Case Report

New Indian Journal of Surgery
Volume 7 Number 2, May - August 2016
DOI: https://dx.doi.org/10.21088/nijs.0976.4747.7216.23

Ectodermal Dysplasia with Spontaneous Palatal Perforation: Case Report of a Rare Occurance

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Abstract

Background: Ectodermal Dysplasia is a rare disorder. The otolaryngological manifestations range from chronic upper respiratory tract infection to more severe infections of the para nasal sinuses including nasal myiasis. *Purpose:* The present report describes the rare occurrence of palatal perforation in a case of ectodermal dysplasia, leading to difficulty in feeding and regurgitation. *Conclusion:* Early detection and addressal of otolaryngologicalproblems associated with ectodermal dysplasia is suggested, so that complications like palatal perforations do not occur.

Keywords: Ectodermal Dysplasia; Otolaryngological Manifestations; Nasal Myiasis Palatal Perforation.

Introduction

First described by Thurman in 1843 [1], ectodermal dysplasia embraces a long series of abnormalities whose common denominator is a morphological alteration of ectoderm derived organs and tissues [2]. The main manifestations include defects in hair, teeth, nail and sweat glands. The features are very well described in the literature and remain consistently similar, characterized by absent or reduced sweating, hypotrichosis, dysodontia with episodes of hypothermia. The otolaryngeal manifestations include otitis, chronic upper respiratory tract infections, dysphagia, bronchitis [3]. There may be sometimes severe infections of the nose and paranasal sinuses secondary to stasis and crust formation. Unusual features include atrophic rhinitis aural or nasal myiasis, which may lead to destruction of nose, face and intracranial structures, sometimes causing meningitis and even death [4]. Here we report the occurance of palatal perforation in one of the patients of ectodermal dysplasia with history of nasal myiasis.

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Case Report

Our patient was an 8 years male child who presented with complains of difficulty in feeding with regurgitation and appearance of a defect in the oral cavity for last 7 days. The child was a product of non co-sanguineous marriage and was born of a normal delivery. His psychomotor development was normal. In infancy he had sparse and fine scalp hairs with abnormal nails. He had abnormal temporary and permanent dentition and also gave history of heat intolerance and hypohidrosis from early childhood. He had no history of any seizure disorders or any other significant illness. The child gave history of increasing itching in the nasal cavity with appearance of foul smelling discharge from the nose 7 days back. The parents never sought any medical attention for the same. After 1 day he had bouts of sneezing with passage of few maggots and noted appearance defect in the palatal region with passage of few more maggots. Since then he had history of discharge of liquids from the nose while eating and difficulty in eating. Physical examination revealed abnormally widened nose with depressed nasal bridge. There were loss of eyebrow hairs and eyelashes. The scalp hairs and the body hairs were markedly reduced. The skin felt dry and soft with periorbital pigmentation. Oral cavity examination revealed absence of multiple teeth and only two malformed (conical) teeth existing. Palatal defect seen at the

junction of soft and hard palate with infected ragged margins. Examination of the nasal cavity revealed mucopurulent discharge with slough and crusts with foul smelling odour and presence of a dead maggot in the nasal cavity. The palms, digits, soles and the genitalia were normal. Other systemic examination was also normal. The patient was admitted and Intravenous antibiotics were started. Laboratory examination revealed normal serum biochemistry. Histopathological examination of the skin was consistent with ectodermal dysplasia. The patient was put on oral and nasal saline washes with high oral feeding to avoid regurgitation. He was subsequently discharged with the same advise for a later palatal repair.

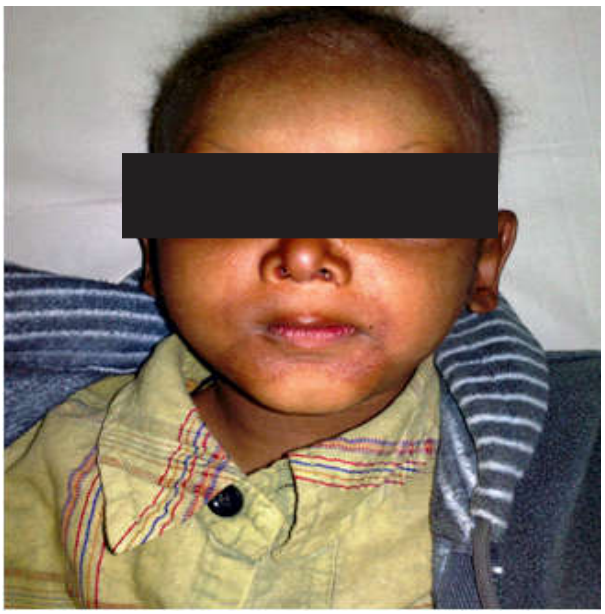

Fig. 1: Typical facies of the patient with blond and scanty hairs. Eye lashes and eyebrows are reduced in number. There is depressed nasal bridge with xerostomia and periorbital pigmentation

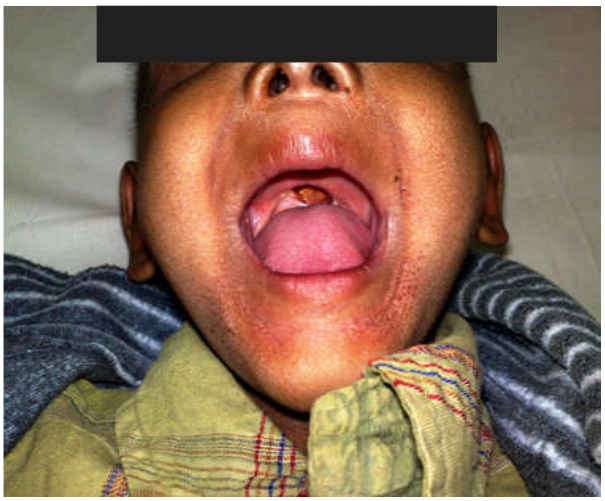

Fig. 2: The intra-oral photo showing the palatal defect with crusts in the nasal cavity and xerostomia.

Discussion

Ectodermal dysplasia is a rare condition with an estimated incidence of 7 in 10,000 live births [5]. The ectodermal dysplasias are heterogeneous heritable developmental disorders with manifestations and defects affecting other organs and tissues6. Structures most commonly involved are the ecccrine glands resulting [6] in hypohydrosis or anhidrosis, the hair (wispy), the teeth (fewer in number and conical in shape), and the nails [7]. The facial features of the individuals suffering from the ectodermal dysplasia are consistently similar. They usually exhibit fine blond and scanty hair hairs. Eye lashes and eyebrows are reduced in number or absent. The skin is fine and smooth with deficient sweat and sebaceous glands andeczematoid periorbital pigmentation can be seen [8]. The nasal bridge is usually depressed with resultant saddle nose configuration. Also found in ectodermal dysplasia patients is hypoplasia or absence of the mucous glands linning the upper aerodigestive tract. It results in chronic upper respiratory tract infections, otitis, dysphagia, hoarseness, and bronchitis and some to me shaemoptysis [9]. The tenacious consistency of nasal secretions promotes mucous stasis with secondary infections of the nose and the paranasal sinuses. Nasal myiasis has been described in these patients with increased prevalence in tropical countries with atrophic rhinitis is the most common predisposing factor. If remain uncontrolled these maggots may cause destruction of adjoing structures as palate in the present case. There may be associated decreased mucosal sensitivity of these regions secondary to ED, leading to almost painless destruction of the palate as in this case. An increasing awareness about this disease is needed to prevent these complications like this to occur.

Because manifestation of the ectodermal dysplasia is related to pathological defects that cannot be corrected, alternative measures must be utilized to minimize symptoms. Gentle mechanical removal of the nasal crusts should be educated and done by the patients. Alternating irrigations with both saline and gentamicin drops helps in decreasing crusts and odour coming from the mouth. Saline nose drops should be used as often as necessary. Removal of the crusts can also be done under general anaesthesia. Proper antibiotic cover is recommended to prevent secondary infections.

Conclusion

Ectodermal dysplasia is a rare but serious disease that has to be diagnosed as soon as possible because of the important problems it can cause. Features of the oronasal cavity along with general features are very helpful in providing a provisional diagnosis. Early addressal to these problems are suggested so that complications like palatal perforations do not occur.

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