CASE REPORT

Ectodermal Dysplasia presenting with Atrophic Rhinitis: A Report of Two Cases and a Review of the Literature

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ABSTRACT

Aims: To present two rare cases of ectodermal dysplasia presenting with maggot infestation due to atrophic rhinitis.

Study design: Case report.

Setting: A tertiary care referral hospital.

Presentation: Two male children aged about 5 and 8 years presented to the ear, nose, and throat emergency with a history of maggot infestation of the nose.

Results: Clinical examination was suggestive of ectodermal dysplasia. Intranasal examination was suggestive of atrophic rhinitis. Maggots were removed in the conventional manner. Following a conservative treatment, skin biopsy and nasal mucosal biopsy were done, which confirmed the diagnosis of ectodermal dysplasia.

Conclusion: Any ectodermal dysplasia patient should be suspected of having atrophic rhinitis and intranasal conservative therapy should be initiated at the earliest to prevent complications like maggots in the nose.

Keywords: Atrophic rhinitis, Ectodermal dysplasia, Nasal myiasis.

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INTRODUCTION

Sir Charles Darwin had mentioned in his book "The Variation of Animal and Plants under Domestication" about an Indian family with defective dentition and the inability to sweat properly in 1838. Although Thurnam¹ published the first report of a patient with ectodermal dysplasia in 1848, the term "ectodermal dysplasia" was not coined until 1929 by Weech.² Ectodermal dysplasia comprises of a large group of 192 distinct inherited disorders that are clinically and genetically heterogeneous with the common features of abnormal, absent, or delayed embryonic development of one or more of the epidermal and mucosal appendages. Among them the most common disorders is hypohidrotic or anhidrotic ectodermal dysplasia (Christ variety), which has an estimated prevalence of 1 per 100,000 births.³ The clinical triad of this disorder consists of hypotrichosis, anodontia or hypodontia, and hypohidrosis or anhidrosis.

CASE PRESENTATION

An 8-year-old boy attended the ear, nose, and throat (ENT) emergency with the complaint of live insects coming out of his nose; he also had fever and general weakness. Detailed history was taken from his mother, who revealed that her son suffered from recurrent attacks of high fever and heat intolerance along with occasional epistaxis, anosmia, and purulent discharge from the nose. He had frontal bossing with sparse thin hair on the scalp and eyebrows, saddle nose, and only three conical teeth (Figs 1 and 2). His developmental milestones and IQ level were in the normal range. Anterior rhinoscopy showed lots of crusting in the nasal cavity with maggots and with atrophic turbinates. Thermoregulation was achieved after admission and maggots were removed. Skin biopsy was performed, which revealed absence of eccrine sweat glands, and biopsy from nasal mucosa showed the absence of mucous glands (Figs 3 and 4). Thus, the child was diagnosed to be suffering from the rare disorder anhidrotic ectodermal dysplasia. He had no family history of a similar disease, and his male sibling was not affected.

A second similar case of maggots in the nose that presented to our department was that of a 5-year-old boy who came from a remote village with nasal myiasis

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Fig. 1: Clinical photograph of the 1st patient

Fig. 2: Conical teeth of the 1st patient

Fig. 3: Histopathological slide from skin biopsy showing absence of eccrine glands

Fig. 4: Nasal mucosal histopathology slide showing absence of mucous glands

and bilateral ear discharge. His body temperature was 103°F, and he was in a very irritable state. After admission, the maggots were removed and thermoregulation restored by repeated cold sponging. He had the same facies as the previous case but with complete alopecia and very dry skin. Interestingly, his mother also had sparse thin hair, dry skin, and the inability to sweat properly (Fig. 5).

DISCUSSION

Most of the patients of ectodermal dysplasia attend a dermatology or dental clinic first. But these rare cases may present with otorhinological manifestations as the chief complaint and must be diagnosed by the clinical features and proper history-taking. The clinical features are not evident in neonates. Dental, hair, and nail anomalies become evident only in early childhood. Other signs and symptoms include xerophthalmia,⁴ xerostomia,⁵ frequent otitis, rhinitis, pharyngitis,^{6,7}

Fig. 5: Clinical photograph of the second patient with his mother

growth failure,⁸ etc. Hypoplasia, or the absence of mucous glands in the upper aerodigestive tract, is responsible for these manifestations. Till now, only few cases have presented with nasal symptoms.⁹⁻¹³

For anhidrotic or hypohidrotic ectodermal dysplasia, the inheritance pattern is almost always X-linked recessive. Three genes that are related to the ectodysplasin protein and its receptor have been identified in the X chromosome at the q13 position. Like all other X-linked recessive disorders, only male children will manifest the disease and female carriers will be asymptomatic. But carrier females may sometimes show partial manifestation of the disease, which can be explained by lyonization or inactivation of one of the X chromosomes.¹⁴ That is why the mother of the second child in our case was mildly affected and the child being a male suffered from the whole stigmata of the disorder. But the same is not true for the first case because neither his mother nor his brother showed any features suggestive of this rare disorder. This must be a case of autosomal dominant inheritance. The gene for autosomal dominant hypohidrotic ectodermal dysplasia has been mapped to 2q11–q13.

Ectodermal dysplasia is a nonprogressive disorder with good prognosis. Morbidity can be decreased with orthodontic reconstruction; proper care of nose, ears, and oral cavity; and maintaining body temperature by lifestyle modification. There are several other forms of the disorder that need special attention like anhidrotic ectodermal dysplasia with immunodeficiency, which may give rise to recurrent systemic infection. Hidrotic ectodermal dysplasia patients may suffer from eye changes like cataract or strabismus. An erosive scalp dermatitis may be seen in AEC syndrome (ankyloblepharon, ectodermal dysplasia, cleft lip/palate), whereas ectrodactyly is pathognomonic in EEC syndrome (ectodermal dysplasia, ectrodactyly, cleft lip/palate). Carrier mother must be counseled about the risk in male child. Prenatal diagnosis is possible by fetal skin biopsy or by linkage analysis.

CONCLUSION

Ectodermal dysplasia is a rare entity that can first present to the ENT clinic with atrophic rhinitis, nasal myiasis, or recurrent ear infection. Proper history-taking, general examination, and family history are the cornerstones of

the diagnosis. Otorhinolaryngologists play a vital role in the health care team managing such patients.

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