Treatment with removable prosthesis in hypohidrotic ectodermal dysplasia. A clinical case

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Summary

Ectodermal dysplasias form part of a wide range of syndromes presenting abnormal development of two or more tissues derived from the ectoderm.

Hypohidrotic ectodermal dysplasia is a congenital syndrome, characterized by hypotrichosis (hair is sparse, fine and weak; anomalies in the skin and nails), hypohidrosis (due to the paucity of sweat glands which in turn gives rise to sweat disorders) and hypodontia (partial, and occasionally total, absence of primary and/or permanent dentition). A case of a child with hypohidrotic ectodermal dysplasia with oligodontia and marked resorption of the maxillary and mandibular alveolar ridges is presented. A prosthetic rehabilitation in the form of a removable acrylic prosthesis was made, achieving excellent esthetics, functionality and adaptation, thanks to which a considerable improvement in self-esteem has been obtained.

Key words: *Hypohidrotic ectodermal dysplasia, ectodysplasin A, oligodontia, removable prosthesis.*

Introduction

Ectodermal dysplasias are found within the extensive group of diseases which in common present abnormal development of two or more tissues derived from the ectoderm. They are non-progressive, diffuse, congenital genodermatoses, characterized by a lack or scarcity of hair, teeth, nails and eccrine sweat glands, to which can be added defects in the external morphology (nose, outer ears, and lips), disorders of the CNS; alterations of the eyes; anomalies in the oronasal mucosa and in melanocytes (1).

Ectodermal dysplasia is a relatively rare disorder, with a frequency varying between 1:10,000 and 1:100,000 live births, and is more frequent in males. The majority of cases follow the autosomal-recessive mode of inheritance, but it can also be autosomal-dominant or X-linked (2).

The most frequently observed types of ectodermic dysplasia are the hypohidrotic-anhidrotic, and the hidrotic.

The hypohidrotic-anhidrotic type, or Christ-Siemens-Touraine syndrome was first described in 1848 by Thurman, and is characterized by the triad of hypotrichosis (skin, hair and nail anomalies), either hypodontia or anodontia, and hypohidrosis (partial or total absence of eccrine sweat glands) and other features such as frontal bossing, saddle-shaped nose, everted lips etc. (3).

The hidrotic type was first defined in 1929 by Clouston, and is distinguished by hypotrichosis, ungual dystrophy and hyperkeratosis of the palms and soles (4).

Numerous combinations of clinical alterations can present in ectodermal dysplasia, observing diverse syndromes and up to 154 different types of ectodermal dysplasias and 11 subgroups, labeled from 1 to 4 according to whether they affect the hair, teeth, nails or sweat glands (5).

Recently, a new classification for ectodermal dysplasia has been proposed, based on the alterations in the proteic molecular functions that lie behind it (6).

The diagnosis of patients with ectodermal dysplasia is based fundamentally on the clinical history (ungual dystrophy, hypotrichosis, anodontia, oligodontia, hypodontia); on a skin biopsy in cases of changes in sweating (reduction in pilosebaceous units and sweat glands); hair study showing thin, fine hair; panoramic radiography (clearly showing dental dysmorphia and agenesis); molecular genetic analysis (studying genetic mutation, genetic locus, EDA, EDAR, NEMO, etc.).

Hypodontia, oligodontia, or anodontia, are of great interest when diagnosing the dysplasia. Oligodontia is most frequently associated with dominant type hereditary factors, polygenic factors, or X-linked (7). Malformed teeth, delayed eruption and dental impaction are also very frequent in these patients.

The differential diagnosis of ectodermal dysplasias should be made against pathologies such as: congenital syphilis, familial simple anhidrosis, aplasia cutis congenita, dyskeratosis congenita, fever of unknown origin, progeroid syndromes such as Werner syndrome or Rothmund-Thompson syndrome, pachyonychia congenita, recurrent infant pneumonia, etc. (3,8,9).

The prognosis is usually good, except in cases with hypo- or anhidrosis, where mortality rates can reach 30% in first infancy, due fundamentally to respiratory infections resulting from the absence of mucosal glands in the respiratory system.

Clinical Case

A 5-year-old boy was seen in the Faculty of Odontology, referred by the Genetic Service, Central University Hospital, Asturias, presenting oligodontia, with 7 primary teeth; sparse, fine silky hair, thinly covering the scalp, thin eyebrows, narrow nose at the tip, fine dry skin on the palms and soles, folded lower eyelid, thick lower lip. Normal nails, and no genital, breast or ocular anomalies.

Family history: mother, grandmother and aunts on the mother's side present retention of primary dentition, impaction of permanent teeth (Fig. 1). Mother with excess of secondary folds on palms and forearms, generally dry skin.

Personal history: normal pregnancy and normal birth at time. Controlled vaccinations.

The intraoral examination revealed bone atrophy of the alveolar ridges, on both the maxilla and the mandibula, and numerous dental gaps. Some of the existing teeth were short, conical and pointed (Fig. 2).

Orthopantomograph: displaying numerous agenesis in the primary dentition, having less than half the normal number (oligodontia), delayed eruption, and misshapen teeth (Fig. 3).

Therefore, this is a case of recessive, X-linked hypohidrotic ectodermal dysplasia. The mutation perhaps took place in the gamete of the maternal great-grandmother, who was already 'elderly' when pregnant with her daughter (the boy's grandmother).

An acceptable solution to this complex case was sought, given the young age of the boy, and the emotional state of the mother, worried by the absence of numerous teeth in such esthetically compromised areas.

Under these conditions, the earliest possible prosthetic treatment was evaluated, on the one hand to limit the resorption of the alveolar ridges, and on the other to minimize the emotional and psychological impact created by the lack of teeth on the child's relationships (2,10).

Treatment began with morphological modifications using photopolymerizable composite material to some of the conical teeth in order to improve the esthetics and to aid retention of the prothesis.

It is usually problematic to take dental impressions on a 5-year-old; the patient is unwilling to cooperate, being, as in this case, unaware of the need to wear a dental prothesis, which apart from being uncomfortable, can initially complicate the patient's social life: games, sporting activities, etc.

Heavy silicone (for better handling) was used to take some preliminary impressions and make a personalized tray and later an alginate impression was taken under more favorable conditions.

It was considered appropriate to make a lower and upper acrylic partial prosthesis (Fig. 4) using steel retainers on some of the teeth present in the arcade. This design allows modifications to be made as and when necessary, as well as providing a reasonable esthetic result, an acceptable masticatory function on complementing the lack of dentition, at the same time as attempting to maintain the alveolar ridges free of atrophy, and allowing an almost normal social life, which is so important at this stage in a child's personal and psychological development.

The importance of oral hygiene as a fundamental aspect of dental prognosis for these patients was stressed to the parents, in the sense that they monitor brushing, provide daily fluoride drops according to body weight, and control the eating of sweets and snacks between meals as far as possible. Periodic check ups were prescribed, approximately once every six months, at which topical applications of fluoride gel were to be applied as well as making small adjustments and modifications to the prosthesis, especially the lower one (11).

At the present moment, we are following the eruption of a still unerupted tooth, as well as insisting on good hygienic measures of daily brushing and mouth rinse, topical fluoride applications every six months, periodic check ups etc. (Fig. 5).

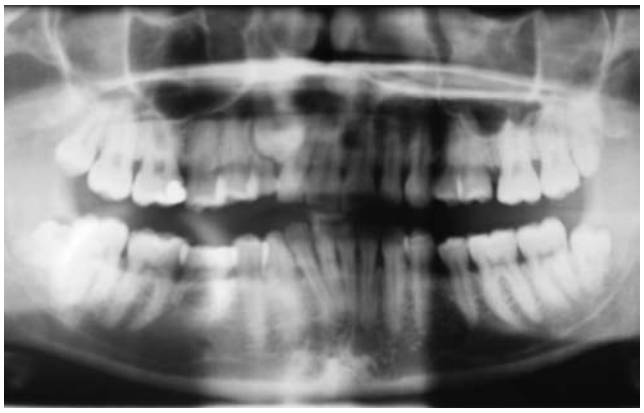

Fig. 1. Mother's orthopantomograph present retention of primary dentition and impaction of permanent teeth.

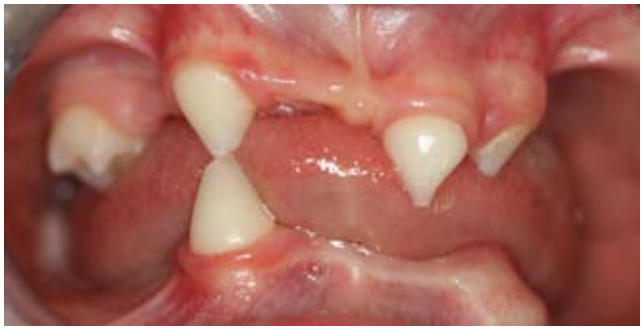

Fig. 2. The intraoral examination. Some of the existing teeth were short, conical and pointed.

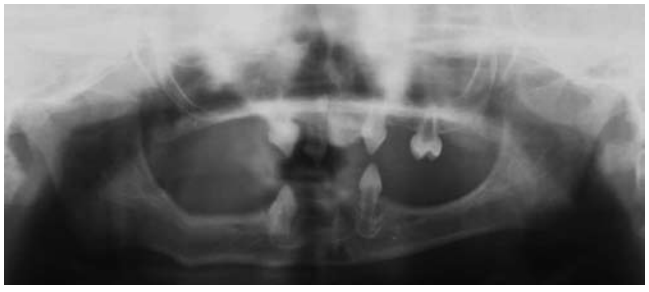

Fig. 3. Orthopantomograph: displaying numerous agenesis in the primary dentition, oligodontia, delayed eruption, and misshapen teeth.

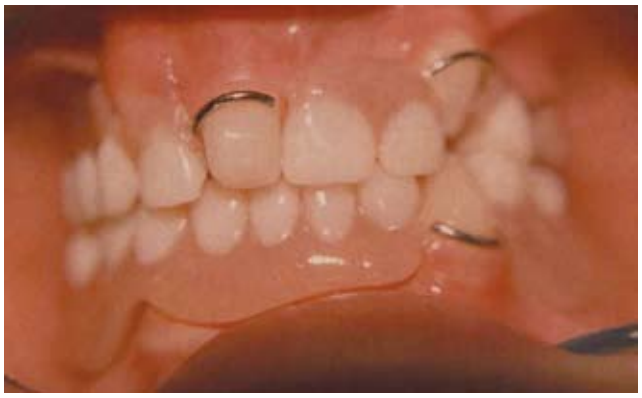

Fig. 4. A lower and upper acrylic partial prosthesis.

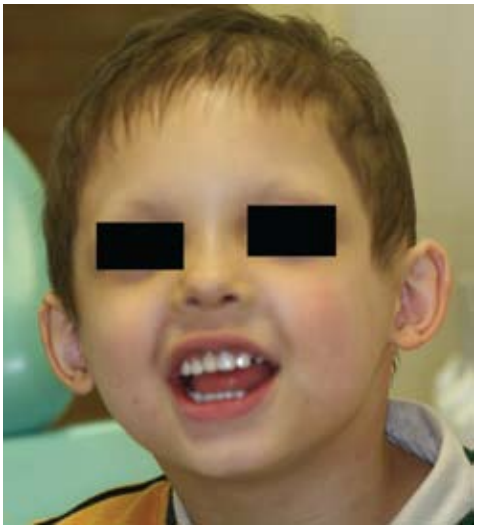

Fig. 5. Front view of the patient´s face.

Discussion

Transmission of hypohidrotic ectodermal dysplasia is, in general, X-linked (females carry the responsible gene, and males suffer from the disease, although the carrying mothers usually bear some typical characteristic feature of the disease) and at times in autosomal recessive form. Mosaic expression is rare. Currently, hypohidrotic ectodermal dysplasia is related with a mutation of the protein ectodysplasin-A, related with the EDA gene in the q12-q13 locus of the X chromosome (consisting of 12 exons, 8 of which are responsible for encoding the EDA-A1 transmembrane protein which is related with ectodermal growth). Mutations in one or various genes, including EDA, EDAR (EDA receptor) and NEMO (NF-kB essential modulator: encodes the NEMO protein, regulator of NF-kB transcription factor activity that intervenes in the control of stratified epithelial growth, allowing the cells to respond to external stimuli, etc.) are associated with

hypohidrotic ectodermal dysplasia, with or without immunodeficiency. Mutations in NEMO that suppress the protein synthesis (amorphic mutations) cause incontinentia pigmenti, however, the hypomorphic mutations in NEMO that do not eliminate its entire function cause ectodermic dysplasias and immunodeficiency in man (12-16).

Orally, the disease is characterized by hypodontia, oligodontia or anodontia, which can, moreover, affect both the maxilla and mandible; delayed eruption, malformed teeth, producing a small, pointed, conical appearance; and resorption or atrophy of the alveolar border, thus complicating the fundamental rehabilitation procedure in these patients.

Dystrophic, hypoplastic or absent nails, and sparse hair may also present. Some individuals may lack eyebrows or eyelashes, and present hyperkeratosis of the knees, elbows, knuckles and under the free border of the nails.

In general, physical development is normal, although cases of short stature, epilepsy, deafness, drumstick fingers, multiple poromas and bilateral cataracts have been observed (17).

From the point of view of the dentist, an acceptable solution is sought for these young patients with complex cases of congenital lack of teeth.

A prosthodontic rehabilitation is fundamental in these situations, attempting to provide a functional and esthetic solution that will allow the child as normal a life-style as possible, without damaging self-esteem or psychological development and ensuring that behaviour is unaffected.

The prosthetic treatment should be carried out on an individual basis, aimed always towards providing good occlusal stability.

Treatment should be commenced as soon as possible in order to avoid possible resorption and atrophy of the alveolar ridges, and to control the vertical dimension, which can be severely affected by the total or partial lack of teeth.

Different authors have proposed different rehabilitation possibilities for these patients. In general, almost all agree in recommending the use of removable prostheses during the first stages of growth (3-5 years), allowing the adjustment of the vertical dimension or maxilla / mandible interrelationship, so as to later opt where possible for provisional fixed prostheses until the patient finishes growing when a more stable and fixed situation is established and the possibility of implant treatment can be considered (4, 18-21).

From this point on different rehabilitation options can be considered, from classic, conventional solutions such as complete mucosa supported prostheses (as in the case of complete edentulism, frequent in anhidrotic-hypohidrotic ectodermal dysplasia) resin or chrome-cobalt tooth and mucosa supported prostheses, provisional fixed protheses, and the concomitant problems that sometimes occur due to the shape of the teeth and poor development of the edentulous alveolar crest; to the most up-to-date cemented-on-implant prostheses, in addition to ceramic crowns and composite reconstructions and even orthodontic treatment (with fixed multi-bracket apparatus allowing the modification of the position of the teeth, creation of spaces which can be useful in order to facilitate implant supported or mucosa and implant supported prostheses, closure of diastema, making occasional use of titanium implants or mini-screws to carry out certain movements, etc.) (22-29).

From our point of view, the use of partial acrylic prostheses is an interesting and practical alternative that provides a relatively quick, easy, acceptable and economical solution to the functional and esthetic oral rehabilitation in patients with pronounced edentulism. This solution improves the patient's quality of life and optimizes social integration; furthermore, it permits stimulation of the alveolar ridges for later treatment with an implant supported prosthesis as a more stable and esthetic solution for patients with multiple dental agenesis.

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