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10.2478/AMB-2024-0023

CASE REPORT

HEREDITARY ECTODERMAL DYSPLASIA IN TWO IDENTICAL SIBLINGS

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Abstract. *Primary defects in two or more ectodermally-derived tissues during embryonic development characterize ectodermal dysplasia, a vast, varied group of inherited illnesses. Skin, hair, nails, eccrine glands, and teeth are the primary tissues affected. Most cases of ectodermal dysplasia are caused by the X-linked recessive form of the disease (also known as Christ–Siemens–Touraine syndrome), which is passed down from female carriers to their male offspring. It is characterized by an absence of sweat glands (hypohidrosis or anhidrosis), malformed teeth (anodontia or hypodontia), and scant hair (atrichosis or hypotrichosis). Lack of teeth and unusual look were cited as major causes for alarm. The usual manifestations of hypohidrotic hereditary ectodermal dysplasia have been described in two case reports. Two identical siblings with possible typically X-linked recessive hypohidrotic ectodermal dysplasia are described here. Despite the lack of a cure, patients can benefit from a multidisciplinary approach to treatment planning and an expedient diagnosis.*

Key words: *anodontia, consanguineous siblings, hereditary disease, hypotrichosis, hypohidrosis*

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Received: *07 June 2023;* ***Revised/Accepted:*** *04 April 2024*

INTRODUCTION

Ectodermal dysplasia (ED) refers to a group of rare genetic disorders characterized by a wide variety of primary developmental abnormalities. Two or more ectodermal structures, such as epidermis, nails, teeth, hair, or sweat glands, are affected. Additionally, it can influence the development of other embryonic ectodermal organs, including the eye, ear, neural, and adrenal tissues, to variable degrees [1]. Based on the level of sweat gland function, there are two basic forms of ectodermal dysplasia. The first type of hypohidrotic disorder is both autosomal and X-linked and is defined by the typical trio of hypodontia, hypohidrosis, and hypotrichosis. The hydrotic type, in contrast, is an autosomal trait that is characterized by the absence of sweat gland involvement [1, 2]. The original ectodermal dysplasia categorization system was established in 1982 by Freire-Maia and Pinheiro, with updates in 1994 and 2001. The group A of ED included cases where a minimum of two of the four typical ectodermal structures were affected; the group B of ED had a defect in one classic ectodermal structure plus a deficit in another ectodermal structure (e.g., ears, lips). Hypohidrotic (anhidrotic) (HAED) and hidrotic (HED) syndromes are the most prevalent types of ectodermal dysplasia [3, 4]. Defective ectodysplasin A, caused by mutations in the EDA, EDAR, or EDAR-

ADD genes, is involved in a structure that hinders the regular development of teeth, sweat glands, and hair by blocking the regular contacts between the ectoderm and the mesoderm. The traits of hypohidrotic ectodermal dysplasia (HED) are caused by the aberrant production of these ectodermal structures [5]. Cleft lip and palate are not the only possible oral signs of ectodermal dysplasia; anodontia and hypodontia are also prevalent. Other distinguishing features include prominent cheekbones, thick lips, a broad, depressed nose bridge, pigmentation around the mouth and eyes, and deformed ear [6].

Two identical siblings with possible typically X-linked recessive hypohidrotic ectodermal dysplasia are described here.

CASE PRESENTATION

Two male patients of Indian origin, aged 17, nontwins, presented to our Department of Oral Medicine, Diagnosis, and Radiology, with the major complaint of toothlessness from infancy (Fig. 1).

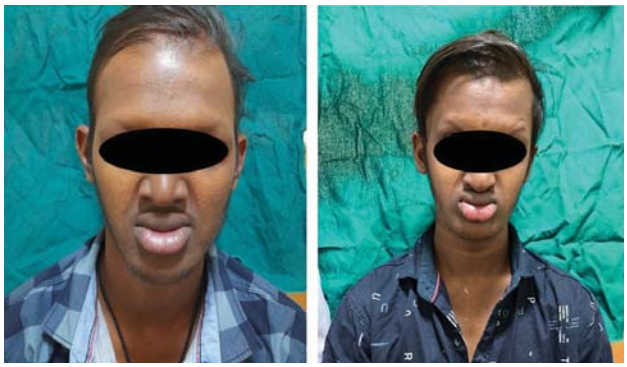

Fig. 1. Two 17-year-old siblings showing similar features of ectodermal dysplasia

There was no history of difficulties during the delivery of either sibling, and they both presented with identical symptoms. The parents also reported a favorable family history, with no indication of the previously listed features on either the mother's or father's sides of the patients and with no other healthy siblings. Both patients had average IQ scores and no visual or hearing impairments. Immune status of the patients was not evaluated. The clinical examination revealed dry, scaly skin, with a somewhat elevated fever. The patient hair was fine, sparse, and lusterless, and the eyebrows were thinner in the patient 1 compared to patient 2. The eye examination indicated corneal dryness and reduced tear production/lubrication. The finger inspection revealed fingers with thin, weak nails (Fig. 2).

The examination of the ears showed big, low-set ears with midface hypoplasia, which gave the patients an older appearance in comparison to other individuals of the same age; the patients are with average intelligence. The patient microsomia was characterized by a saddle-shaped nose, thick, outwards curved lips, and a noticeable double lip. Both the patients exhibited very significant intraoral examination findings as dry mucous membrane, full anodontia of both the maxilla and mandible, and decreased vertical heights of both arches (Fig. 3).

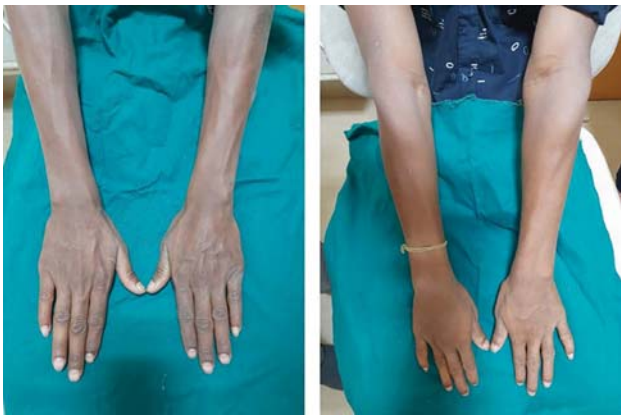

Fig. 2. Forearms with scanty or no hair, with dry, scaly skin and thin brittle nails

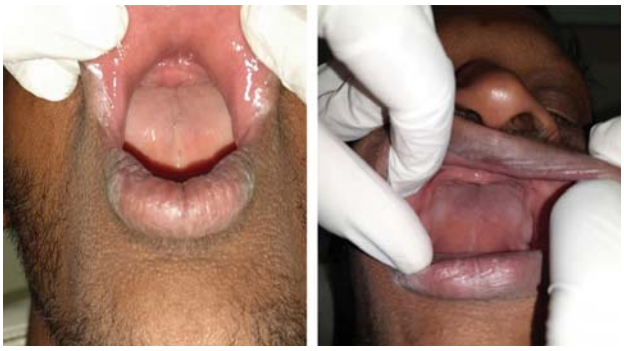

Fig. 3. Intraoral examination of both siblings with anodontia of both arches.

The patients underwent radiographic studies, in which orthopantomograph revealed a complete absence of teeth on both the maxillary and mandibular arches with hypoplastic alveolar ridges (Fig. 4).

Hypoplastic nasal conchae along with deficient nasal septum were the other significant findings noted along with the gross alteration of both condylar and coronoid processes.

DISCUSSION

Ectodermal dysplasia (ED) refers to a rare set of conditions as heterogeneous, inherited disorders distinguished by the aberrant development of many ectodermal tissues during embryonic development. Congenital ED is a nonprogressive condition that can

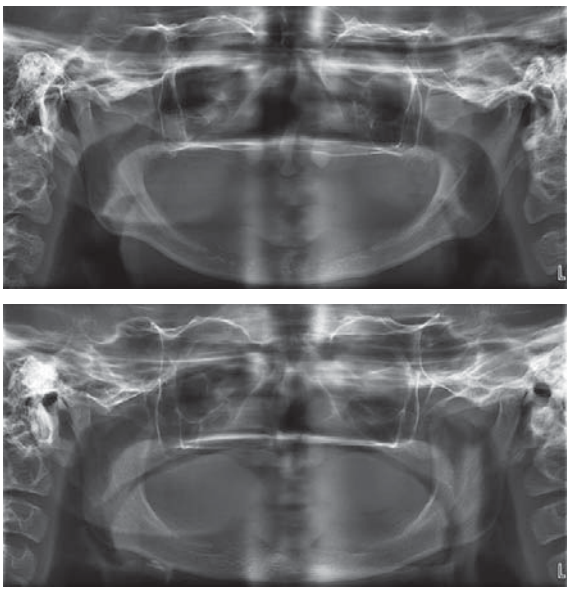

Fig. 4. Panoramic imaging of both siblings with anodontia of the upper and lower arches with hypoplastic alveolar crest

be a syndrome when other issues are present or an isolated disorder when the impairment is confined to the ectodermal tissues. The most prevalent forms of ectodermal dysplasia are X-linked recessive hypohidrotic ectodermal dysplasia and hidrotic ectodermal dysplasia [7]. Ectodermal dysplasia is a common kind of congenital malformation that does not worsen with time. Thurnam described the illness, but Weech used the term 'ectodermal dysplasia' in 1929 [4].

Malformed teeth and severely sparse hair are only two examples of the aberrant ectodermal features that must be available to diagnose ectodermal dysplasia. When missing teeth or a delay in teething generate parental anxiety and prompt the doctor to assume ectodermal dysplasia, ectodermal dysplasia is usually diagnosed. It is well known that dental findings at the emergency department can vary from hypodontia/oligodontia to anodontia of primary and permanent teeth. However, delivery without primary teeth is quite unusual [2, 3, 8]. In this instance, however, both the maxillary and mandibular arches had full anodontia including both primary and permanent dentitions along with the history of consanguinity. Conical maxillary incisors also contributes to the diagnosis of ectodermal dysplasia.

Radiographic examination revealed undeveloped alveolar ridges in both cases, despite the fact that growth of the jaws was unaffected (orthopantomograph); this is because the alveolar process does not grow when there are no teeth. This makes the usual vertical measurement reduced in size. Identifying probable female carriers is facilitated by phenotypic tests, such as evaluation of perspiration and dental abnormalities [6].

Two perspiration measurement methods have been developed: On the back of the female carrier, the initial sweat test reveals a V-shaped pattern of streaks that matches to the lines of Blaschko. Counting sweat pores along the ridges of the fingertips or hands is the alternative way. Hypotrichosis can be documented by means of pilocarpine iontophoresis and sweat pore counts with yellow starch iodine [9]. The combination of a dental exam and a sweat test considerably boosts the chance of accurately identifying female HED carriers. Radiographs of the teeth can give useful supplemental information and act as a simple screening test for carrier status. Differential diagnosis includes illnesses such as alopecia areata, regional dermal hypoplasia, incontinentia pigmenti, and dyskeratosis congenital [10, 11]. Early intervention is crucial to the effective and successful management of ED regardless of the affected organ. Physical cooling strategies, such drinking cold liquids often and wearing special cooling vests to decrease heat generation during exercises, are essential for managing heat exposure. Improvements in both the health and appearance of teeth can result from prompt dental care. Bone grafting/sinus lift procedures, dental implants, and dental prostheses are all treatments that should be performed under the guidance of an orthodontist. The use of Minoxidil 3% to stimulate hair growth in people with ectodermal dysplasia is a relatively new treatment strategy. Topical and systemic emollients are the drugs of choice for treating xerosis and wounds [12].

In addition; this disease requires thorough observation and regular follow-up part of the treatment approach. In the above clinical case, complete upper and lower prostheses were recommended until the implant evaluation was complete. Supportive treatments such as drinking water at regular intervals, wearing loose clothing, and using artificial saliva, were also recommended. Additionally, the patients were instructed to consult a genetic counsellor for genetic analysis.

CONCLUSION

The clinical manifestations of ectodermal dysplasia in afflicted individuals lead to substantial social challenges and decreased quality of life. Because of the difficulty of diagnosing and treating these congenital conditions early in life, the involvement of the oral physician is crucial. A multimodality approach is required for treatment planning, which ultimately helps the patient. Regarding dental issues, oral rehabilitation remains the cornerstone of treatment. Examining the function and effects of gene mutations will be a fascinating endeavor that will provide a new door for

the advancement of therapeutic treatments. With the exception of cases of ectodermal dysplasia coupled with immunodeficiency, the prognosis for ectodermal dysplasia is favorable, and the patient life expectancy is generally normal.

Acknowledgements: *None.*

Disclosure Summary: *The authors have nothing to disclose.*

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