSN 1307-394X

Case Report
DOI: 10.6003/jtad.1483c6

Olmsted Syndrome Associated with Pseudomonilethrix: A New Case from Turkey

Kıymet Handan Kelekci,[1]* MD, Kadriye Koç,[2] MD, Mihriban Gürbüzel,[3] MD, Mehmet Özeren,[2] MD

Address: [1]Department of Dermatology, Katip Çelebi University School of Medicine, İzmir, [2]Department of Dermatology and [3]Department of Pathology, Haseki Research and Traning Hospital, İstanbul, Turkey.

E-mail: drhandankelekci@yahoo.com

** Corresponding Author:* Kıymet Handan Kelekci, MD, İzmir Katip Çelebi University School of Medicine, Department of Dermatology, Haseki Research and Traning Hospital, Department of Dermatology, İstanbul, Turkey

Published:
J Turk Acad Dermatol 2014; **8 (3)**: 1483c6
This article is available from: http://www.jtad.org/2014/3/jtad1483c6.pdf
Key Words: A..

Abstract

Observation: Olmsted syndrome (OS) is a very rare genetic disorder, which includes diffuse alopecia, leukokeratose of oral mucosa, onychodystrophy , hyperkeratotic linear streaks, follicular keratosis and anhidrosis. Pseudomonilethrix shaft anomaly is characterized by hair shafts with elliptical nodes at variable intervals and separated by narrower internodes. To the best of our knowledge, we present here the first documented case of OS associated with pseudomonilethrix hair shaft abnormality.

Introduction

Olmsted syndrome (OS) is a very rare genetic disorder, whose original description of the disorder included a combination of bilateral, mutilating, palmoplantar keratoderma (PPK) and periorificial hyperkeratotic plaques with flexion deformities of the digits, leading to constriction or spontaneous amputation [1]. Most cases are sporadic, but there is occasional familiar occurrence. The genetics underlying this syndrome have not been elucidated yet [2]. Clinical features of OS include diffuse alopecia, leukokeratose of oral mucosa, onychodystrophy, hyperkeratotic linear streaks, follicular keratosis and anhidrosis. Systemic associations include large joint laxity, absent premolar teeth, hearing loss for high frequencies, corneal dysplasia and primary sclerosing cholangitis [3].

Pseudomonilethrix shaft anomaly is characterized by hair shafts with elliptical nodes at variable intervals and separated by narrower internodes. No follicular papules can be seen on physical examination. Inheritance is usually autosomal dominant with penetrance and variable expressivity. Pseudomonilethrix usually starts in the first months of life, alt-

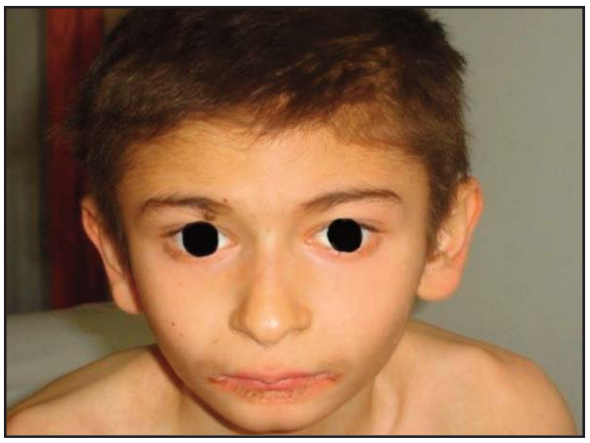

Figure 1. Erythematous hyperkeratosis of the perioral and sparse, and brittle hair.

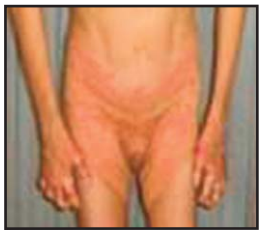

Figure 2. Bilaterally perigroin localised symmetrical, sharply defined thick plaques and diffuse palmar keratoderma

hough, in some cases, it does not become apparent until childhood [**4**].

Case Report

A 13-year-old boy was admitted to the outpatient clinic with the complaints of painful thickened skin over her palms and soles. After 6 months, the patient's hand skin under the nail and finger ends inflamed and started peeling off. Although he started walking when he was 18 months old, he had difficulty in walking due to the thickening and ragat formation of the feet skin. Both fifth fingers of each feet had developed autoamputation 5 years ago. Dermatologic examination revealed sparse, lusterless and brittle hair, erythematous hyperkeratosis of the perioral, and perigroin areas, axilla and knee, bilaterally symmetrical, sharply defined thick and diffuse palmoplantar painful keratoderma with flexion deformities of the digits of hands and feet. All the nails were hyperkeratotic, with transverse fissuring and there was clubbing. No abnormality in teeth, oral, genital and ocular mucous membranes was detected, hearing functions were normal. (**Figures 1, 2**). There was some growth retardation. Detailed pediatric examination was completely normal including electrocardiographic, echocardiographic and abdominal ultrasonographic examinations. His brother, who was died eight years ago also had palmoplantar keratoderma and periorificial keratotic plaques like our patient. We learned that his parents are within the third degree of consanguinity. Laboratory findings, including serum zinc level were in normal limits. A biopsy of the skin performed from the inguinal area. Histopathological study showed hyperkeratosis, parakeratosis, mild acanthosis, and papillomatosis. There was mild perivascular lymphocytic infiltration in the dermis. Another biopsy specimen from right planter area revealed massive hyperkeratosis, parakeratosis, acanthosis and prominent papillomatosis. The dermis showed moderate acute and chronic inflammation. Dermatoscopic examination of his scalp hair resulted in scanty and thin hair from birth with gradually amelioration. Light microscopic examination of his scalp

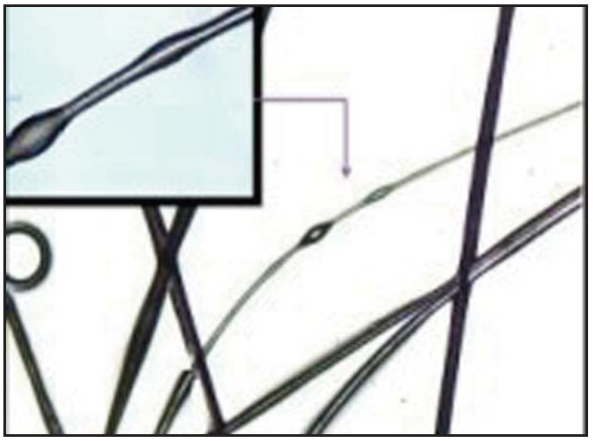

Figure 3. Pseudomonilethrix hair shaft abnormality (100 X)

hair showed pseudomonilethrix hair shaft abnormality (100 X) (**Figure 3**).

Discussion

OS was first described by *H.C. Olmsted* in 1927. The syndrome was characterized by the combination of periorificial plaques and bilateral palmoplantar transgredient keratoderma [**5**].The exact mode of inheritance is unknown, but the majority of instances are thought to be sporadic occasional familiar [**1, 6**]. An autosomal dominant or X- linked dominant trait or X- linked recessive transmission have been suggested. Finally, there may be more than one mode of inheritance in OS [**7**]. OS usually begins in the first 6 months of life by PPK. However, a late onset at the ages of 13 and 30 has been reported [**2, 5, 8**]. The pathogenesis of this disorder is still poorly understood, but there is cytochemical evidence of hyperproliferation of the epidermis [**1**]. Several genes implicated in the pathogenesis of other PPKs including KRT1, GJB2, SLURP1, and LOR were found normal at least in one patient [**7**]. Kress et al found a defect in the expression of mature epidermal keratins (types 1 and 10) and persistence of acidic keratins (types 5 and 14) in the involved epidermis. They suggested that the alteration in keratin expression is seen to be specific to the epidermis and these keratins are expressed normally in other sites [**6, 9**]. The failure to differentiate into mature keratinocytes is responsible for excessive deposition of acidic keratin 5 and 14, which is detected by monoclonal antikeratin antibody AE1 [**10, 11**]. Cutaneous findings of OS' which are less common include leukokeratosis

of the oral mucosa, diffuse alopecia, sparse hair, nail distrophy, hyperhidrosis of the palms and soles, hypohydrosis, hypotichosis, anhidrosis, hyperkeratotic linear streaks on the elbows, knees, axillae and antecubital fossae [**6, 10, 11**]. Other associations are variable that there have been flexion deformity of the digits, autoamputation of digits with constriction, large joint laxity, absent premolar teeth, hearing loss for high frequencies, chronic blepharitis, corneal epithelial dysplasia, primary sclerosing cholangitis and small stature [**3, 5, 12**].

The growth development delay in patients with OS is unclear whether the growth delay is a feature of the syndrome, secondary to the chronicity of the disease or the severe pain associated with PPK [**8**]. Growth of our case was in border-line. PPK may be the cause of restriction of social activity because painful walking and manuel skills. There is no satisfactory treatment for this condition. Acitretin has been reported with a result of partial improvement. For the patients unresponsive to acitretin treatment, full thickness excision of hyperkeratotic plaques followed by skin grafting is another therapeutic option to alleviate the pain and to arrest the progress of the disease [**9**].

Dry hair, lanugo, hypotrichia and congenital universal alopecia are reported as related hair anomalies in OS. Dogra et al. reported that light and scanning electron microscopic examination of the hair revealed several hair shaft abnormalities [**13**]. *Cambiaghi* et al. has reported that scanning electron microscopy of hair revealed changes such as twisting of hair shafts, trichorhexis nodosa, transverse fractures, and disturbance of the cuticle cell pattern, ranging from the presence of abnormal cells to complete absence of the cuticle; transverse fractures across the hair shaft through the cuticle cells were observed in two monozygotic twins [**14**]. *Mevorah* et al. has reported that seven specimens showed pili torti and another seven trichorrhexis nodosa type defects among 100 hairs from the sparse area but such defects were not seen in 100 hairs from a normal area [**7**].

Pseudomonilethrix is a rare hair shaft abnormality characterized by small beaded globular swellings. Pseudomonilethrix has been classiffied as familiar, acquired, and iatrogenic. Pili torti or partial forms of trichorrhexis nodosa have been associated with the acquired type of this hair disorder. Differential diagnosis with monilethrix is needed. Monilethrix is a disorder characterized by beaded appearance of the hair due to periodic thinning of the shaft disorder with hair fragility and patchy dystrophic alopecia. Keratosis pilaris is almost invariably associated with monilethrix unlike pseudomonilethrix. Therapy of psuedomonilethrix is recommended to avoid all trauma [**4, 15, 16**]. Our case has mild course hair problems with mutilan palmoplantar keratoderma.

We believe that the explanation of OS genetic constitution in the future which is encountered rarely will shed light on the explanation of the hair anomalies that accompanies OS frequently. To the best of our knowledge, we present here the first documented case of OS associated with pseudomonilethrix hair shaft abnormality. Although pseudomonilethrix may be have different causes, we wanted to emphasize that OS may be associated with hair shaft anomaly with similar to the pathogenesis. However this relationship needs to be supported by new cases. By means of this rare and challenging case, the relationship between OS and hair shaft abnormality especially pseudomonilethrix has been discussed.

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