INTRODUCTION

Ambras syndrome (AS) is a rare and special form of congenital hypertrichosis, characterized by dysmorphic facial features and familial pattern of inheritance. It is rarely associated with gingival hyperplasia. We report such a rare entity in a 38-year-old female patient with a history of consanguinity and positive family history.

CASE REPORT

A 38-year-old female patient reported to the department with a chief complaint of pain associated with decayed tooth in the lower right back region of the jaw, since 2 days. On general physical examination entire body including the face, hands, back, and external ear were covered with fine, light colored hair sparing the palms, soles, and mucosa. She gives a history of daily removal of hair by shaving. She also had dysmorphic facial features such as triangular facies and coarse features, hypertelorism, a wide and prominent nasal root, a large interalar distance, round nasal tip, and anteverted nostrils, Figures 1 and 2 depict these features.

The patient gives a history of her parents had second degree consanguinity. The antenatal period was uneventful, where there was no history of drug intake by her mother or radiation exposure. She gave a positive family history of her male sibling and maternal grandfather.

On intraoral examination, there was generalized gingival hyperplasia causing displacement of maxillary and mandibular anterior teeth with high arched palate and dental caries in relation to 46 [Figure 3].
Baseline investigations such as complete blood picture and hormonal analysis were noncontributory. Genetic analysis was advised, but the patient did not get it done due to financial constraints. Radiographical examination such as orthopantamogram [Figure 4] revealed impacted teeth in relation to 12, 22 and generalized interdental bone loss. Based on the history and clinical findings correlating with the previous literature, it was diagnosed as AS with gingival hyperplasia.

**DISCUSSION**

The first recorded case of AS is believed to be that of Petrus Gonzales in 1556, and Ambras name was given as his family portraits were discovered in Ambras Castle among an art collection started by the Archduke Ferdinand II (1529–1595). This syndrome was first reported by Baumeister et al. in 1993, and the second case by Balducci et al. The proposed genetic etiology for AS results alterations in chromosome 8 as analyzed in 2 out of 10 cases so far with pericentric inversion, inv(8)(p11.2q22), or paracentric inversion inv(8)(q12q22), or more complex insertion of the q23-24 region into a more proximal region of the long arm of chromosome 8. Downregulation of TRPS1 expression is the another probable cause of hypertrichosis in AS.

This condition affects both the sexes without any racial and geographical distribution. Clinically, it manifests mucocutaneous as long, fine, vellus hair covering the entire body, sparing the palms, soles, mucous membranes, dorsal terminal phalanges and associated with dysmorphic facial features such as triangular, coarse face, broad palpebral fissures, bushy eyebrows, hypertelorism, prominent bulbous nose, round nasal tip, large interalar distance, anteverted nares, and flat sulcus mentolabialis. Dental abnormalities such as anodontia, delayed primary, and secondary dentition may occur, and rarely associated with postaxial rudimentary hexadactyly and multiple exostosis, apart from above features the present case is associated with gingival hyperplasia, which is a rare entity. The diagnosis of AS is strictly based on clinical features.

The management of AS patients requires multidisciplinary approach, as it is associated with multiple system involvement. The main goal of therapy for AS is to improve the appearance (esthetics) of the affected person and concerned to hypertrichosis management, long-term...
removal of hair poses to be challenging to patients. Based on the degree of hair growth, the patient’s psychology and issue of social acceptance, the various methods for hair removal are epilatory methods such as mechanical and electronic tweezers, depilatory methods which include chemical bleaching and new therapies like lasers. The most promising therapy which induces retardation of hair growth is by eflornithine hydrochloride, ornithine decarboxylase inhibitor 13.9% cream.\textsuperscript{[1,8,9]} In this case reported, has all the clinical features of AS of Baumeister type with gingival hyperplasia and positive family history.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

**Conflicts of interest**

There are no conflicts of interest.

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