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Olmsted Sydrome Accompanıed by Immune Deficiency

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Abstract: Olmsted Syndrome, OS, (congenital palmoplanter and periofical karetoderma) is a very rare condition and is associated with palmoplanter karetoderma and perioral hyperkaratotic plaques, deformation and loss of the flexibility of hands. On rare occasions it could lead to amputation of the severely affected digits or hands. The condition has also been associated with growth impairement and joint laxity and described to be inherited from the famility or effect patient's sporadicaly. Specific types of skin lesions are formed with OS. New types of leasions and plaques are still being identified with new patients. In our sunjest we have seen depletion of IgG2 levels (immune deficiency) severe pneumonia and empyema during skin deformation. Although all reported cases have been described to have genetic links or appear sporadicly, our findings show that the depletion of the immune responses should also be considered and not ignored when identifying and diagnosing the condition.
Keywords: Olmsted syndrome, Child, immune deficiency

INTRODUCTION

Olmsted syndrome (OS) still remains as a rarely reported condition. OS (congenital palmoplantar and perioral keratoderma) is a very rare disorder and was described by H. C. Olmsted in 1927 [1] whose original description included the combination of bilateral, mutilating, palmoplantar keratoderma and periorificial hyperkeratotic plaques with flexion deformities of the digits, leading to constriction or spontaneous amputation [1, 2]. Bilateral palmoplantar keratoderma and periorificial hyperkeratotic plaques are the hallmarks for diagnosis of this syndrome [1, 3]. Many features have been subsequently associated with this syndrome and new features continue to be reported. Generally these new features includ cuteneaus finding [4]. Immundeficiency and pnomenue, ampiame havent been described but only one has been describedso far. We report here the case of a six year-old male child, who had ampiame and immun deficiency with OS

CASE REPORT

Our subjest is a 6 year old male child, who had suffered with a high temperature and a cough for a week before a long antibiotic treatment and refered to our intensive care unit where we have diagnosed him with pneumonia after a PA scan. His parents were healthy and his family had no mediacel history of any skin condition or disease.

During the medical inspection, we have found that the subject had breathing problems on the right lung auscultation and seen hyperketatotic plaques arouns palmo-planter reagions and fingers of both hands (figure 1), soles of the feet, edges of the mouth and genital regions. We have also seen fragile hair strands which were easily broken and hair loss in pathches. The PA scan also confirmed pneumonia on the right lung (CRP : >300). After the diagnosis a combined Sefotaksim (Eqıtax) + Amikasin (Amiketem) + Vankomisin (Vancomycin HCL) treatment was given to our subject and his parents were informed and advised on the condition and the treatment. A urea (15%) solution was used for lesion and plaque treatemnt on skin in addition of the drug coctail treatemnt given. As the treatment described above was found to be unsuccessful we have decided to check the immune responses of the subject and found his IgG2 levels to be depleted to 65 mg/dl (75-350 mg/dl) . The subject did not seem to have leucopenia and the PA scans have confirmed empyema (figure 2). After the diagnosis of the empyema the subject was fitted with a thorax tube and further PA thorax scans and tomografies were performed to show pleural thickening (figure 3) followed by camera aided thoracoscopic abscess drainage and reformation of the scared regions of the affected areas which was finally found to be effective. The subject was discharged however monitored regularly after the treatment described above.

DISCUSSION

Olmsted described a five year-old male who developed sharply marginated, palmoplantar keratoderma during his first year of life. The thick keratoderma led to flexion deformities and autoamputation of the digits. Periorificial, harply marginated, hyperkeratotic plaques were also present [1]. Reports of new cases have lengthened the list of cutaneous and systemic features that may be seen in this

syndrome. However, it is widely accepted that two major findings that are prerequisites for diagnosis of this syndrome are the symmetrical involvement of the palms and soles with keratoderma, and symmetrical hyperkeratotic plaques in the periorificial areas [1, 4].

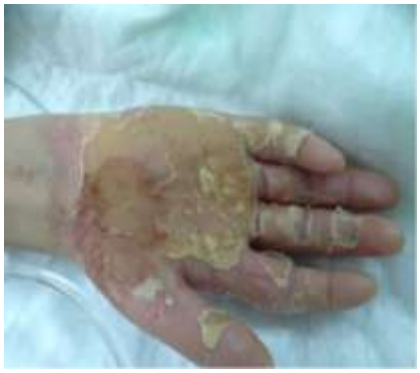

Fig. 1: Hiperkeratotic plak of palm

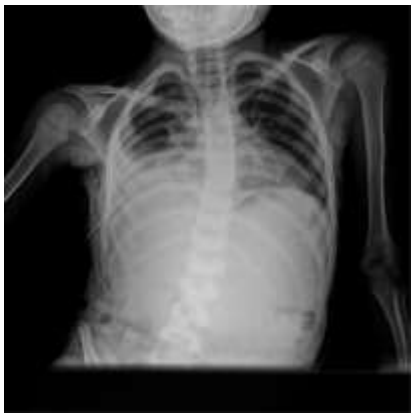

Fig. 2: PA scan showing pneumonic infiltration

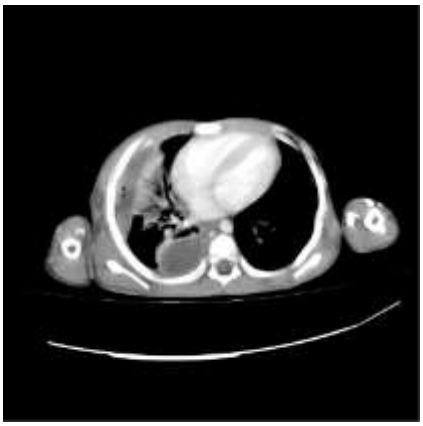

Fig. 3: Right lung empyema

Although Herein has demostrated autosomal dominant inheritance [5] of the condition in 3 patients, the incidences have been described mainly as sporadic. Cambiachi et al, have shown the condition to be X chromosome linked and dominantly inherited in twin studies. The pathogenesis of the condition has not been fully understood however the TRPV3 gene in OS patients was found to be mutated by Lin et al. As this gene is involved in expression of the skin, the brain, the hair follicules and the spinal cord, it was found to be playing a major role in growth and formation of the healthy skin and hair. Therefore loss of full of partial activity of this gene is therefore associated with apoptosis of the skin cells and formation of skin hyperkeratosis [7].

Kress *et al.* have identified defects in epidermal keratin formation [8]. The effects of the OS are usually identified in todlers however the plamoplanters are usually formed durin the first 6 month of their life followed by yellow-brown periorificial plaques which are usually caracterised as skin thickening around the axilla and the neck regions.

Keratoderma is initally thick, hard and erythematous. At later stages the keratoderma coveres the dorsal regions of the hands and becomes itchy and painful resulting in loss of flexibility of hands. In some patients the lesions can lead to epidermal tomours and cancers. Other findings include hair loss, white plaques in the mouth, chronic paranosia and hyperhydrosis in palms and soles [3, 6, 9].

There is no satisfactory treatment for this condition. Topical treatments offer only symptomatic relief of pain and fissures by reducing the thickness of the keratotic palmoplantar skin lesions. Various topical agents like salicylic acid, urea, boric acid, corticosteroids, shale oil, other emollients, retinoic acid, antimicrobials, wet dressings and prolonged soaking of the affected parts in water, have been tried with varying success. Systemic treatment with antihistamines, vitamins E and A, antimicrobials, corticosteroids have also been used anecdotally with no consistent benefits [1, 2, 4].

Acrodermatitis enteropathica, congenital paronisia, papillon lefever syndrome and hereditary keratoderma mutilans, ectodermal dysplasia should be considered when diagnosing this condition. Unfortunately there are not many treatment options for these patients however local application of salisic asit, boric asit, ürea, retinoic asit, steroid and warm water have been found to be effective. A skin graft could also be considered for stabborn lesions. All these treatments have been found to be improve the flexibilty of the hands howeve as they do not completley cure the disease does not stop repalse of the condition [3, 10, 11].

There are 4 subclasses of IgG (classified according to position and number od the disuphide bonds). The Total IgG is composed of 60-70% of IgG_1, 14-25% of IgG_2, 4-10% of IgG_3 and 2-6% of IgG_4.

Depletion of IgG is one of the most common types of the humoral deficiencies. Repetative severe or chronic upper respiratory conditions, bronchial infections, sinusites, Otitis Media and atopic or nanotopic obstructive respiratory disease all have been associated with depletion of IgG subclasses [12].

In our subject we have identified plaques around the mouth with defined edges, plamoplanter hyperkerotoctic plaques, hair loss and damage, genital region plaques and a severe lung infection which was intended to be intended to be treated with coctail of antibiotics. As the treatemnt was not found to be effective, the IgG levels of the subjest was evaluated and the IgG2 level was found to be depleted. As this was not defined in litearture previoully we think it should be considered and not ignored when diagnosing and identifiying the condition.

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