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Case Report

Oral manifestation of pachyonychia congenita type 1: Jadassohn lewandowsky syndrome

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ABSTRACT

Pachyonychia congenital (PC), is a rare genetic disorder, autosomal dominant, disorder of keratinization. This condition is characterized by cutaneous manifestation mainly hyperkeratosis of skin and mucosae and hypertrophy of nails. In this condition, almost 50% of the patients will have oral leukokeratosis. The case report here is of a 15 years old girl, presented with dystrophic, thickened fingernails and toenails with subungual hyperkeratosis, palmoplantar keratoderma, hyperkeratotic plaques in buccal mucosae. Histological examination shows acanthosis, parakeratosis and ballooning of epithelial cells, these features were consistent leukokeratosis, and has been diagnosed as Pachyonychia Congenita type 1. This is a rare condition hence, has been reported.

1. Introduction

Pachyonychia Congenita is a rare disorder of keratinization,[1] inherited as an autosomal dominant trait. In 1904,[2] the first documentation was done by Muller and the first description of the disease was by Jadassohn and Lewandowsky in 1906.[3] This disease is classified into four types, in which important once are Type 1: Jadassohn Lewandowsky type and Type 2: Jackson Lawer type.[4] In type 1, hyperkeratosis of palm, soles, knees and elbows is usually found with leukokeratosis and follicular keratosis involving mouth, anus, upper airway and may involve larynx in few cases.

In type 2, the keratoderma and mucosal changes are absent or less found. The common finding in type 2 is neonatal teeth and multiple epidermoid cyst.

In type 3 (Schafer-Brunaver), in this type, the features are similar to type 1, but the most common finding in this is corneal leukokeratosis.

The 4^{th} type is called as PC Tarda or late onset PC, because the clinical features appear during the second or third decade of life and has features similar to type 1 PC.[5]

This is a case report of a 15 years female patient with type 1 PC with oral manifestation.

2. Case Report

A 15 years old girl of with normal developmental milestones according to her age, born to non consanguineous parentage, presented with oral lesion in the last 10 years, which turned out to be painful in the last 3 months and also complained of hoarseness of voice in the last 3 months. Further, she also gives history of thickened fingernails and toenails associated with occasional pain, she noticed all these changes when she was 5 years old and at the age of 7 years, she was bought to the notice of dermatologist. There was no similar complains or history in her family.

On oral examination, she had white hyperkeratotic plaques in buccal mucosa [Figure 1] and also involving both lateral borders of her tongue which was multiple

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and painful. Leukokeratosis of dorsum of tongue was present, which was not possible to scrape off the lesion and was sensitive to touch. On video direct laryngoscopy, leukokeratosis of laryngeal mucosa and edematous vocal cords was seen [Figure 2]. On physical examination, discolored, dystrophic and thickened fingernails and toenails with subungual hyperkeratosis was present [Figure 3]. Marked hyperhidrosis of palms and soles was present. Palmoplantar keratoderma with painful hyperkeratotic plaques was noted. Numerous pin head follicular papules were present on entire body.

Fig. 1: Dystrophic and thickened fingernails with small white follicles over the mucosa

A biopsy of white hyperkeratotic plaque on right buccal mucosa and right lateral border of tongue was done. Histopathological examination shows acanthosis, parakeratosis and ballooning of epithelial cells when stained with hematoxylin and eosin. A biopsy of leukokeratosis of dorsum of tongue was also done which shows epithelial hyperplasia with no dysplastic cells. KOH microscopy and culture of nail clipping was negative. Routine laboratory investigations like complete hemogram, lipid profile, LFT and RFT were normal. No evidence of malignancy was found during the thorough work up.

Molecular biological and genetic studies were not done due to lack of infrastructure facilities. Based on the findings, she was diagnosed as Pachyonychia Congenita type1.

The patient is currently being treated on outpatient basis. She is been prescribed with vitamin A and E with emollients, mouth was containing corticosteroids (0.01% dexamethasone clixir) and for hyperkeratosis of palms and soles keratolytic that is topical 10% urea and salicylated

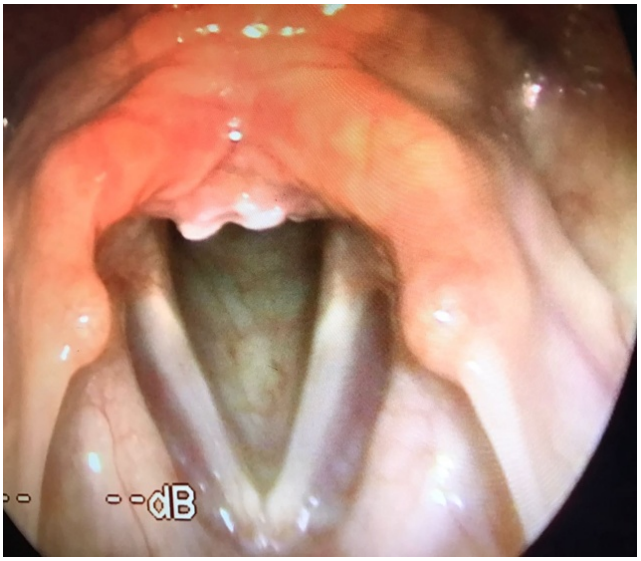

Fig. 2: Laryngeal mucosa showing hyperkeratosis edematous vocal cords

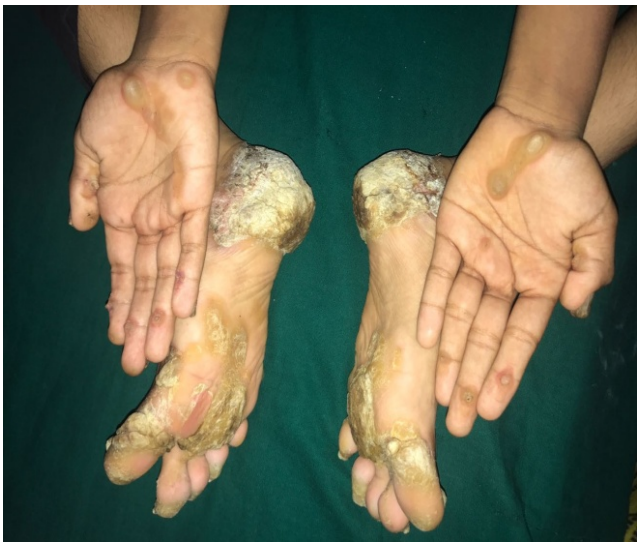

Fig. 3: Palmoplantar keratoderma with hyperkeratotic plaques

lignocaine jelly on lesions to reduce pain was prescribed. Based on above treatment, there was improvement of local sensitivity. Her cutaneous and mucosal lesions are being clinically monitored twice a year and biopsies are performed, if required. No malignancy was evident till date in the patient.

3. Discussion

Pachyonychia congenital (PC) is a rare genetic disease transmitted as an autosomal dominant trait. It has high degree of penetration. There are few reports of AR inheritance.[6] Sporadic case has also been reported from

spontaneous mutation. An estimated 5,000 to 10,000 cases have been reported worldwide.[4] PC has been classified into 4 types based on clinical features, the common clinical features in these types are palmoplantar keratoderma, characteristic hypertrophic toenail dystrophy, oral leukokeratosis, palmoplantar hyperhidrosis, variety of epidermal cysts. The nail, skin, oral mucosa, larynx, hair and teeth are involved in variable combination of which nail involvement is prominent. Few cases only nail involvement has been reported.

Patients with type 1: Jadassohn Lewandowsky syndrome, have symmetric hyperkeratosis of palms and soles over the pressure sites. Follicular keratotic papules on knees and elbows, some cases throughout the body.[7] Oral leukokeratosis, palmoplantar hyperhidrosis, sometimes painful blisters may also develop over the palms and soles. Rarely, verrucous lesion may be present over elbows and knees. Hoarseness of voice with laryngeal mucosa involvement is an important feature of PC type 1, in addition it may rarely involve nasal, esophageal or tympanal mucosa.[8,9]

Type 2: PC Murray Jackson Lawler syndrome, presents with mild palmoplantar keratoderma, natal teeth, hair anomalies like pili torti, bushy eyebrows, multiple pilosebaceous cyst, mild oral leukokeratosis, steatocystoma multiplex, in this type, development of epidermal cyst or steatocyst are hallmark findings.

Type 3: Schafer Branauer syndrome, has features of type 1 along with corneal dyskeratosis.

Type 4: PC Tarda, it is manifested in second or third decade of life.[10]

Pathogenic mutation in keratins CK6a or CK16 are associated with PC type 1 and CK6b and CK17 are associated with PC type 2. Because of these keratin mutation leads to skin fragility and hyperkeratotic disorders.[4] The CK6 and CK16 cytokeratins are responsible for hyperproliferation which are normally found in hairy areas, epidermis, non-keratinized stratified epithelium. If any stimulus like injury or mutations occur in the keratocytes that expresses CK6 or CK16, they migrate to the affected site (injured site), these cytokeratins play a role in allowing keratinocytes to have pliability and plasticity to migrate and leads to re-epithelization.[11] At molecular level, mutation in keratin K16 and K6a has been demonstrated in PC type1. Although these cytokeratins are found in squamous cell carcinoma, there is no confirmation of this relationship between pachyoychia and malignant neoplasm.[11]

The cardinal features of PC (seen in >90% patients) are thickened toenails, plantar keratoderma and plantar pain.[4] Sometimes PC may be associated with unusual features. An association between PC and median rhomboid glossitis reported by Karen and Schaffer,[12] PC may be associated with unusual dental finding or B cell lymphoma[13] have also been reported.

In this case, the clinical features suggestive of PC type 1with characteristic feature subungual hyperkeratosis with follicular papules over entire body since the age of 5. Histopathological examination shows changes as acanthosis, parakeratosis and ballooning of epithelial cells, similar to other case report, confirms the diagnosis.[14]

Till date there is no treatment for PC, thus treated symptomatically.[15] The patient is currently being treated on outpatient basis. The patient is prescribed with vitamin A and E along with emollients and keratolytic.[16] Vitamin A stimulates differentiation, which normalize the accelerated epidermopoiesis of pathological keratinocytes of epidermis of skin and nails. Topical corticosteroids were used leading to regression of pain in oral lesion that is mouth wash containing corticosteroids (0.01% dexamethasone elixir) and for hyperkeratosis of palms and soles, keratolytic like topical 10% urea and salicylated lignocaine jelly to reduce pain. The patient was advised limitation of walking and standing, use of soft shoes, control of body weight and use of appropriate clothing which has improved the local sensitivity.

Periodic oral examination is important and the lesions are being clinically monitored twice per year and annual biopsies are performed if required. As there is a lack of studies for the treatment of oral lesion in PC, henceforth studies must be undertaken in order to improve the quality of life of these patients.

4. Summary

Pachyonychia congenital (PC) is a rare genetic disease transmitted as an autosomal dominant trait, disorder of keratinization. This condition is characterized by cutaneous manifestation mainly hyperkeratosis of skin and mucosae and hypertrophy of nails. This is a case report of a 15 years female patient with type 1 PC with oral manifestation. She was diagnoses based on detailed physical and ENT examination and histopathological examination. Lesions are being clinically monitored periodically. As there is a lack of studies for the treatment of oral lesion in PC, henceforth studies must be undertaken in order to improve the quality of life of these patients.

5. Source of Funding

None.

6. Conflict of Interest

None.

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