Hypotrichosis in a Child with Olmsted Syndrome

Sir,

Olmsted syndrome is a rare and unique keratinizing disorder which presents with bilateral mutilating transgradient palmoplantar keratoderma and periorificial keratotic plaques. Other reported features include leukokeratosis of the tongue, icthyotic lesions, pain, itching, absent premolar teeth, hearing loss for high frequencies, sclerosing cholangitis, short stature, and laxity of the large joints, linear hyperkeratotic follicular streaks, and acral hyperhidrosis.[1,2] Hypotrichosis has rarely been reported in Olmsted syndrome.

A 5-year-old male child presented with periorificial keratotic plaques associated with painful fissures and thickening of bilateral palms and soles [Figure 1]. He had flexion contracture in both hands for the past 2 years. The patient was the only child of a second-degree consanguineous marriage. There was no history of similar complaints in the family. On general examination, there was pallor and gradeIII IAP (Indian Association of Pediatrics) protein energy malnutrition. On examination, the palms and soles showed keratoderma with flexion contracture of bilateral fingers [Figure 2]. The child was unable to walk because of the associated pain. There were hyperkeratotic plaques with fissuring around the perioral region, intranasal, external auditory canal, and in the intergluteal region [Figure 3]. The intranasal plaquescaused difficulty in breathing. Scalp examination showed hypotrichosis with sparse, short, and light-colored hair [Figure 4]. Light microscopic examination of hair shaft showed reduced pigmentation, reduced hair shaft diameter, and trichoschisis. Similar findings along with folliculocentric papules and empty follicles were seen in trichoscopy [Figure 5]. Ophthalmic examination showed

decreased eyelashes in both eyes and corneal epithelial defect in the left eye [Figure 6]. Skin biopsy from the palm revealed parakeratosis, irregular acanthosis, and papillary dermis showed vascular proliferation with edema. He was started on oral acitretin 1 mg/kg, emollients, passive extension exercises for hands, and nutritional supplements. Four weeks later, there was mild reduction in plaque thickness along with reduced pain on walking and the child was able to resume walking.

Olmsted syndrome was first described by Olmsted in 1927 in a 5-year-old boy. It is a rare genodermatosis and the exact prevalence is unknown. Only 73 cases were reported till 2014.

Olmsted syndrome may be sporadic or familial with autosomal dominant, recessive or X-linked inheritance. Mutations in *TRPV3*(Transientreceptorpotentialvanilloid-3) gene is associated with sporadic, autosomal dominant, and recessive Olmsted syndrome, and mutation in *MBTPS2* (membrane-boundtranscriptionfactor2) is associated with recessive X-linked Olmsted syndrome.[1]

There is a paucity of reports of hair abnormalities in Olmsted syndrome. Our patient had hypotrichosis, scanty eyebrows and eyelashes, reduced hair shaft diameter, and trichoschisis. Dogra *et al*.[3] described hypotrichosis with hair shaft defects in a 5-year-old child. Poulin *et al*.[4] described a case of Olmsted syndrome who presented with congenital universal alopecia along with absence of premolar teeth and leukokeratosis of oral cavity. Other reported changes include sparse, brittle, lustreless hair, woolly hair, congenital absence of eyebrows and eyelashes along with hypotrichosis of the scalp, which eventually progressed to alopecia totalis, pilitorti, piliannulati, pseudomonolethrix, and reduced number of hair follicles with minimal scarring.[5-8] *TRPV3* plays an important role in skin keratinization, hair growth, and itching sensation in humans, and *TRPV3* mutation can elevate intracellular calcium concentrations, induce apoptosis, and inhibit hair growth.[9] Thus, our patient could have had *TRPV3* mutation resulting in hypotrichosis. However, genetic testing could not be done since the patient could not afford the investigation.Congenital alopecia with atrichia or hypotrichosis with palmoplantar keratodermas can be seen in other syndromes including Clouston syndrome, HOPP (hypotrichosis, acrosteolysis, onychogryphosis, periodontitis, and palmoplantar keratoderma) syndrome, keratosis follicularis spinulosa decalvans, odonto-onycho-dermal dysplasia, Lelis syndrome, Schopf–Schulz–Passarge syndrome, KID (keratitis-ichthyosis-deafness) syndrome, palmoplantar keratoderma-congenital alopecia syndrome, andVohwinkel syndrome.[10-12] Woolly hair with palmoplantar keratoderma can also occur in ectodermal dysplasia with skin fragility, Carvajal syndrome, and Naxos syndrome.[10] No beaded appearance was seen in light microscopy or trichoscopy.

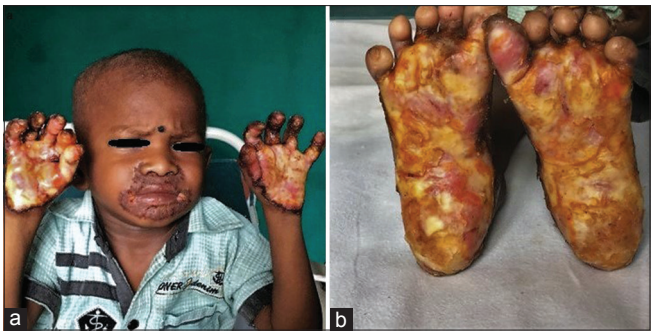

Figure 1: (a) Perioral hyperkeratosis with hyperkeratosis and fissuring of palms; (b) hyperkeratosis and fissuring of soles

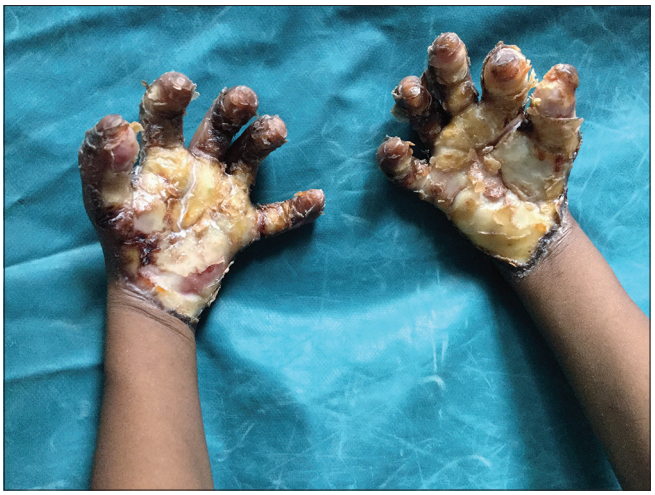

Figure 2: Flexion contracture of bilateral fingers

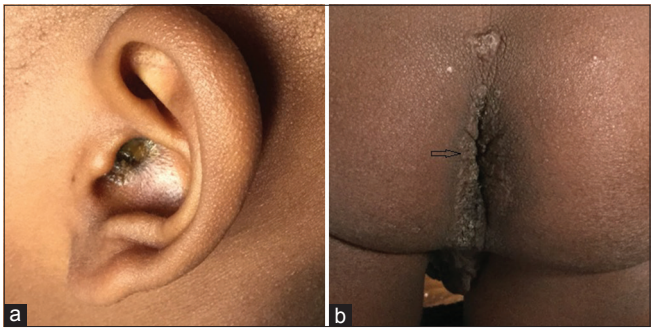

Figure 3: (a) Hyperkeratotic plaques in the external auditory canal; (b) hyperkeratotic plaques in the intergluteal cleft

There is no definite treatment available for Olmsted syndrome. Cutaneous lesions are recalcitrant to therapy.[1,2] Secondary bacterial infections and candidiasis may occur in macerated plaques. Malignancies such as squamous cell carcinoma and malignant melanoma may occur on keratoderma plaques.[7] Various systemic medications such as systemic retinoids, corticosteroids, and methotrexate have been tried with minimal or no response.[1,7] Topical medications such as calcipotriol, emollients, and keratolytics have also been tried.[5] Surgical treatment modalities that have been tried are full thickness excision of keratoderma followed by split skin graft.[2,13]

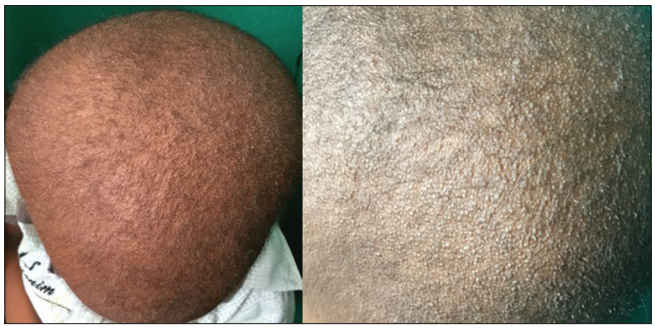

Figure 4: Hypotrichosis with sparse, short, and light-colored hair

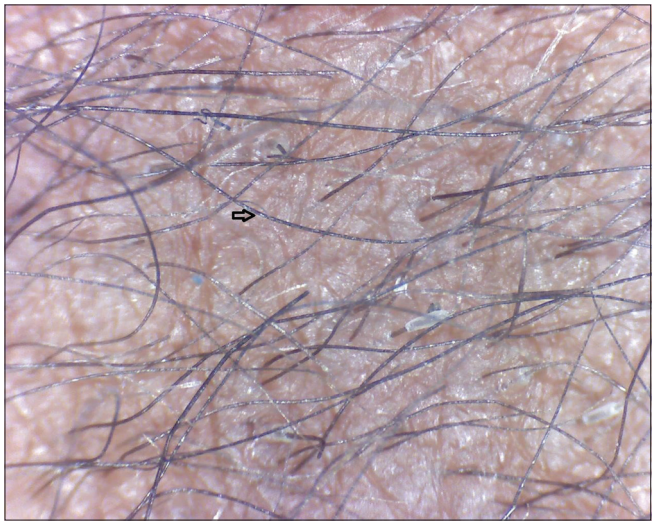

Figure 5: Trichoscopy revealed short sparsehair×25 (nonpolarized light)

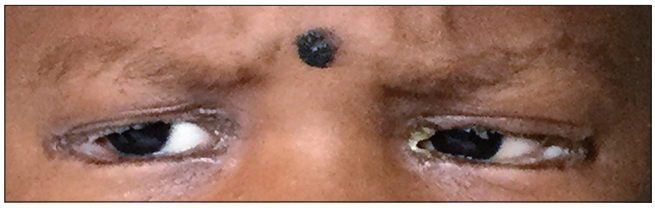

Figure 6: Decreased eyelashes in bilateral eyes

Conclusion

Our case is a rare case of Olmsted syndrome with hypotrichosis. Only a few case reports of Olmsted syndrome have been reported from India. Further elucidation of the variable clinical phenotypes and genetics of this rare syndrome is needed.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patients mother has given consent for her sons images and other clinical information to be reported in the journal. The patients mother understands that name and initial will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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