Case Report

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Oral Manifestations of Lamellar Ichthyosıs; A Case Report

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Abstract

Background: The name ichthyosis is derived from the Greek ikhthus meaning "fish" and refers to the similarity in appearance of the skin to fish scale. The ichthyoses are a heterogeneous group of disorders. There are few studies about the oral manifestations of these disorders. But early reports of ichthyosis in the Indian and Chinese literature date back to several hundred years.

Case Presentation: Oral manifestations of the 14-year-old female patient with ichthyosis are presented. Physical examination revealed thick, brownish scales covering the entire body surface including all larger body flexures and corneae. She had short and dry hair. There were no nail abnormalities and hearing loss.

Conclusion: We consider that this patient represents a new manifestation of lamellar ichthyosis disease, because congenitally teeth missing and cephalometric analysis measurements have not been reported before.

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Key Words: Lamellar ichthyosis; Teeth anomalies; Cephalometric analysis; Caries; Hypoplasia

Introduction

Ichthyoses are a heterogenous group of hereditary keratinization disorders that share in common the accumulation and shedding of large amounts of hyperkeratotic epidermis[1,2].

Depending on gene mutation that causes the disease, the skin problems later in life may range from a severe lamellar or bullous ichthyosis to mild or only focally expressed hyperkeratotic lesions[3]. There are several clinical signs of lamellar ichthyosis but there is little information about the oral manifestations of ichthyosis. To our knowledge, the case appears to be unique in the literature because there was no research on

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the congenitally missing teeth and cephalometric analysis in lamellar ichthyosis disease. This report presents oral symptoms and cephalometric measurements of a patient with lamellar ichthyosis.

The name ichthyosis is derived from the Greek ikhthus meaning "fish" and refers to the similarity in appearance of the skin to fish scale. Early reports of ichthyosis in the Indian and Chinese literature date back to several hundred years[4].

Case Presentation

A 14-year-old girl was admitted to our hospital for treatment of her teeth. The patient's medical history showed that lamellar ichthyosis had persisted since birth. Topical moisturizers were the only treatment the patient had received. There was consanguinity and a family history of the same disease. The patient consented to take part in this study. Ethical approval was obtained from the relevant local research ethics committees.

Physical examination revealed thick, brownish scales covering the entire body surface including all larger body flexures and corneae (Fig 1A). Her hair was short, dry and sparse (Fig 1B). There were no nail abnormalities. She did not suffer from deafness.

In the intraoral and radiological examination, upper and lower lateral incisors; second premolars and upper right first premolar were congenitally missing (Fig 2). All second deciduous molars, upper right deciduous canine and lower left deciduous canine were persistent. Enamel defect was not observed, but two deciduous lower second molars were carious.

The craniofacial growth of the patient was evaluated with cephalometric analysis. Cephalometric landmark definitions and locations are mentioned in table 1. In our patient, these parameters were:
SNA: 82°, SNB: 77° and ANB: 5°. Angles showing sagittal values were in normal ranges[5] with the normal ranges of vertical measurements (SNGoGn: 30°); the mandible of the patient rotated posteriorly (Y axis angle: 65°). The patient whose soft tissue measurements were in normal ranges (SD-UL: 12 mm, LI-LL: 13 mm) had a convex profile (Convexity angle: 156°). The nasolabial angle (132°) was highly increased (Fig 3, Table 2).

Fig 1: A-Thick, brownish scales covering the entire body surface including all larger body flexures and corneal involvement; **B-** Short, dry and sparse hair

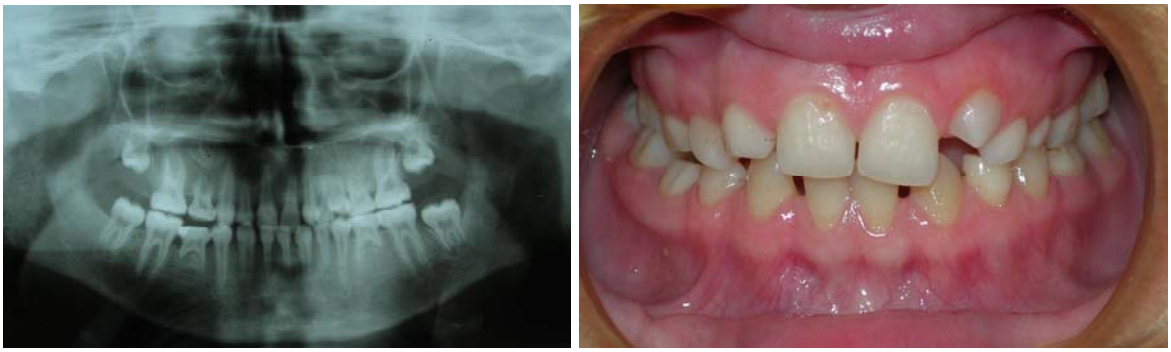

Fig 2: Panoramic view Figure (A) and intraoral photography (B) in a 14-year-old girl with lamellar ichthyosis

Table 1: Cephalometric landmark definitions and locations

Cephalometric landmark	definitions or locations
Sella (S)	is located in the center of the outline of the sella turcica
Nasion (N)	is located at the most inferior, anterior point on the frontonasal suture
Point A	is located at the most posterior part of the anterior shadow of the maxilla
Point B	is located at the most posterior point on the shadow of the anterior border of the mandible
Gonion	is the midpoint of the angle of the mandible.
Gnathion	is located at a point on the shadow of the chin midway between pogonion and menton
Menton	is located at the most inferior point on the shadow of the chin
ANS	anterior nasal spine
SNA	SNA (sella-nasion-point A): angle formed by intersection of S-N line and N-A line at N
SNB	SNB (sella-nasion-point B): angle formed by intersection of S-N line and N-B line at N
ANB	Point A-nasion-point B: difference between angles SNA and SNB
SN-GoMe Angle (mandibular plane angle)	The angle formed by intersection of S-N plane and Go-Me plane
Y axis Angle	anterior inferior angle formed by intersection of horizontal plane with S-Gn line
ANS-Me/N-Me	the ratio between ANS-Me plane and N-Me plane
Nasolabial Angle	Nasolabial angle: the angle between the Sn-Ls plane and a tangent drawn to the columella
SD-UL	the distance between the contact point of maxillary bone with upper central incisor and upper lip
LI-LL	the distance between the lower central incisor and lower lip

Discussion

Lamellar ichyhyosis is apparent at birth, and the newborn usually presents encased in a collodion membrane, a translucent covering that desquamates over the subsequent 10 to 14 days[3]. It is obviously important, but

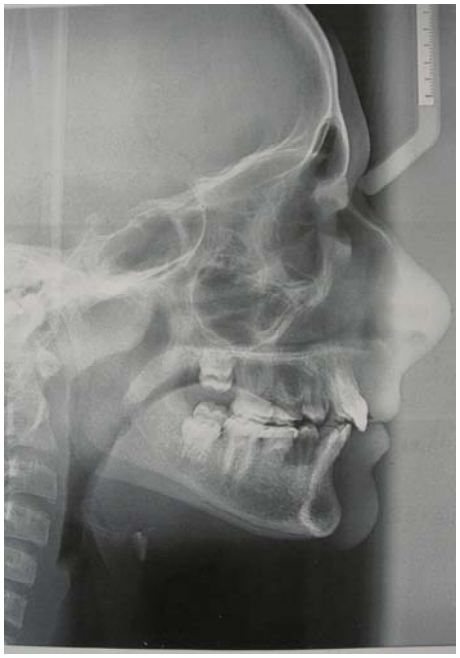

Fig 3: Cephalometric view in our patient with lamellar ichthyosis

Table 2: Cephalometric measurements

Cephalometric parameter	Our patient's parameter	Clinical norms
SNA (°)	82	82±2
SNB (°)	77	80±2
ANB (°)	5	1-5
SNGoMe (°)	30	32±4
Y axis angle	65	59.5
Convexity angle	156	160-165
ANS-Me/N-Me (%)	52.3	52-56
SD-UL (mm)	12	10-14
LI-LL (mm)	13	10-14
Nasolabial angle (°)	132	102±8

sometimes painstakingly difficult to make a correct diagnosis already in infancy.

Fortunately, recent advances in our understanding of the molecular genetics of ichthyosis have led to several new diagnostic tools that are continuously being updated[3]. In lamellar ichthyosis, the scales are large, adherent, dark and pigmented with no skin erythema[6]. There are periodic exacerbations and remissions, but complete clearing of the skin was never noted [7]. Scalp hair has always been sparse and short [8]. The clinical features of patients with lamellar ichthyosis are generally similar to our case, which is typical.

There is little knowledge about the oral manifestations of these disorders. In some patients teeth are normally developed[9,10], but in others they are defective and likely to develop caries [11,12]. Miteva noted both hair and dental abnormalities in his patient [6]. List et al determined abnormal deciduous and permanent teeth[13]. Basel-Vanagaite et al described conical (deciduous) teeth or notched and pitted (permanent) teeth in three individuals with ichthyosis and hypotrichosis[14]. However, they reported that matriptase is highly expressed in the enamel-producing ameloblasts of develop-ing teeth, and they implicated impaired matriptase proteolytic activity as the underlying cause of ichthyosis with hypotrichosis [14]. Examination at high magnification revealed a rougher enamel surface of ST14 hypomorphic incisors and molars with markedly increased bacterial growth, suggesting a direct role of matriptase in enamelogenesis [15]. Cremers et al observed early childhood deafness, congenital non-bullous ichthyosiform erythroderma, corneal involvement, photo-phobia, chronic blepharoconjunctivitis, hy-potrichosis, anhidrosis, hyperkeratosis of the nails and dental dysplasia in their patient[16].

In another study, it is reported that additional clinical features included abnormalities of the teeth and nails[17]. In our patient, in addition to typical clinical features, nine teeth were congenitally missing and also our patient whose soft tissue measurements were in normal ranges had a convex profile and the nasolabial angle was highly increased.

Respiratory movements are restricted; secondary infection and septicemia are frequent complications. Death usually occurs in the first few weeks of life [18]. Our patient's two cousins had also died due to ichyhyosis in the first week of life.

Conclusion

We consider that this patient represents a new manifestation of lamellar ichthyosis disease, because congenitally teeth missing and cephalometric analysis measurements have not been reported before.

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