Palmoplantar Keratoderma with Keratoconus
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Abstract:
Palmoplantar keratodermas (PPKs) are a rare heterogeneous group of disorders characterized by abnormal thickening of the skin of palms and soles. Ocular manifestations reported with Palmer planter keratosis include scleral melanosis, macular deposits, and congenital cataract. We report, for the first time, a case of bilateral keratoconus in a patient with PPK.

Keywords:
Keratoconus, palmoplantar keratoderma, skeletal abnormalities

Introduction
Palmoplantar keratodermas (PPKs) comprise of a rare heterogeneous group of genetic and acquired keratinization disorders characterized by hyperkeratotic thickening, painful fissures and callosities of the skin of palms and soles.[1] Most PPKs are genetic defects having autosomal dominant, recessive, and X-chromosomal patterns of transmission.[1] Less commonly acquired form occur later in life where other associations such as drug intake, internal malignancy, acquired dermatosis, metabolic disorders, malnutrition, chemicals, infectious, need to be ruled out.[2] Based on the clinical morphology, PPK may be classified as diffuse, focal or punctate types. Various clinical features of hereditary PPK include initial manifestation of disease in childhood, a positive family history, persistent clinical appearance with little variation in the type and severity of symptoms, and relative treatment resistance.[3] Ocular manifestations previously reported with PPKs include scleral melanosis, macular deposits, and congenital cataract.[4‑6] However, to the best of our literature search, no association of keratoconus has been described with PPKs. Therefore, we herein, for the first time, report a case of bilateral keratoconus in a patient with PPK.

Case Report
An 18-year-boy presented with complaints of gradual, progressive, diminution of vision in both eyes (left eye > right eye) for 3 years. He had no other ocular complaints or history of spectacle use. He was simultaneously being treated in the dermatology department for painful fissuring and ulceration of the skin of palms and soles since birth. It started initially from the right sole just proximal to the toes and progressed to involve the other sole and the palms over the next year. The thickening was accentuated on pressure bearing sites. The lesions were associated with deformity of the hand interfering with daily activities of the patient.

Patient’s younger brother also suffered from similar but milder skin lesions. He had no ocular complaints. No other family member seemed to be affected. The past and personal history were unremarkable.

General physical examination revealed severe stunting and wasting with 19 kg weight, 130 cm height, and a body mass index of 11.2. Secondary sexual characters were underdeveloped as evidenced by decreased
Cutaneous examination revealed diffuse mutilating yellow-colored thickening of the palms and soles with background erythema, erythematous borders, and hemorrhagic crusting [Figure 1]. Flexion deformity of all interphalangeal joints of palms and resorption of distal phalanx was noted [Figure 1]. There was erythematous atrophic skin over the dorsum of bilateral digits of hands and feet up to metacarpophalangeal joints in hands and whole dorsum of the foot with sclerodactyly [Figure 1]. Fifty percent hot and cold loss was seen over hands and 100% over palms.

Skeletal examination showed the presence of prognathism, scoliosis, pectus carinatum, increased arm span (128 cm), upper segment: lower segment ratio of 0.83, hallux valgus, and increased femoral anteversion (W-sign) [Figure 1]. However, no joint laxity was seen. Radiological examination revealed skeletal age lagging behind chronological age.

Pulmonary function tests showed moderate restriction on spirometry, mild restriction in lung volume, and a normal diffusion capacity. The electrocardiogram showed sinus tachycardia and a normal ultrasound abdomen. Ear and dental examination were within normal limits.

The best corrected visual acuity was 6/36 in the right eye and 1/60 in the left eye. Both eyes had corneal ectasia as evidenced by the presence of Munson’s sign, Rizzuti’s sign [Figure 2], and scissoring reflex on retinoscopy. Slit lamp examination showed corneal thinning and ectasia in both eyes (left eye > right eye) and corneal opacity at the level of the Descemet’s membrane in the superior half of the cornea. The diagnosis of early keratoconus in RE and advanced keratoconus in LE was confirmed by corneal topography with Orbscan [Figure 2]. Corneal thickness was 484 µm OD and 372 µm OS at the thinnest points. Keratometry values in right eye showed maximum power of 53.3 D at 175° and minimum power of 46.2 D at 85°, while in the left eye, maximum power was 62.1 D at 66° and minimum power was 56.6 D at 156° [Figure 2]. Rest of the ocular examination was within normal limits.

Discussion

Ocular involvement in PPKs is extremely rare. Ocular associations have been reported in “Mal de Meleda syndrome” (MdM) which is an extremely rare autosomal recessive congenital form of palmoplantar hyperkeratosis with extended involvement of the dorsa of the hands and feet (transgrediens behavior) in a “glove-and‑stocking” distribution. PPK did not involve dorsa of the hands and feet in our patient. Our patient had diffuse type of palmoplantar keratosis since birth. Ocular associations of MdM have been described by Durmuş et al., in a case presenting with ocular melanosis and bilateral macular deposits.[4] There are only two reports of congenital posterior subcapsular cataract in association with Mal de Meleda including a consanguineous Tunisian family with two female siblings aged 45 and 30 years, who presented with a clinical association of mal de Meleda and bilateral congenital posterior subcapsular cataracts.[5,6]

However, no association of keratoconus has been described with PPKs. Keratoconus is a bilateral asymmetric noninflammatory corneal ectasia characterized by progressive stromal thinning and corneal protrusion resulting in irregular myopic astigmatism.[7] The isolated sporadic disorder is the most common presentation. It may be associated with various ocular and systemic conditions such as vernal and atopic keratoconjunctivitis,[7] retinitis pigmentosa, Laurence–Moon–Biedl syndrome, aniridia, Leber’s congenital amaurosis, ectopia lentis, floppy eyelid syndrome, Down’s syndrome, Ehlers–Danlos syndrome, Marfan’s syndrome, atopic dermatitis, mitral valve prolapse, and neurofibromatosis. Significant risk factors include history of atopy, contact lens wear, and constant eye rubbing.[7]

The patient presented to us with palmoplantar keratosis with keratoconus and many skeletal and joint abnormalities which have not been mentioned in literature. We believe that their co-occurrence could be more than coincidental finding as the origin of corneal epithelium and epidermis of skin is from the ectoderm, while that of keratocytes, corneal endothelium, melanocytes and dermis in the skin, craniofacial bones and tendons from neural crest cells.[8,9] Thus, we postulate that the co-occurrence of PPK, keratoconus, and skeletal abnormalities in this patient could be attributed to the
origin of these structures from the same embryological layer, i.e., ectoderm and neural crest.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

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