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Case Report

Olmsted Syndrome Managed with Surgery-A Case Report

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Abstract

Olmsted disease is a rare genetic disorder, which is characterized by symmetrical mutilating palmoplantar keratoderma and perioral hyperkeratosis. First reported and described by Olmsted in 1927. Here we describe 3 cases from a family, who presented with palmoplantar, perioral and perianal hyperkeratosis, with one patient had spontaneous mutilation of right 5th toe.

Keywords: Olmsted syndrome; Anorexia; Parakeratosis

Case Presentation

A 12 year, eldest daughter born out of non consanguineous marriage, to healthy parents by normal vaginal delivery hailing from khordha, Odisha, India. She presented with hyper scaling and thickening of her bilateral sole for past 11 years. A small erythematous lesion at the age of 1 year on her right sole, noticed by her parents which got gradually replaced by itchy scales, over a period of 2 years affecting the entire sole followed by left sole. At the age of 4 years her palms got affected followed by perioral and perianal skin. The mutilating lesion is progressing and interferes with walking for past 3 years with spontaneous amputation of Right 5th toe. She was primarily treated with medicines in form of oral zinc and topical keratolytics. Examination revealed massive, verrucose, uneven, fissured, horny hard hyperkeratotic plaques with erythematous border over plantar aspect and extending to dorsal aspect of foot. Dystrophic toe nails with subungual hyperkeratosis. Both palms show hyperkeratosis extending to the dorsum of hand. Well defined hyperkeratotic plaques in perioral areas and perineum.

Her other 2 siblings, 6-year-old monozygotic twins, had a similar history, progression of disease and similar examination findings. Provisional diagnosis was made as Olmsted Syndrome. The medical treatment offered to patients in the form of zinc supplementation, topical keratolytic. As the patients were not responding to medical treatment, tangential excision of keratoderma layer was done up to subcutaneous fat using dermatome followed by resurfacing with intermediate thickness split thickness skin graft. The donor site healed normally. Patient was followed up for 6 months; there after they lost to follow up. Histopathology reveals epidermal hyperplasia, ortho hyperkeratosis, focal parakeratosis and hypergranulosis.

Discussion

The disease was first described by H.C Olmsted in 1927 in a 5year old boy developing bilateral palmoplantar keratoderma and periorificial hyperkeratosis during initial year of his life [1]. Mostly the reported cases are sporadic and very few have family history [2], but our patients were siblings. As per the literature, males are predominantly affected [2] but in our case all 3 patients were females. Till date 73 cases have been reported [3]. The etiopathogenesis of the disease as described in literature is likely due to gain of function mutation of TRPV3 on chromosomal region 17p13, causing apoptosis of keratinocytes and consequent hyperkeratosis of skin in the diseased individuals [4]. Another study showed defective expression of mature epidermal keratin type 1 and 2 and persistence of acidic keratin [5].

The diagnosis of disease is mainly clinical and is made when the following criteria met- "symmetrical involvement of the palms and soles with keratoderma, symmetrical hyperkeratotic plaques in the periorificial areas" [6]. The progression of disease is slow but debilitating. As the keratoderma extends the patient has difficulty in walking. Fissuring around toes leading to auto amputation has been reported and it was seen with one of our case as well. The differential diagnosis to be considered in this case are- Acrodermatitis enteropathica, pustular dermatitis around mouth/anus, chronic diarrhoea, Vohwinkel keratoderma and these were ruled out. At present there is no satisfactory treatment for the disease. Topical therapies with keratolytics have been tried with varying success. Debulking surgery is also options with coverage of raw area using skin grafting have been tried in many cases but there are recurrences [7]. Recently sirolimus which is mTOR inhibitor and erlotinib which is EGFR inhibitor has shown promising result in the management of this disease [8]. The researchers have stated that "all 4 patients (median age of 7) had sustained improvements in pruritus and pain with transient hair loss and acneiform eruptions that responded well to topical medications, without any adverse effects" [8]. With erlotinib, after 3 months therapy "all three patients had resolution of hyperkeratosis and pain, allowing them to wear shoes and walk," the researchers said. Anorexia, insomnia resolved, and growth improved [8] and the benefits persisted for 12 months of treatment.

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Conclusion

The disease is rare with very few cases from India. There is currently no satisfactory treatment for the disease. For surgical treatment currently debulking and skin grafting is being done and long term follow up is required. The anti neoplastic drugs have shown promising results but risk benefit ratio must be taken into account before starting the drug. As for our patients the targeted therapy was not considered in view of financial constraints, surgery was done and patients were prescribed silicon cushions for foot wear to avoid pressure and frictional damage. After 6 months postoperation, there is satisfactory healing and no recurrence as of now.

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