CASE REPORT

Rhinology, 43, 233-235, 2005

Atrophic rhinitis in a patient with anhidrotic ectodermal dysplasia*

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SUMMARY

We would like to present the rare case of a now 37-year old female patient with autosomal-recessively inherited anhidrotic ectodermal dysplasia being treated in our ENT department for atrophic rhinitis. The clinical appearance very much resembled the picture of an "empty nose" with distinct hypoplasia of the turbinates and extensively wide nasal cavities. We want to point out the possible existence of atrophic rhinitis against the background of an underlying syndromatic disease in adults and also the pediatric patient.

Key words: atrophic rhinitis, saddle nose deformity, empty nose, ectodermal dysplasia, autosomal-recessive inheritance

INTRODUCTION

Ectodermal dysplasias (ED) represent a large and very heterogeneous group of disorders with defects of ectodermal structures [1]. Practically, various clinical expressions had been described [2]. According to McKusick [3], the incidence is estimated to be seven in 10.000 births. Lamartine [4] stated that the inheritance is in conformity with Mendelian modes although sporadic cases are possible as well. Several types of ED had been explored on the molecular level identifying the causative genes. In cases of the X-linked recessive form of hypohidrotic ED (type Christ-Siemens-Touraine Syndrome), mostly males are severely affected whereas female carriers might only suffer from minor symptoms. The responsible gene had been mapped to Xq12-q13 causing mutations of the ectodysplasin anhidrotic receptor. Autosomal recessive inheritance is also possible for hypohidrotic ED. Then, both sexes might be equally affected. The gene map locus in these cases is 2q11-q13 causing mutations of the ectodysplasin-A transmembrane protein [5]. Patients suffering from ED mostly present more or less distinctive features entailed by absent or reduced skin appendages and defective dentition [6]. Frontal bossing, periorbital wrinkles, prominent lips, disturbed dentition, hypo- or anhydrosis as well as partial or total alopecia may be obvious. The otorhinolaryngologist comes into play for the treatment of nasal mucosal atrophy, recurrent nose bleeding, saddle nose deformity, recurrent pharyngeal or laryngeal infection due to mucosal dessication. In particular the existence of atrophic rhinitis (AR), also referred to as ozena, producing a more or less intense fetid nasal odour, may be a debilitating factor reducing the patients` quality of life. Huizing et al. [7] already reported in detail about ozena as part in syndromes. We would like to present the rare case of a now 37-year old female patient with anhidrotic ED being treated in our ENT department for AR.

CASE REPORT

After cytogenetic exploration and thorough examinations by the pediatric department, University of Ulm, Germany, a then 15-year old female patient was diagnosed with autosomal recessive anhydrotic ED. Cytogenetic workup revealed the autosomal-recessive type of anhidrotic ED. No X;9 translocation could be found. The chromosome set presented no aberrance. Her three years older brother also had been diagnosed with the autosomal recessive form of anhidrotic ED, presenting almost identical characteristics. Both parents did not show any characteristics typical in anhidrotic ED, any consanguinity between them was not known.

She had then been presented in the Department of Otorhinolaryngology, University of Ulm, Germany, due to intense fetid nasal odour. The examination revealed a saddle nose deformity from the outer aspect. Endonasally, signs of chronic inflammation and hypoplasia of the nasal conchae were obvious. Conservative measures including saline solution inhalations had been recommended. Other main features of the patient at that age had been fine, brittle hair, partial alopecia, dysodontia provided with dental prosthesis, absence of proper eccrine sweat gland function, dry mucosa within the upper airways, recurrent hoarseness of the voice, frontal bossing, a depressed nasal bridge, prominent, bulging lips as well as absent mammary glands. At the age of 17 a rhinoplasty without any endonasal interventions had been performed due to increasing unhappiness with the outer aspect of her nose. The turbinates had not been surgically treated.

The patient had last been examined in our department at the age of 37 (Figures 1 and 2) then. Her major complaints had been feeling of a dry nose with crusting and awkward nasal odour. Our findings revealed that both nasal cavities were

*Received for publication: March 5, 2004; accepted: November 11, 2004

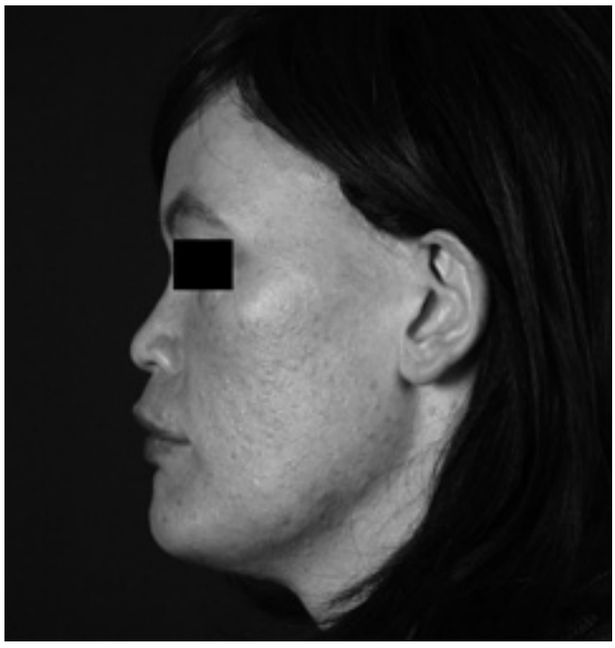

Figure 1. Outer appearances with typical features like frontal bossing and saddle nose deformity in lateral view. Due to alopecia totalis, the patient is wearing a wig.

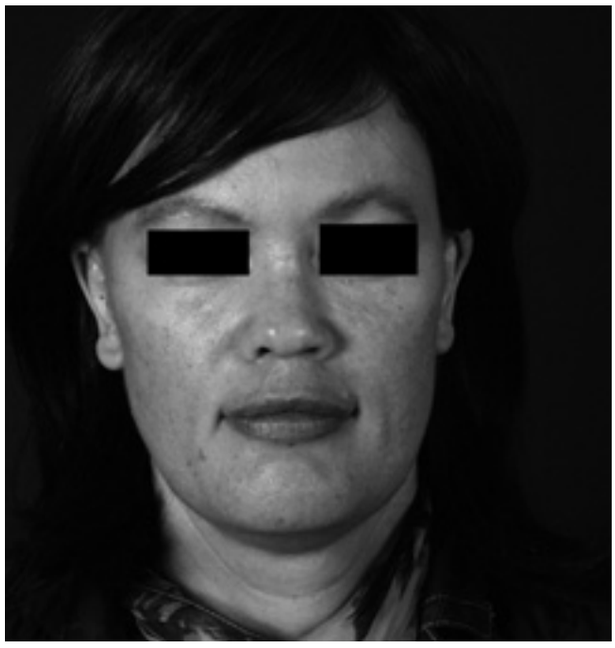

Figure 2. Outer appearances with typical features in frontal view.

extremely wide with distinctive hypoplasia of the nasal turbinates, almost presenting the appearance of an empty nose after radical turbinate surgery contrary to the fact that the turbinates had remained untouched. Nasal swab showed no colonisation with *Klebsiella ozena*. Physiological bacterial flora as well as *Enterobacter* species were present. Regarding therapy, intensive nasal hygiene involving saline solution inhalations and application of dexpanthenol nasal spray were recommended. Furthermore, antibiotic therapy with oral application of ciprofloxacin for three weeks and topical employment of mupirocin was started. After six months subjective complaints, especially nasal odour had not improved. Computer tomography of the paranasal sinuses revealed an almost empty nose with pronounced hypoplasia of the turbinates and chronic maxillary sinusitis (Figures 3 and 4). Consequently, endonasal sinus surgery and reduction of the nasal cavity volume by submucosal implantation of allogenic rib cartilage was performed. We followed the surgical technique of narrowing the nasal cavity described by Huizing et al. [8]. Hemitransfixion incision was done, as the common endonasal access for septal surgery. Perichondrial flaps were created on either side of the septum, especially along the bottom of the nasal cavity. Allogenic rib cartilage was then cut into little slices to be placed between nasal mucosa and septal cartilage. By doing so, narrowing of the large nasal cavity could be achieved in order to recreate close to physiological conditions for nasal airflow. Histology revealed one typical feature existent in atrophic rhinits: the absence of mucinous glands and cilia, as atrophic rhinitis is practically defined by the lack of characteristic traits of healthy mucosa [9].

Since then, the patient's complaints had clearly improved. Nasal hygiene involving regular intranasal irrigation is continued. The last examination in our department was performed one year after surgery, after an overall course of 22 years of treatment. Our patient then reported about noticeable fewer problems with odour and the feeling of a dry nose due to thorough nasal hygiene involving lavage and inhalation with saline solution and occasional application of dexpanthenol ointment.

DISCUSSION

Ectodermal dysplasias (ED) constitute a large and very heterogeneous group of disorders mostly involving defects of the sebaceous, submucous and eccrine sweat glands. The otorhinolaryngologist`s attention is primarily drawn to pathological changes including the nose, pharynx and larynx. According to Ibanes-Carcamo [10], particularly the absence or hypoplasia of the nasal mucous glands may lead to atrophic rhinitis (AR) with its inevitable, often intolerable odour. Turbinate hypoplasia may be so distinctive that one might get the impression of an empty nose as present in patients after extensive turbinate surgery. Concerning AR, its etiology is still not yet completely understood. Generally, it is mainly differentiated between primary and secondary AR, which is referred to as a result of trauma, surgery, particularly extensive turbinate resection, granulomatous diseases, infection or radiation exposure. In the presented case no prior turbinate surgery had been performed. The problem seemed to lie within the disease of ED itself showing extreme hypoplasia of the turbinates. On the other hand, Moore et al. [9] find that the origin of primary AR still remains unknown. In respect of the treatment of AR two main approaches, conservative or surgical measures, exist. Though,

surgery should not be the therapy of choice, but only be taken into consideration after the failure of conservative measures. Intensive care of the nasal mucosa is one priority. Systemic application of antibiotics seems to specifically improve the nuisance of fetid nasal odour. Nielsen et al. [11] reported about oral therapy with ciprofloxacin for one to three months which seemed to make odour, crusting and growth of Klebsiella ozenae disappear. When it comes to surgery, different procedures might be applied. Huizing [12] reported about their various experiences with surgical treatment of ozena. El-Kholy et al. [13] described a septal mucoperichondrial flap for closure of the nostril as an easy technique to improve typical symptoms of AR. Fang et al. [14] applied endoscopic sinus surgery in patients with AR due to sinus infections with appropriate results in carefully selected candidates. Even closure of the nostril is reported by Gadre et al. [15]. The surgical approach applied in our patient after unsuccessful conservative measures was to scale down the size of the vast nasal cavity volume by implanting chips of allogenic rib cartilage submucosally [9]. Thereby, endonasal mucosal surface is restored and enhanced enabling better function of nasal mucosa again, particularly when it comes to heating and humidifying air influx.

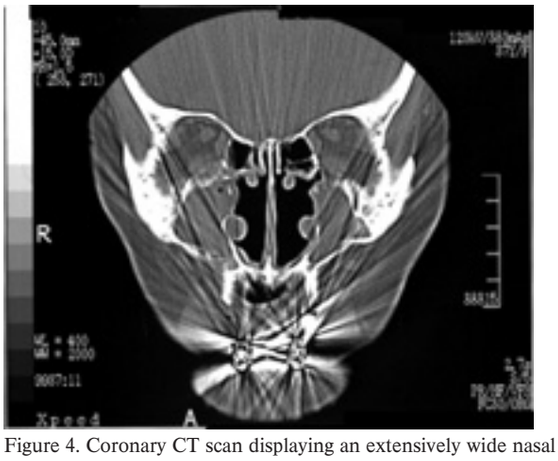

Figure 4. Coronary CT scan displaying an extensively wide nasal cavity with complete hypoplasia of the inferior and middle turbinates.

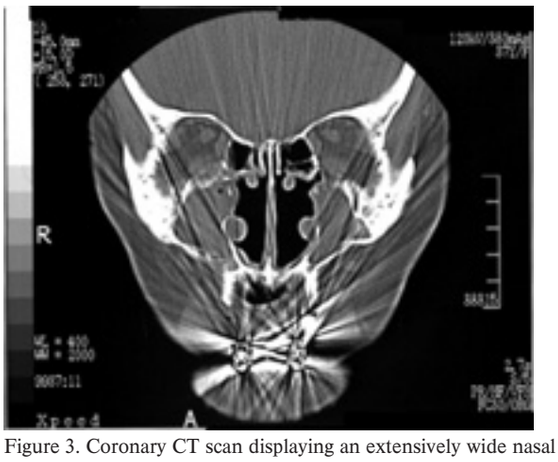

Figure 3. Coronary CT scan displaying an extensively wide nasal cavity with complete hypoplasia of the inferior and middle turbinates even though turbinate surgery had never been performed. Additionally, signs of chronic maxillary sinusitis are present.

CONCLUSIONS

The aim of the presentation of this case of anhidrotic ER is the allusion to the fact that AR does not have to be of primary unknown origin or secondary to endonasal surgery or radiation therapy. It may also be one symptom of syndromatic diseases, like the very heterogenous group of ERs. In the presented case the main problem was extreme hypoplasia of the inferior and middle turbinates without precedent turbinate surgery, creating a similarly wide open nasal cavity as the empty nose after aggressive turbinate resection. Whenever confronted with AR in addition to particular and obvious syndromatic features, a more thorough examination is inalienable, especially in the pediatric patient.

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