ELSEVIER

Archives of Medical Research 37 (2006) 403–409

Archives of Medical Research

BRIEF REPORT

Ectodermal Dysplasia: Retrospective Study of Fifteen Cases

Izzet Yavuz,[a] Zelal Baskan,[b] Refik Ulku,[c] Turksel Coruh Dulgergil,[d] Osman Dari,[e] Aydin Ece,[f] Yasemin Yavuz,[g] and Kadriye Oya Dari[h]

[a]*Department of Pediatrics,* [b]*Department of Prosthetic Dentistry,* [c]*Department of Orthodontics,* [h]*Department of Periodontology, Faculty of Dentistry, Dicle University, Diyarbakir, Turkey*

[c]*Department of Thoracic Surgery,* [f]*Department of Pediatrics, Faculty of Medicine, Dicle University, Diyarbakir, Turkey,*

[d]*Department of Operative Dentistry, Faculty of Dentistry, Kirikkale University, Kirikkale, Turkey*

[g]*Community Centre of Oral and Teeth Health, Diyarbakir, Turkey*

Received for publication March 24, 2005; accepted June 29, 2005 (ARCMED-D-05-00113).

The aim of this article is to review possible cranio-maxillofacial deformative consequences associated with hypohidrotic ectodermal dysplasia and embryonic malformations, which include dental ageneses, and describe the oral habilitation. Hypohidrotic ectodermal dysplasia patients had a clinical examination and underwent radiographic and Steiner's analyses and a respiratory capability test before assessment and treatment. Fifteen patients (eight males and seven females, aged 5–45 years) had tooth ageneses (from hypodontia to anodontia) associated with cutaneous dyshydrosis and hair and nail dystrophy. Most patients had sparse or absent hair, a short face with an unusual facial concavity, a maxillary retrusion and a relative mandibular protrusion. Dentists must conduct a comprehensive and multidisciplinary approach to these patients in order to improve their dental, masticatory, growth and orthognathic conditions. © 2006 IMSS. Published by Elsevier Inc.

Key Words: Hypohidrotic ectodermal dysplasias, Denture, Tooth ageneses.

Introduction

Ectodermal dysplasia (ED) is a large and complex group of disorders defined by the abnormal development of two or more structures derived from the ectodermal layer. The most frequently reported manifestation of ED is hypohidrotic dysplasia (HED), also termed Christ-Siemens-Touraine syndrome, and anhydrotic dysplasia, as in our cases. The ectoderm, one of three germ layers present in the developing embryo, gives rise to the central nervous system, peripheral nervous system, sweat glands, hair, nails, and tooth enamel (1,2).

As a result, HED patients exhibit the following clinical signs: hypotrichosis, hypohidrosis, and cranial abnormalities. Patients often exhibit a smaller than normal face because of frontal bossing, a depressed nasal bridge, the absence of sweat glands resulting in very smooth, dry skin and/or hyperkeratosis of hands and feet. Oral traits may express themselves as anodontia, hypodontia, and conical teeth. Anodontia also manifests itself by a lack of alveolar ridge development (1,3,4).

The earliest recorded cases of ED were described in 1792 (1). Since then, more than 200 different pathologic clinical conditions have been recognized and defined as ED. These disorders are considered relatively rare, 1 in 10,000–1 in 100,000 births (1–3,5,6).

Clinical manifestations of HED also cause considerable social problems in affected patients. Dental treatments of the clinical traits of HED can have a profound impact on these patients. The ability to look and feel like their peers is imperative for the psychological development of these patients. The literature has demonstrated the benefits that corrective dentistry has for the self-esteem and social well being of these patients (1,7,8).

Our major goals of providing dental and medical management were to provide comfort to patients to be as other healthy individuals. Depending on their ages and their abnormalities, patients underwent periodontal therapy, caries

Address reprint requests to: Dr. Izzet Yavuz, Assistant Professor, Department of Pediatric Dentistry, Faculty of Dentistry, University of Dicle, Diyarbakir 21280, Turkey; E-mail: iyavuz@dicle.edu.tr

0188-4409/06 $–see front matter.
doi: 10.1016/j.arcmed.2005.06.016

management or prosthodontic or orthodontic treatment. Implanting and orthognatic surgery was reserved for full-grown patients.

Patients and Methods

This retrospective study was carried out using patients applying to our university dental clinic from 1997–2005. In our dental faculty, 15 cases (eight males and seven females, aged 5–45 years) with a diagnosis of HED were included.

All major signs of HED were studied, such as sparse hair (trichodysplasia), smooth skin (hypohidrosis), abnormal fingernails and toenails, and cranial and facial abnormalities. The pedigree of the patients was researched (Table 1, Figures 1–9).

Each patient had the benefit of a rigorous clinical examination for diagnosis and therapy: minor or major abnormalities had to be detected in both patients and family. Examination included the skull, face, hair, teeth, nails, skin, lungs, sweat glands, etc. (Table 2).

Results of the maxillofacial cephalometric analyses are summarized in Table 3, and Steiner analysis was used to determine abnormalities.

Results

Fifteen cases (eight males and seven females, aged 5–45 years) had tooth ageneses (all cases: from hypodontia to anodontia), associated with cutaneous dyshidrosis, sparse or absent hair (cases 1–8, 12–15), nail dystrophy (cases 1, 3, 4, 6, 11–15), and hypohidrosis (all except patient 11).

The skin on most of the body was abnormally thin, dry, and soft with an abnormal lack of pigmentation (hypopigmentation: patients 4, 6, 7, and 12); however, the skin around the eyes (periorbital) was darkly pigmented (hyperpigmentation: cases 1, 6, 7, 11, 12, 15) and finely wrinkled, appearing prematurely aged (Tables 1 and 2).

Fifteen cases had hypodontia, six cases with fewer than ten teeth (cases 1, 2, 6, 11–13), and nine cases with more than ten teeth (cases 1, 3–5, 7–10, and 15).

Table 1. Report of clinical characteristics for each of the 15 cases

Abnormality	15 cases (100%)
Trichodysplasia (sparse or lack of hair)	12 cases (80%)
Hypohidrosis (from moderate to severe)	13 cases (86%)
Abnormal finger and toenails	12 cases (80%)
Protuberant lips	14 cases (93%)
Saddle nose	11 cases (73%)
Fever history	12 cases (80%)
Asthma and difficulty in breathing	7 cases (46%)
Peeling skin	14 cases (93%)
Deafness (hearing loss from moderate to severe)	8 cases (53%)
Relationship of parents to each other	8 cases (53%)

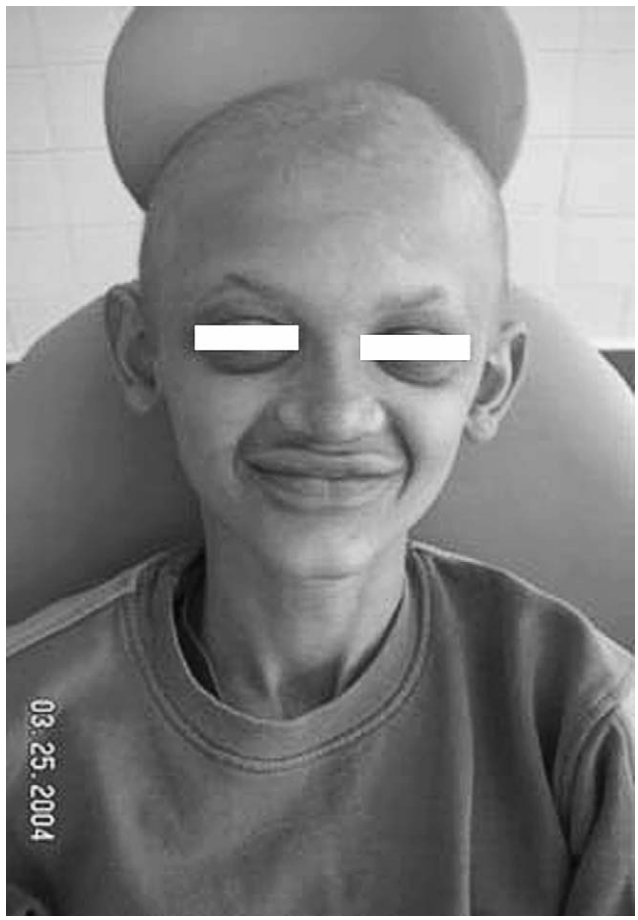

Figure 1. Maxillofacial examinations point out a significantly reduced anterior facial height caused by the collapse of the distance from anterior nasal spine to chin. The presence of a prognatic mandible also contributes to the mild or severe facial profile concavity. Also, HED is presented with an abnormal bulging forehead with high-implanted brittle hair (patient 6).

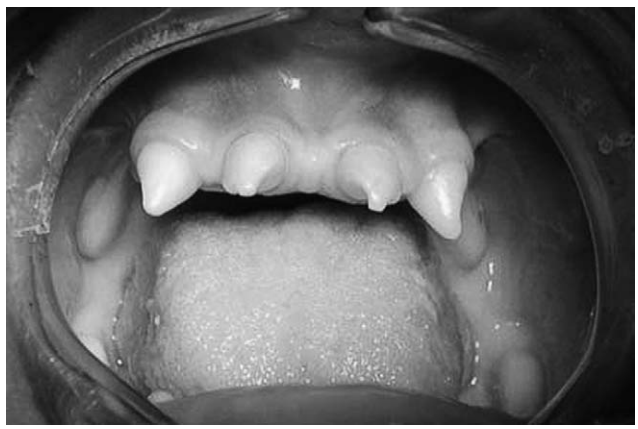

Figure 2. Mouth examination is of primary necessity. It allows mucosal assessment (dryness, thickness, and brittleness), dental inspection (number, form, and dysenamelogeneses) and an alveolar crest height estimation (patient 6).