Rhinology, 20, 231-235, 1982

The nose in anhidrotic ectodermal dysplasia

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SUMMARY

A case is presented of anhidrotic ectodermal dysplasia, of extreme rarity. This is distinghuished by disorders in the structures of ectodermal origin. The clinical characteristics – hypohidrosis, hypodontia, hypotrichosis –, and the genetic features – genealogy, karyotipe – are examined first. The patient's face appears to be small, due to the combination of frontal bossing, hypodontia, and a depressed nasal dorsum, somewhat resembling the saddle nose of congenital syphilis.

The case is studied from various perspectives: clinical, radiological, anatomicopathological and ultrastructural. The study concentrates principally on the anomalies of the nasal cavities, where there is a notable atrophy of the nasal mucosa. This atrophic rhinitis displays all the features of ozaena: severe crusting, fetid green secretion, nasal obstruction, etc.

In 1929 Weech reported a hereditary disease which affects the structures of ectodermal origin. In view of the important decrease in the number of eccrine sweat glands, he proposed for this complaint the name of Anhidrotic Ectodermal Dysplasia. This syndrome, which is very infrequent, is characterized by the triad of hypodontia, hypotrichosis and hypohidrosis.

It is transmitted in a recessive way linked to the chromosome X, the gene being carried by the mother and displayed in the males. Females may present a minor syndrome with moderate hypodontia and/or conical teeth.

In our case, we examine principally the structure of the face, and the structural, functional, histopathologic and ultrastructural pecularities of the nasal cavities.

CASE REPORT

D.L.A., male, was first examined by us at the age of three years and one month. His mother, aged thirty-seven, displays no evidence of the disease. There is no possibility of collecting data about the maternal family.

The patient was delivered in a normal childbirth, but problems immediately be-

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Paper presented at the 9th Congress of the European Rhinologic Society and 3rd ISIAN, Stockholm (Sweden), September, 1982.

gan with the appearance of an intense dryness of the skin and violent outbreaks of hyperpyrexia. Already during lactation nasal stuffiness was notable. After the age of two, crusting appeared in both nasal cavities, difficult to extract, of considerable discomfort for the child and producing intense fetidness. Epistaxis was frequent and abundant. When the boy reached the age of three, the absence of normal dentition began to cause the family some concern. Only two teeth with conical crowns appeared in the upper group, and none in the lower jaw.

The facies is as described for this syndrome by Gorlin, Pindborg and Cohen (1976). The disproportionate combination of frontal bossing and a sunken dorsum and root of the nose gives the observer the impression that the face is small. The distance from the chin to the inferior nasal spine is very reduced, especially in relation to the distance between the inferior nasal spine and the forehead; this is due to the contact between the maxilla and the mandible, which is direct because of the virtual lack of teeth. The osseous external nose is clearly sunken and broad.

With anterior rhinoscopy the nasal cavities can be seen to be greatly expanded, fundamentally at the expense of a sideways extension of their lateral walls and a extraordinary atrophy of the turbinates. The inferior turbinate is totally diminished, and barely protrudes in the nasal cavity; the middle turbinate is almost unnoticeable. Prior to the nasal cavity examination, an intensely malodorous scab measuring 4×2 cm was extracted from each cavity.

The skin is dry and thin, of seemingly brittle and papery texture. Very fine wrinkles show up in various parts of the face, becoming obvious around the mouth. The hair is sparse, stiff, very fine. The eyebrows are almost totally missing and the eyelashes are sparse, thin and short.

A Roentgen Ray diagnosis is essential to determine exactly whether a genuine hypo- or anodontia is being dealt with and not a pseudoanodontia or a case of retarded dentition. Metson and Williams (1952) and Gorlin, Pindborg and Cohen (1976) draw our attention tot the deficiency of the alveolar process due to the inexistence of teeth. The absence of tooth germs is a fundamental fact, for it precludes any possibility of retarded eruption.

The orthopantomograph shows the two conical teeth already in existence in the upper dental group and four tooth germs, which are the maximum possibility of definitive dentition.

In the anteroposterior polytomographs we find a big increase in the transversal diameter of the nasal fossae at the expense of the lateralization of the external wall; in none of the shots can the middle or inferior turbinates be seen.

Histopathologic studies of the nasal mucosa show that the ciliated epithelium has suffered an epidermoid metaplasia to squamous epithelium, and that the ciliated cells typical of respiratory mucosa have almost completely disappeared. We car-

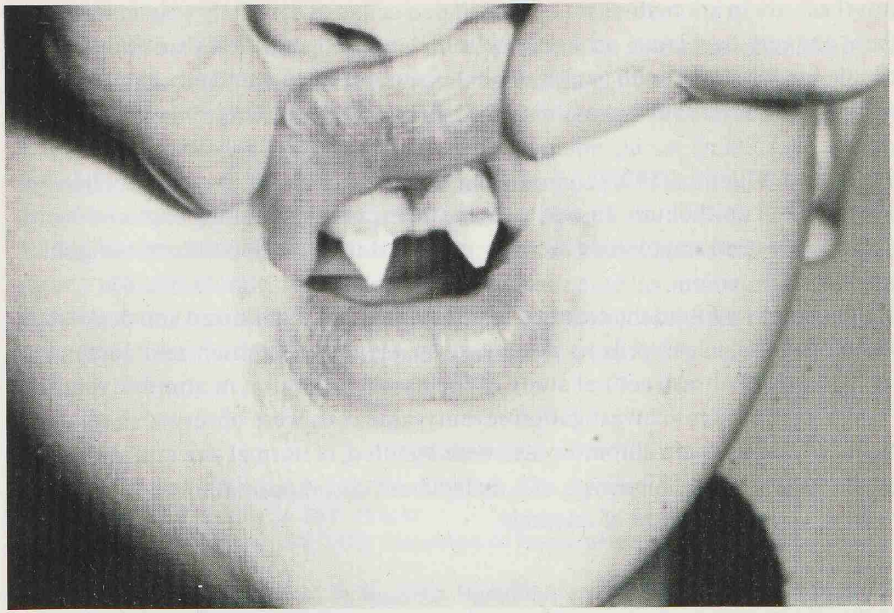

Figure 1. Only two teeth with conical crowns appeared in the upper group, and none in the lower jaw.

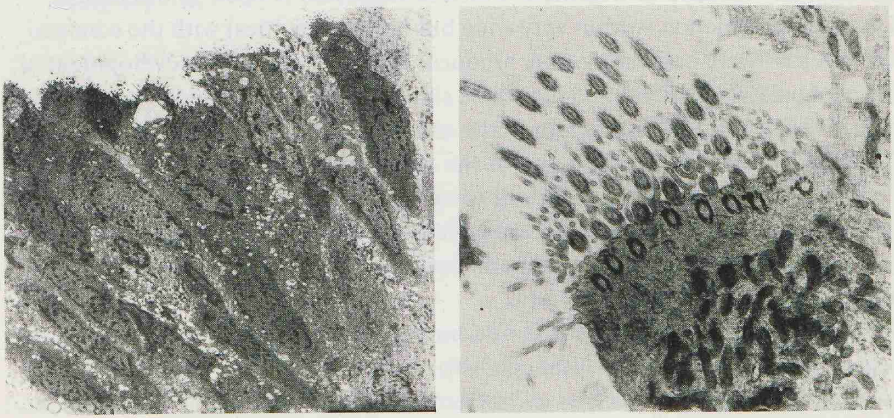

Figure 2. The ciliated cells are definitely scarce; however there are very numerous columnar cells in a pseudo-stratified situation.

ried out tincturing with PAS, studying the corium beneath the epithelium, and observing only very sparse glandular formations.

By transmission electron microscopic studies we located the cells typical of pseudo-stratified respiratory epithelium: ciliated cells, goblet cells, basal cells and "brush" cells.

But these are in anomalous ratio: the ciliated cells are definitely scarse; however there are very numerous columnar cells, in a pseudo-stratified situation, devoid of cilia but with microvilli as exterior extensions of their cytoplasm, these microvilli being shorter and more abundant than usual interciliary microvilli.

Melon and Willemot (1972) counted four of five ciliated cells per goblet cell in the normal nasal epithelium. In our case, in the numerous sections explored by us with the electron microscope we only observed the occasional scattered goblet cell.

In no section we found the external stratum corneum, keratinized and deep twelve layers of anuclear cells as described by Mygind, Thomsen and Jorgensen (1974) in their ultrastructural study of the nasal epithelium in atrophic rhinitis.

For the chromosomic investigation seventy-nine cells were observed in mitosis: in all of them forty-six chromosomes were counted, of normal size and shape; we are dealing with the karyotype of a male whose chromosomal formula is 44XY, with no observable type of anomaly.

THERAPEUTIC COMMENTS

In this final section, dealing with possible courses of therapeutic action, the absence of practical and bibliographical experience must be pointed out. Consequently, any method of treatment which is suggested is open to discussion.

The nasal obstruction (within very wide but blocked cavities) with the concomitant alternative of oral ventilation, produces disturbances in the development of the hard palate, upper jaw and the whole sinus system. This is made worse by the defective mastication resulting from the anodontia, and which affects in turn the development of the mandible and of the whole face.

Palliative therapy should direct itself towards an extreme care of the nasal cavities: meticulous extraction of the scabs, frequent washing and pumping, vitaminotherapy, advising the patient to spend periods in geographical zones with a climate of high relative humidity, etc.

When the growth of the skull is considered complete, surgical treatment of the ozaena could be considered although with reservations. Surgery could be by implantation of sustances such as chips from the iliac crest, deproteinated bovine chips, dolomite or silicone, with techniques derived from that of Eyries; or by moving the external wall of the nasal cavities.

Following our advice, the patient was fittid with a maximallary appliance at a premature stage. The boy began to use it at the age of four, his splendid acceptance of the orthoprosthetic appliance being a pleasant surprise to us. In the three years since then, this appliance has had to be replaced by another one adapted to the natural growth of the jaws.

RESUMEN

Se presenta un caso, muy poco frecuente, de displasia ectodérmica anhidrotica. Se tipifica por transtornos en las estructuras de origen ectodérmico. Sus caracteristicas clinicas (hipohidrosis, hipodoncia, hipotricosis) y sus connotaciones genéticas, son revisadas previamente. La combinación de un frontal abombado, hipodoncia y una depressión del dorso nasal parecida a la de la sífilis congénita, hacen que la cara del paciente parezca muy pequeña.

El caso es estudiado desde puntos de vista: clinicos, radiológicos, anatomopatológicos y ultrastructurales. Consideramos principalmente las anomalias de las fosas nasales, en las que existe una notable atrofia de la mucosa. Esta rinitis atrófica muestra las peculiaridades del ocena: costras, secrecion fétida, obstrucción nasal, etc.

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