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. 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 grade III 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 plaques caused 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 (Transient receptor potential vanilloid-3) gene is associated with sporadic, autosomal dominant, and recessive Olmsted syndrome, and mutation in MBTPS2 (membrane-bound transcription factor 2) 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, pili torti, pili annulati, 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, and Voelwinkel syndrome.[10-12] Woolly hair with palmoplantar keratoderma can also occur in ectodermal dysplasia with skin fragility, Carvalajal syndrome, and Naxos syndrome.[10] No beaded appearance was seen in light microscopy or trichoscopy.

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]
Letters to the Editor

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Conflicts of interest
There are no conflicts of interest.

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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 son’s 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.

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

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

Figure 6: Decreased eyelashes in bilateral eyes
Sir,

Perioral dermatitis is an uncommon facial eruption of women and children presenting as tiny papules and pustules often in a perioral distribution. It is now more correctly referred to as "periorificial dermatitis" as it frequently involves perinasal and periocular skin. Granulomatous periorificial dermatitis (GPD) is a distinct facial eruption in prepubertal children characterized by a monomorphic papular eruption occurring in the perinasal, perioral, and periocular areas.

A 30-year-old unmarried female, saleswoman by occupation, presented with appearance of reddish, scaly lesions on the face since 2 months. There was history of mild photosensitivity associated with the lesions. She did not give a history of introduction of new creams or cosmetic products, drug ingestion, or parlour procedures prior to appearance of lesions. Patient received treatment with oral doxycycline and topical antibacterial creams for 1 month without significant improvement. She had also received topical steroids for 7 days in the recent past with worsening of lesions requiring discontinuation.

Cutaneous examination revealed multiple erythematous, scaly papules and plaques on the supra orbital, periorbital, perioral, and perinasal area with classical sparing of upper forehead and butterfly area of the face [Figure 1a and b]. A provisional diagnosis of periorificial dermatitis was considered.

Laboratory investigations of the patient did not reveal any abnormality. Skin biopsy from the inflammatory papule revealed perifollicular and perivascular granulomatous inflammatory infiltrate composed of lymphocytes, epithelioid cells, and giant cells suggesting the diagnosis of granulomatous type of periorificial dermatitis [Figure 2a and b].

Considering unresponsiveness to doxycycline in the past, patient was started on oral isotretinoin 20 mg daily with topical application of metronidazole cream. The patient followed up after 5 days with acute exacerbation of lesions [Figure 3a and b]. She was counselled and advised to continue the treatment. After 3 weeks of treatment with isotretinoin, there was remarkable improvement in the lesions with disappearance of papules and significant reduction in the erythema [Figure 4a-c].

Periorificial dermatitis (POD) is an acneiform eruption of unknown origin, most commonly found around the orifices. POD occurs worldwide, especially in the fair-skinned population; predominantly women between the ages of 15 and 45 years are affected. Peak incidence is in the second and third decade of life. POD is also observed in children, where in contrast to the adult form, males are predominantly affected.

The exact etiology is unknown; however, it is related to the impairment of barrier function and dryness of the skin as well as proliferation of the skin flora. It may be induced by topical application or inhalation of corticosteroids, allergic response to amalgam, and mercury in dental fillings, toothpaste containing fluorides, cosmetics, fusiform bacteria, Candida albicans, and Demodex folliculorum. Nikkels and Pierard noted POD in females after stopping oral contraceptives. These patients also had premenstrual flares.

It usually presents as grouped follicular reddish papules, papulovesicles, and papulopustules on an erythematous base with a possible prospect of confluence of lesions, predominantly in perioral distribution with classical sparing of vermilion border of lips. Granulomatous variant of periorificial dermatitis (GPD) is a well-recognized entity, presenting most commonly in prepubertal children as monomorphic small papules limited to the perioral, perinasal, and periocular regions. The primary lesion is a discrete 1–3 mm dome‑shaped flesh‑colored, yellow‑brown, or red papule. Mild scaling of the lesions or surrounding erythema may occur. Extra facial