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Case Report

Oral manifestations of lamellar ichthyosis: A rare case report and review

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Abstract Ichthyosis is a heterogeneous family of hereditary disorders mostly characterized by the accumulation of hyperkeratotic scales. Although these disorders affect tissues of epidermal origin, there is little evidence regarding the involvement of oral and dental structures in ichthyosis. Perioral tissues, however, are frequently affected both by disease and drugs used for treatment. A case report of dental involvement in lamellar ichthyosis is presented and the dental management is discussed.

Key words
Ichthyosis, teeth abnormalities, caries, dental management.

Introduction

Ichthyosis is a heterogeneous family of hereditary disorders mostly characterized by the accumulation of hyperkeratotic scales on the skin surface. It is caused by abnormality in keratinization and exfoliation of the horny cell layer.[1] The severity of symptoms can vary enormously, from the mildest types such as ichthyosis vulgaris which may be mistaken for normal dry skin up to life-threatening conditions such as harlequin ichthyosis. The most common type of ichthyosis is ichthyosis vulgaris, accounting for more than 95% of cases.[2,3]

Lamellar ichthyosis (LI) is relatively rare type of ichthyosis with an incidence of approximately 1:200,000 to 300,000 live births. The synonyms include nonbullous congenital ichthyosiform erythroderma, congenital ichthyosiform erythroderma, non-erythrodermic autosomal recessive lamellar ichthyosis, ichthyosis congenita, and classic LI. The onset of LI is at birth.[4] Infants with LI are often born preterm as "collodion baby".[5] Later in life, the skin is covered with large, thick, dark scales that have a plate-like appearance. There can be hair and nail involvement along with a number of deformities. This involves a mutation in the gene for transglutaminase 1 (TGM1). The transglutaminase 1 enzyme is involved in the formation of the cornified cell envelope. The formation of the cornified cell envelope is an essential scaffold upon which normal intercellular lipid layer formation in the stratum corneum occurs. Thus, mutations in the TGM1 secondarily cause defects in the intercellular lipid layers in the stratum corneum, leading to defective barrier function of the stratum corneum.[6] To date, 6 genes for lamellar ichthyosis have been localized and 5 of them identified, as follows[2]: *TGM1* (14q11), *ABCA12* (2q34), 19p12-q12, 19p13, *ALOXE3-ALOX12B* (17p13), *ichthyin* (5q33).

Ichthyosis affects tissues of epidermal origin, there is scanty data about oral and dental involvement. These tissues are frequently affected both by disease and drugs used for

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treatment.[2] We, herein, report dental involvement a case of lamellar ichthyosis.

Case report

An 8-year-old girl reported to the Department of Pedodontia, with the chief complaint of pain in maxillary anterior region for two days. She was born to a nonconsanguineous couple as a collodion baby. She had generalized scaling since her early infancy and was diagnosed as a case of lamellar ichthyosis by dermatologist and had been treated with different moisturizers. No history of such disease was reported in the family. Patient did not suffer from any ocular and otolaryngeal abnormalities. No other systemic abnormalities detected.

The patient's IQ and physical growth was normal. On cutaneous examination, there were thick, armour-like brownish scales covering the entire body surface including all larger body flexures. Her hair was dry, sparse and eyebrows were scanty (**Figure 1**). There were no nail abnormalities. The mouth opening of the patient was reduced with multiple fissures on corner of the mouth bilaterally (**Figure 2**). On intraoral examination multiple carious lesions were found in the oral cavity involving all first and second deciduous molars, maxillary canines, central and lateral incisors (**Figure 3**).

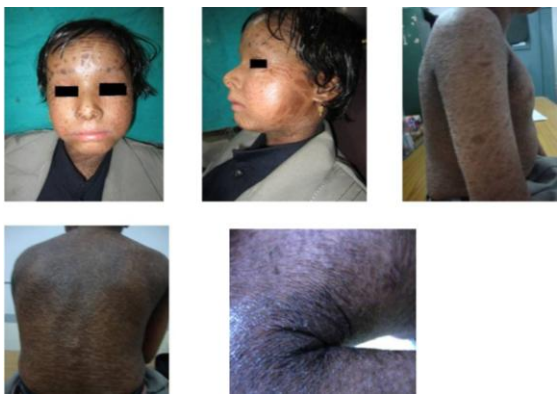

Figure 1 Clinical presentation of patient with lamellar ichthyosis.

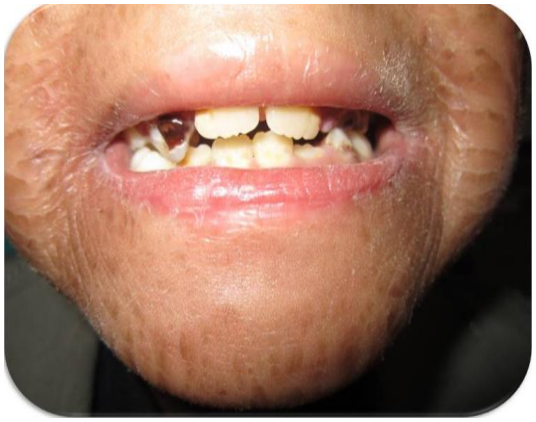

Figure 2 Reduced mouth opening with multiple fissures on corner of the mouth.

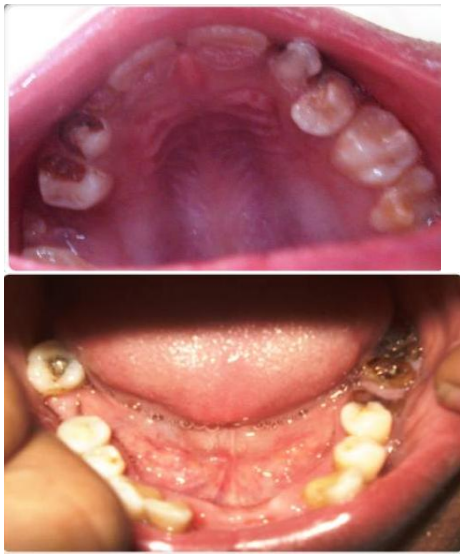

Figure 3 Multiple carious lesions in the oral cavity involving upper and lower jaw.

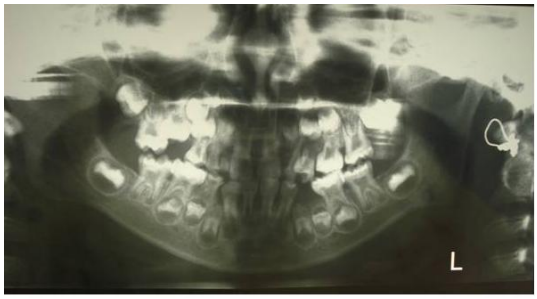

Figure 4 OPG shows mixed dentition without any enamel defects.

Radiological examination showed mixed dentition without any enamel defects; however, multiple carious lesions were seen on radiograph (**Figure 4**). No soft tissue abnormalities were appreciated. The dental management was initiated with a preventive approach as the oral hygiene was compromised and patient was under high

caries risk. Full mouth scaling and polishing was done followed by topical fluoride application. Pit and fissure sealants were applied. Glass ionomer restoration of 53,62,74,75 was done. Stainless steel crown was placed with 84 due to multi-surface decay. Adequate oral hygiene instructions and demonstration of brushing technique was performed for the patient.

For the management of his skin problem, he was referred to dermatologist.

Discussion

There is little knowledge about the oral manifestations of these disorders. In some patients teeth are normally developed but in others they are defective and likely to develop caries.[7,8] Oral and dental findings reported in persons with ichthyosis have included gingivitis, periodontitis, enamel hypoplasia, high caries incidence, delayed primary and secondary eruption, bruxism, alveolar ridging, bifid teeth, irregular morphology of teeth, and hyperkeratotic plaques on the tongue. The perioral, face and neck areas may be affected by ichthyosis. Angular cheilitis and facial dermatitis may occurs as side effects of oral retinoid theraphy.[9]

Our patient was a known case of lamellar ichthyosis, however, he had never undergone dental evaluation. On intraoral examination, there were multiple carious teeth. The patient had difficulty in opening his mouth secondary to angular cheilitis. Variable dental findings have been reported previously. Miteva[10] recorded both hair and dental abnormalities in his patient. List *et al.*[11] noticed abnormal deciduous and permanent teeth. Basel-Vanagaite *et al.*[12] described conical (deciduous) teeth or notched and pitted (permanent) teeth in three individuals with ichthyosis. Cremers *et al.*[13] observed early childhood deafness, congenital nonbullous ichthyosiform erythroderma, corneal involvement, photophobia, chronic blepharoconjunctivitis, hypotrichosis, anhidrosis, hyperkeratosis of the nails and dental dysplasia. Similarly, Bolgül *et al.*[14] reported missing teeth, carious teeth and persistent deciduous teeth in a 14-year-old boy with LI. In our case, there was no enamel defect, missing or persistent teeth.

Most of the times, patients with ichthyosis do not require any modification in dental treatment; however, the dentist should be aware of the concurrent medical problem and its treatment, as there is possibility of hepatic toxicity with the use of retinoids, which can affect the choice of local anesthetic agents during dental treatment. During dental treatment care must be taken to avoid manipulating the patient's skin, particularly in the perioral areas, since affected areas can be tender or friable.

References

1. Okulicz JF, Schwartz RA. Hereditary and acquired ichthyosis vulgaris. *Int J Dermatol.* 2003;**42**:95-8.
2. Vinzenz OJI, Heiko T. Ichthyoses: Differential diagnosis and molecular genetics. *Eur J Dermatol.* 2006;**16**:349-59.
3. Shwayder T, Ott F. All about ichthyosis. *Pediatr Clin North Am*. 1991;**38**:835-57.
4. Tor Shwayder. Ichthyosis in a nutshell. *Pediatr Rev*.1999;**20**:5-8
5. Schachner LA, Hansen RC, eds. *Pediatric Dermatology. 2n ed*. New York: Churchill Livingstone; 1996. P. 413-64.
6. Huber M, Rettler I, Bernasconi K. Mutations of keratinocytes transglutaminase in lamellar ichthyosis. *Sciences*. 1995;**267**:528.
7. Avrahami L, Maas S, Pasmanik-Chor M *et al.* Autosomal recessive ichthyosis with hypotrichosis syndrome: further delineation of the phenotype. *Clin Genet.* 2008;**74**:47-53.
8. List K, Hobson JP, Molinolo A *et al.* Co-localization of the channel activating protease prostasin/(CAP1/PRSS8) with its candidate activator, matriptase. *J Cell Physiol*. 2007;**213**:237-45.

9. Çakmak A, Baba F, Shermatov K *et al*. Treatment of congenital ichthyosis with acitretin. *Internet J Pediatr Neonatol*. 2008;**8**(1).
10. Miteva L. Keratitis, ichthyosis, and deafness (KID) syndrome. *Pediatr Dermatol*. 2002;**19**:513-6.
11. Basel-Vanagaite L, Attia R, Ishida-Yamamoto A *et al*. Autosomal recessive ichthyosis with hypotrichosis caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase. *Am J Hum Genet*. 2007;**80**:467-77.
12. Cremers CW, Philipsen VM, Mali JW. Deafness, ichthyosiform erythroderma, corneal involvement, photophobia and dental dysplasia. *J Laryngol Otol*. 1977;**91**:585-90.
13. Bolgül B, Hamamci N, Akdeniz S, Çelenk S. Oral manifestations of lamellar ichthyosis; a case report. *Iran J Pediatr*. 2009;**19**:298-302.