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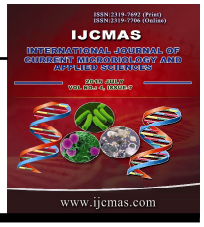

Case Study

Hydrotic or Hypohydrotic Ectodermal Dysplasia: Diagnostic Dilemmas (Case Report)

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Keywords

Ectodermal dysplasia, Hypohidrosis, Anhidrosis

A B S T R A C T

Ectodermal dysplasia is a rare genetic syndrome, caused by developmental defects in ectoderm-derived tissues. Patients with ectodermal dysplasia often suffer such symptoms as hypodontia or anodentia, thin and sparse hair, lack of perspiration glands, dry skin, and deformed nails of the fingers and toes. Its incidence rate is about 7 in 1000 births. This study describes a case of ectodermal dysplasia without loss of teeth, but with other ectodermal impairments.

Introduction

Ectodermal dysplasia includes a large group of syndromes that are clinically and genetically heterogeneous, and are identified by anomalies in structures of ectodermal-origin, and can present with disorders in such structures as hair, nail, teeth, sweat glands, sebaceous glands and conjunctiva and nervous system (Ramesh *et al.,* 2010). This syndrome was first reported by Charles Darwin (1860) (Mirkarimi *et al.,* 2010).

Its incidence rate is estimated at 7 cases per 1000 births, and more than 192 different disorders have been identified in this disease (Ramesh *et al.,* 2010). It is usually inherited as a X-linked recessive trait (Makdonald *et al.,* 2011; Casamassimo *et al.*,2013), and is more prevalent in men than in women (Sharma and Mamatha, 2008).

A major problem of patients with ectodermal dysplasia that is common in almost all patients is hypodentia or anodontia. If any teeth are present, they are conical or peg-shaped. Patients' hair is thin and sparse, and their skin is dry due to lack of perspiration. Thus, these people can barely tolerate heat. Also, in many cases, finger and toe nails are deformed (Mirkarimi *et al.,* 2010). The disorder may occur in the first trimester of pregnancy, and it will affect the teeth if severe and appear before the sixth gestational week. After the 8th gestational week, other ectodermal

structures may also be affected (Sharma and Mamatha, 2008). Prenatal diagnosis of ectodermal dysplasia is performed by fetal skin biopsy with photoscopy in the 20thgestational week (Babu *et al.,* 2011). The disease is seen in two hydrotic and anhydrotic (hypohydrotic) forms. Felsher in 1944 changed the adjective anhydrotic to hypohydrotic because the persons with hypohydrotic form are not truly devoid of all sweat glands. A definitive classification of ectodermal dysplasia (ED) is difficult to formulate since many of the syndrome that involve ED have overlapping features. A simple attempt made by Nelson included five categories, namely Hypohydrotic (anhydrotic), Hydrotic (Clouston's syndrome), EEC (Ectodactyly ectodermal dysplasia) syndrome, Rapp –Hodgkin syndrome and Robinson's disease (Sharma and Mamatha, 2008).

Case Report

The patient was a 7-year-old girl, admitted to pediatrics ward in School of Dentistry of Tehran University of Medical Sciences for treatment of her teeth. Intraoral examination revealed several dental caries (Figure 1), and patient suffered from xerostomia. After milking test, no saliva was excreted from parotid and submandibular glands (Figure 2). Patient history revealed symptoms like absence of tears and perspiration as well, and patient consumed a lot of water due to hyperthermia. Dry skin and cracked lips were also evident in patient's clinical examination (Figure 3). Radiographic examination showed presence of all of the patient's primary and permanent teeth, and no dental deformities or shortage of dental buds. Patient's nails were also normal (Figure 4). Considering involvement of a number of ectodermal structures, ectodermal dysplasia took diagnostic priority. Skin biopsy was done by Dermatologist in Razi Hospital in Tehran. According to skin biopsy report, mild hyperkeratosis, pigmentation of basal layer, lymphocytic infiltration melanophages and mastocytes were observed around vessels with cell. A number of sweat glands existed in the dermis, but no sebaceous glands were found in the sample. Biopsy resulted in diagnosis of Hydrotic ectodermal dysplasia. Sweat test was also performed on the patient, and insufficient sweat was reported.

History of no disease was found in patient's medical history, and patient's parents did not mention any family history of ectodermal dysplasia. Patient's father and mother were consanguineous relatives (cousins), but no symptom of the disease was observed in them.

In the present patient, dental caries were repaired, and Biotene mouth-wash was administered for xerostomia. Fluoride-therapy and regular periodic examinations were also recommended.

Discussion

Ectodermal dysplasia is an extremely rare genetic disorder characterized by faulty development of ectodermal structures (Babu *et al.,* 2011). People with ectodermal dysplasia may present with different combinations of ectodermal structural defects. In a person, hair and nails may be involved, while in another person, teeth and sweat glands. Each combination is considered a separate type of ectodermal dysplasia (Bari and Rahman, 2007). Freire Maia and Pinheir proposed a numerical system for this disorder, in which a number is assigned to every ectodermal structure involved: hair (1), teeth (2), nails (3), and sweat gland (4). A total of 10 different ectodermal dysplasia subgroups were proposed in their review article (for

example: 1-2-3-4, and 1-2-3) (Babu *et al.*, 2011).

These patients' main concern was lack of teeth and particular appearance associated with this disease. The most common finding is reduced number of teeth and their deformed shape, as well as delayed growth of teeth, which is often the first step in diagnosing the disease (Babu *et al.*, 2011). Such attributes were not found in the present patient, and unlike previous studies, all primary and permanent teeth were present in clinical and radiological assessment (Sharma and Mamatha, 2008; Babu *et al.*, 2011; Bari and Rahman, 2007; Mangalore, 2002; Mohammed *et al.*, 1994), instead, xerostomia, and lack of tears and sweat clinically confirmed the diagnosis of ectodermal dysplasia. Other ectodermal structures had not been affected.

In these patients, lack of tear could be caused by complete or partial loss of lacrimal glands or deformity in their ducts (Mohammed *et al.*, 1994). Studies by Shaw and Basserman Nielsen report xerostomia and cracked lips can be due to full or partial lack of secondary salivary glands (Mohammed *et al.*, 1994), and the patient in the present study had such symptoms, as well as normal shape of nails, which is similar to that in studies by Shaw and Sharma (2008).

Skin biopsy of the present patient showed only a few sweat glands in the dermis, and no sebaceous glands. Hydrotic Ectodermal Dysplasia was diagnosis resulting from biopsy. However, since patient suffered from dry skin and his sweat glands did not produce any moisture, diagnosis was also confirmed after performing sweat gland function test. Therefore, hypohydrotic ectodermal dysplasia was diagnosed. Table 1 shows clinical differences between hypohydrotic and hydrotic ectodermal dysplasia (Sharma and Mamatha, 2008; Varghese and Sathyan, 2011).

According to table 1, in the present study patient, smooth dark hair, normal lips, and not flattened nasal bridge are similar to reported attributes in hydrotic type, but due to characteristics like reduced sweat glands and normal nails, it is similar to the hypohydrotic type. As stated earlier, hypodentia or anodontia was not found in the patient, which is a prominent characteristic, for which patient was reported. Thus, it seems, patients with ectodermal dysplasia have a variety of signs and symptoms that do not accurately place them in either hydrotic or hypohydrotic group.

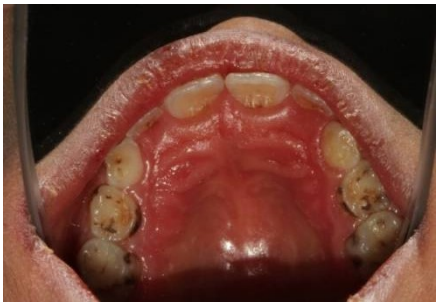
Extraoral characteristics observed in ectodermal dysplasia include prominent and square forehead, prominent supraorbital ridge, depressed nasal bridge, midface hypoplasia, pointed chin, protruberent and everted lips, which were not found in the present patient (Babu *et al.*, 2011; Mortier and Wackens, 2004).

In differential diagnosis of the diseases, it is difficult to differentiate recessive autosomal hypohydrotic ectodermal dysplasia from x-linked hypohydrotic form. Clinical characteristics are similar in both, but because of different inheritance, the autosomal recessive involves both men and women, and heterozygous condition does not show any symptoms. To differentiate between Hypohydrotic x-linked ectodermal dysplasia and autosomal recessive, full family history should be reviewed. Such findings as equal involvement of men and women within the family, as well as consanguineous parents, increases chances of the trait or disease presenting as autosomal recessive inheritance (Mortier and Wackens, 2004).

Table.1 Differences between the hydrotic and hypohydrotic forms of ectodermal dysplasia

	Hydrotic	**Hypohydrotic**
Mode of Inheritance	Most often autosomal dominant	Most often autosomal recessive
Scalp Hair	Soft, dawny,color is darker	Fine in texture,fair and short
Teeth	Anodontia To hypodontia	Anodontia to hypodontia
Lips	No abnormality	Protruding
Sweat glands	Active	Reduced to absent
Nasal bridge	No flattening	Underdeveloped
Nails	Dystrophic nails	No abnormality
Eyebrows	Frequently absent	Absent
Eyelashes/Pubic/Axillar y hairs	Scanty/absent	Variably affected

Figure.1 Intraoral photograph showing several dental caries



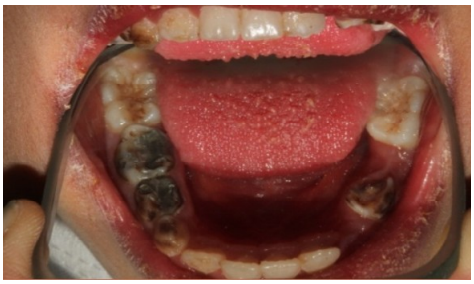


Figure.2 Milking test of parotid glands

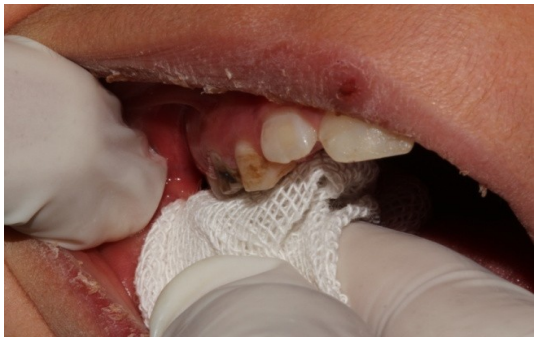


Figure.3 Cracked lips

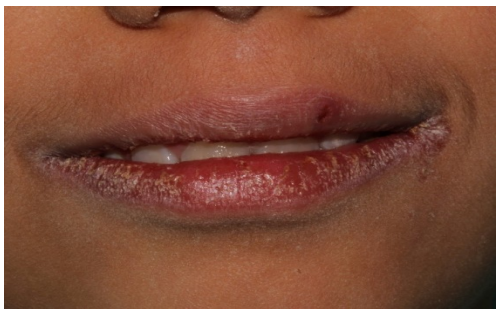


Figure.4 Patient's normal nails

Given consanguineous parents and female gender of the present patient, it seems the disease has been inherited as autosomal recessive. Since girls inherit an X chromosome from their father and another from mother, if the disease is inherited as x-linked in the present patient, then her father should also have some signs of the disease as well. But, neither of the parents showed any symptoms.

There is no definitive pharmacological treatment for these patients, and their management depends on the structures involved. Use of artificial tear and skin moisturizers are recommended for these patients, and they should avoid excessive exposure to warm climates and heavy physical activities (Bari and Rahman, 2007). Early dental examination is required, and dental radiography and measurement of salivary flow should be carried out as soon as child is able to cooperate. Reduced salivary flow is associated with dental caries, and preventive treatments, including fluoride-therapy and regular periodic examinations should also be performed (Bergendal *et al.,* 2011). These patients live just as long as normal people do (Makdonald *et al.,* 2011; Bari and Rahman, 2007).

In conclusion, Ectodermal dysplasia is a rare genetic disease that involves different body tissues of ectoderm origin. Full and accurate examination leads to the diagnosis of these patients. Restoring normal function is the principle aim in treatment of these patients.

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