Ectodermal dysplasia: A review and case report
Anut Itthagartin, DDS, PDip DS*/Nigel M. King, BDS, MSc, PhD**

Abstract
Ectodermal dysplasia is a hereditary disease characterized by a congenital dysplasia of one or more ectodermal structures and their accessory appendages. Common manifestations include defective hair follicles and eyebrows, frontal bossing with prominent supraorbital ridges, nasal bridge depression, and protuberant lips. Intraorally, common findings are anodontia or hypodontia, conical teeth, and, consequently, generalized spacing. The patient may suffer from dry skin, hyperthermia, and unexplained high fever as a result of the deficiency of sweat glands. The present review focuses on the clinical manifestations, classifications, and diagnosis of ectodermal dysplasia. A 6-year-old girl, described in the case report, exhibited many of the manifestations of ectodermal dysplasia as well as behavioral problems and a severe gag reflex. The treatment to improve her appearance and oral function included a removable prosthesis, acid-etch-retained indirect resin composite veneers, and a fixed partial denture. (Quintessence Int 1997;28:595-602.)

Clinical relevance
Although the treatment of a 6-year-old girl with multiple restorative needs resulting from ectodermal dysplasia was difficult to perform, the end result not only improved her appearance but also had a positive effect on her behavior, self-esteem, and attitude toward dental treatment.

Introduction
Traditionally, ectodermal dysplasia (ED) has been the term used to denote a group of disorders characterized by a constellation of defects involving the teeth, skin, and appendicular structures, including the nails and the eccrine and sebaceous glands. Although there are alterations in the various ectodermal structures, disturbances in tissues derived from other embryologic layers are not uncommon. The ectoderm is one of the three germ layers of the embryo; it develops at, or around, the 13th day in utero, which is earlier than the mesoderm and endoderm. The ectodermal germ layer gives rise to those organs and structures that maintain contact with the outside world. They are the central nervous system; the peripheral nervous system; the sensory epithelium of the ears, the nose, and the eyes; the skin, hair, and nails; the pituitary, mammary, and sweat glands; and the enamel of the teeth.

Ectodermal dysplasia was redefined by Freire-Maia as a pathogenic developmental defect, which at the embryologic level, affects the ectoderm and therefore the tissues and structures derived from it. Thus, it affects the epidermis, in which it is responsible for the development of keratinocytes, and causes aberrations in the hair, sebaceous glands, eccrine and apocrine glands, nails, teeth, lenses and conjunctiva of the eyes, anterior pituitary gland, nipples, and the ears. In addition, there are defects of the central nervous system, the adrenal medulla, the oral, nasal, and rectal mucosa, and their associated glands; eg, in the oropharynx the defects may be manifested as a high palatal arch or even a cleft palate. The pharyngeal and laryngeal mucosa may be so atrophic that it results in dysphonia and hoarseness of the voice.
A report of this disease entity occurred as early as 1792, according to Perabo. Another early report was by Charles Darwin, who identified the complex in a closely inbred group of Indians. Since these early reports, various terms have appeared in the literature, such as congenital ectodermal defect and anhidrotic ectodermal dysplasia because of the lack of sweat gland function. Later, Felsher suggested that the adjective hypohidrotic was more appropriate to describe the reduced number of sweat glands, rather than the complete absence of sweat glands, as suggested by the term anhidrotic, which had been used previously.

Genetic studies of more than 300 cases have revealed an X-linked mode of inheritance; the gene is carried by the female but the condition is manifested by the male. However, there are reports of multiple siblings being affected and of females suffering from this condition.

Clinical manifestations

Hypohidrosis is possibly the most remarkable characteristic of ED because it may not be apparent in the first year of life but present later as a "fever of unknown origin." The inability to sweat results in an intolerance to heat, occasionally causing severe incapacitation and hyperpyrexia after only mild exertion or even following just a meal.

Because of the partial, or complete, absence of sweat and sebaceous glands, the phenotype has smooth, soft, dry, and thin skin. Fine linear wrinkles and increased pigmentation are often present around the eyes and mouth. There may be hyperkeratosis of the palms of the hands and soles of the feet. There is an absence of lanugo hair, and, although the beard is usually normal, axillary and pubic hair is generally sparse. The hair over the scalp is often short, fine, stiff, and short, while the eyebrows and eyelashes are frequently missing. The nails appear to be normal or somewhat spoon shaped. In females the mammary glands are usually aplastic or hypoplastic. Impaired lacrimal gland function and occasionally glaucoma have been reported. Other findings have included an increased susceptibility to allergic disorders such as asthma and eczema.

Other reported clinical manifestations have been variations in the shape of the skull, which can resemble an inverted triangle. The face looks smaller because of frontal bossing and depression of the nasal bridge. The lips can be protuberant and the ears may be situated obliquely on the head, causing them to stand out. The facial appearance of affected individuals is so characteristic of the condition that unrelated patients may even be mistaken for siblings.

Jaw development

The presence of teeth produces alveolar thickening but has no influence on the growth and development of the jaws, which in these patients is normal. Because the alveolar process does not develop in the absence of teeth, the vertical dimension is reduced in subjects with ED, resulting in protuberance of the lips. The palatal arch is frequently high, and a cleft palate may be present. Usually, xerostomia is not a complaint of affected subjects, because complete absence of the salivary glands is rare.

Dental abnormalities

The dental findings in ED may range from hypodontia to anodontia of the primary or permanent teeth; however, congenital absence of primary teeth is relatively rare. Many reports have emphasized the value of hypodontia in the diagnosis of ED. Frequently, the teeth that are present have conical crowns.

Diagnosis and classification

All racial groups have been afflicted by this condition. Attempts have been made in the past to develop objective diagnostic criteria based on the number and distribution of sweat pores and the amount of sweat produced. Other diagnostic criteria have been the results of dermatologlyphic analysis, characteristics of the lacrimal secretion, and the distribution and pattern of scalp hair. However, universally acceptable standards for diagnosis have not been forthcoming. Nevertheless, a good way to detect a carrier is by testing the suspected individual's sweating formation, which may reveal areas on the trunk with an absence of the ability to sweat. Missing teeth can also be an important sign.

A definitive and comprehensive classification of ED is difficult to formulate because many of the syndromes that involve ED have overlapping features. Nevertheless, a simple attempt made by Nelson included five categories: hypohidrotic (anhidrotic); hidrotic (Clouston's syndrome); EEC (ectrodactyly-ectodermal dysplasia) syndrome; Rapp-Hodgkin syndrome; and Robinson's disease.
The management of a young girl who suffered from anhidrotic ectodermal dysplasia and the effects that the operative techniques had on her appearance and psyche will be reported.

Case report

History
A 3-year-old Chinese girl was referred because of delayed eruption of her primary teeth, the conical shape of the erupted teeth, and the protuberance of her lips.

She was born after an uneventful full-term pregnancy. Soon after birth, she experienced frequent fevers. At the age of 3 months, she was hospitalized for 1 month to investigate her low body weight and short stature. No specific treatment was provided, and she subsequently developed normally. A comprehensive hematologic assessment was conducted and conditions were found to be normal.

At 18 months of age, she was again hospitalized, this time because of measles and a high fever. A skin biopsy revealed that the depigmented patches contained sweat glands, while the normal-colored skin showed a deficiency of sweat glands. She had sparse hair on her head and eyebrows and experienced mid photophobia. An oral examination revealed that only three of her 20 primary teeth were present; they were the maxillary right lateral and both central incisors, all of which were markedly tapered. The radiographic examination showed that the maxillary left lateral incisor and first molar and all of the mandibular anterior teeth were missing. Consequently, she was diagnosed as suffering from hypohidrotic ectodermal dysplasia.

Initially, only a prophylaxis could be performed because of major management problems and a severe gag reflex. Consequently, arrangements were made to review her regularly every 4 months to monitor her general growth and development and to observe the development of her dentition. It was proposed that when she could cooperate sufficiently to receive treatment, or when she became concerned about her appearance, a restorative phase of treatment would be implemented.

She fell and traumatized her oral structures at the age of 6 years. There were no extraoral injuries, but the maxillary right central incisor exhibited mobility of about 1.5 mm in all directions and the gingivae around this tooth showed a bluish red discoloration. Radiographically, there was evidence of some root resorption and periodontal thickening associated with this tooth. A “fishing line splint,” made of 23-kg breaking strain monofilament fishing line and resin composite, was fitted from canine to canine in the maxilla. This splint was left in place for a period of 4 weeks, until all of the signs and symptoms exhibited by the traumatized maxillary right central incisor had resolved. Shortly after removal of the splint, the patient and her mother expressed concern about her appearance.

Clinical examination

The extraoral examination showed a mildly concave soft tissue profile with protuberant lips (Fig 1). Introraorally, her soft tissues were normal, and xerostomia was not evident. All of the maxillary anterior teeth that were present had tapered or even conical crowns (teeth C, D, E, F, and H [53, 52, 51, 61, and 63]); the maxillary left lateral incisor and first molar were missing. In the mandible, all of the anterior teeth were missing. All of her permanent first molars were in the process of erupting. Her oral hygiene was satisfactory, and she was caries free (Figs 2a to 2c).

Radiographic examination

A panoramic radiograph revealed the absence of numerous permanent teeth. Those present were the maxillary second premolars and left lateral incisor; the mandibular first premolars, and all of the first and second molars. In addition, the primary maxillary left central incisor had a shorter root than did the other anterior teeth (Fig 3).

Restorative procedures

The primary concern in the treatment of this individual was her facial esthetics. After the treatment plan was clearly explained to the girl and her mother, it was necessary to take primary diagnostic impressions to prepare casts for a diagnostic setup. These impressions were extremely difficult to take because of her uncooperative manner and severe gag reflex. The impression material Optosil (Bayer) was used for the impressions because the patient could not tolerate the smell or the taste of alginate. Furthermore, it was only possible to obtain an impression of the buccal and occlusal surfaces of the teeth.

In collaboration with the dental technician, preliminary “waxups” were made to simulate the effects of a mandibular partial removable prosthesis and resin composite restorations to recontour the maxillary anterior teeth. This was performed so that the patient...
Itthagarun/King

Fig 1 (left) The profile of the patient before treatment shows a mild concavity of the face and protuberance of the lips.

Fig 2a (below) All the maxillary anterior teeth are tapered or conical.

Fig 2b The maxillary left lateral incisor and first molar are missing.

Fig 2c All of the mandibular anterior teeth are missing.

Fig 3 The panoramic radiograph reveals the absence of numerous permanent teeth: the first premolars, canines, central incisors, and right lateral incisor in the maxillary arch; and both second premolars and all of the anterior teeth in the mandibular arch.
and her mother could visualize the possible treatment options. The child expressed a strong desire to have a mandibular prosthesis. Therefore, an impression was taken of the mandibular arch using the polyether bite registration material Ramitec (ESPE). The patient cried and vomited while the impression was being taken.

At a subsequent appointment, a mandibular removable partial denture made of heat-cured acrylic resin with C-clasps on the mandibular second molars was fitted (Fig 4). Oral and written instructions about oral hygiene techniques, which emphasized the need for removal of the prosthesis for tissue recuperation, were given to the patient and her mother. In addition, instructions were given about the placement of topical fluoride drops on the fitting surface of the prosthesis. The girl was extremely happy with the prosthesis.

After a period of 1 month, the patient expressed a desire for restoration of her maxillary teeth. The study cast was surveyed to identify the regions of the teeth that were undercut and would affect the path of insertion and so require reduction for the placement of veneers and a fixed partial denture. Minimal tooth reduction was planned because of the low level of patient cooperation. Subsequently, minimal reduction of her maxillary teeth was carried out for the placement of a three-unit laboratory-cured resin composite fixed prosthesis to span from the right central incisor to the left lateral incisor with a pontic to replace one incisor. In addition, the right canine and lateral incisor and the left canine were prepared to receive three individual indirect composite veneers: all of the margins were finished to a feather edge. During the preparation phase, the patient was cooperative but reticent. A special tray without palatal extension had been constructed from clear acrylic resin by means of a vacuum-molding procedure. This tray, loaded with Ramitec, was used to take an impression of the maxillary teeth.

The indirect composite veneers and fixed partial denture were fabricated in the laboratory using a vacuum-cured resin composite system, Dentacolor (Kulzer).

After trial fitting, the veneers and fixed partial denture were bonded to the teeth with Durafill (Kulzer), a photocured microfilled composite material (Fig 5). Minor occlusal adjustments were made to the mandibular prosthesis rather than to the veneers, because the patient’s gag reflex limited the usage of rotary instruments in her oral cavity. The palatal margins of the veneers were finished to a feather edge. Both the girl and her mother were extremely pleased with the fixed maxillary prosthesis and the veneers (Figs 6a and 6b).

Regular recalls were scheduled 1, 2, 4, and 16 weeks postinsertion, to make any adjustments and to monitor the oral hygiene. As the child grows and the succedaneous teeth begin to erupt, the prosthesis will have to be modified and later remade to accommodate growth changes, the exfoliation of her primary teeth, and the eruption of her permanent teeth.

Discussion

In the present case, the proband was the only child of the parents. No similar cases of ED have been identified among the relatives, which suggests that the propositus was probably a fresh mutation.
Some patients who have presented with anodontia or hypodontia have been treated with different types of prostheses. Grinberg and coworkers delivered a complete maxillary and a partial mandibular prosthesis with expansion springs to a 4-year-old child with ectodermal dysplasia who had only one primary mandibular canine. The intention was to stimulate growth of the jaws and to enhance the child’s appearance. Another patient who was treated by the placement of a complete prosthesis was subjected to cephalometric analysis to confirm improvement in the esthetics.

Various other types of prostheses have been used, including overdentures, which provide greater retention and stability than complete dentures and have the added advantage of not requiring extensive flanges, and gold thimbles with Seko attachments. Orthodontic treatment has been used in combination with the fitting of a partial denture. For these procedures, a high level of patient cooperation is required, which was not demonstrated by the patient in this report. In adult patients, osseointegrated implants have also been used and reported to function satisfactorily. However, when the use of osseointegrated implants is planned, dental and skeleton maturation, not the chronologic age of the patient, must be taken into consideration to prevent future infraocclusion of the prosthesis. Furthermore, it is important that there be enough space in the mesiodistal direction for the fixtures to avoid the risk of marginal bone loss around the adjacent teeth.

In this particular child, because of her severe management problems, there appeared to be only one treatment option for the mandibular arch, namely a heat-cured acrylic resin partial denture. The prosthesis was not a tight fit, a feature that did not bother the girl; better retention could have been achieved by placing resin composite on the teeth. This would have provided more undercut for the clasps.

For the maxillary arch, several options were considered, such as the use of resin composite to reduce the space between the teeth by enlarging the anterior teeth. The second option to improve the appearance was the use of an overdenture that utilized the retention from the erupted teeth and the palate. The third option was a three-unit, indirect, laboratory-processed resin composite fixed partial denture and three separate indirect resin composite veneers. All of the options were clearly explained and discussed with the patient and her parents. The first option was rejected because the patient could not be relied upon to sit still in the chair and she could not tolerate the use of the rotary instrumentations necessary to prepare and fit direct composite veneers. Because her severe gag reflex precluded the taking of an impression of the palatal region, it was considered doubtful that she
would be able to tolerate a fully extended maxillary removable prosthesis; therefore, the second option could not be used. Hence, it was decided to adopt the third treatment option.

The margins of the preparations for the composite veneers and the composite fixed prosthesis were finished to feather edges that were located just above the gingival margin, thus avoiding the need for local anesthesia and minimizing the chance of traumatizing the soft tissues. When the patient returned for her recall visits, there was no gingival inflammation, and the margins of the veneers were intact and not discolored.

The main objectives of fitting prostheses in patients with ED are to improve the esthetics, phonetics, and masticatory function. Prostheses may also improve the tone of the muscles of mastication and compensate for the reduced vertical dimension, which can predispose the patient to angular cheilitis.

It is essential to prepare a long-term treatment plan that includes regular reviews to ensure that the patient with ED maintains an adequate level of oral health care. In a patient described by Album, there was a serious breakdown of tooth structure under an overdenture because of a lack of follow-up and inadequate oral hygiene and home-care procedures. This highlights the need for the careful consideration of suitable treatment options for individual patients.

Laboratory-processed resin composite veneers have been shown to be superior to resins cured in the mouth. The veneers that were fitted for this patient can be expected to last until the child has matured further. In the permanent dentition, porcelain veneers and a ceramometal acid-etch-retained prosthesis may be considered.

In this particular case, the patient's increasing age made her social development and social interaction increasingly important. It has been reported that children rejected by their peer groups are more likely to become aggressive and delinquent. In addition, they are more likely to remain unaccepted by their peers and even to experience mental health problems in adulthood. Therefore, the successful treatment of this girl can be expected to help her both physically and psychologically. The fixed and removable prostheses proved to be very popular with the child, and, according to her mother, she was able to enjoy a better-quality diet because of her new ability to chew food with her anterior teeth. Moreover, the girl's attitude, self-confidence, and peer group interaction showed signs of significant improvement.

The successful use of any prosthesis depends on the cooperation and communication between members of the dental team and the patient. It is also important for the technician to share in the planning and to see the outcome of his or her contribution to the treatment process. The positive response of the dental team to this girl and her treatment led her to appreciate and trust all of the members of the dental team.

Conclusion

This case demonstrated the psychological problems that can be experienced by a child with ED. In addition, it highlighted the importance of formulating a customized treatment plan suited to the individual. Although the treatment plan could have been considered a compromise because of the patient's limited level of cooperation, it was rewarding to see the improvements that occurred in her social behavior as a consequence of the dental treatment.

References