

Case Report

Ambras Syndrome with Clinico Radiographic Presentation- A Rare Case Report and Review of Literature

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ABSTRACT

Ambras syndrome, is a form of congenital hypertrichosis lanuginosa, it is characterized by dysmorphic facial features with excessive hair growth on the entire body, with the exclusion of the palms, soles, and mucous membranes. It follows autosomal dominant pattern of inheritance. It is rarely associated with gingival hyperplasia. We are reporting two case of this syndrome with dental and radiographic manifestation.

Keywords- AMBRAS Syndrome, Gingival hyperplasia, Dysmorphic facial features, Vellus hair, congenital hypertrichosis lanuginosa

INTRODUCTION

Baumeister et al^[1] was first defined the case of Ambras syndrome in 1993. Ambras syndrome is a very rare type of hypertrichosis lanuginosa congenita, a congenital skin disease characterized by excessive hair growth on the entire body, with the exclusion of the palms, soles, and mucous membranes. Individuals with Ambras syndrome have excessive growth of vellus hair, especially on the face, ears, and shoulders.^[1] Facial and dental abnormalities may also be present. Ambras syndrome has been recorded to the short (q) arm of chromosome 8₂, involving the region 8q22-24.^[2] It appears to follow an autosomal dominant pattern of inheritance. Until date, only 10 cases of Ambras syndrome have been documented according to literature.^[1,2]

Here we are reporting two case of this syndrome with dental abnormalities presented in mouth

CASE REPORT 1

19 year female patient was referred from a medical hospital to our department with complaint of swelling over upper and lower jaw since childhood. The patient also reported that for the last 3 years she had visited Dermatology Department for treatment for multiple nodules over her nose.

She gave history that her jaw and facial features are normal at time of birth. Her parents were observed, gums are enlarging, when she was around 3-4 years of age. Until date she was not taken any treatment for gingival enlargement. Her prenatal period was uneventful, where there was no history of drug intake by her mother or radiation exposure. She gives positive family history to her father. When she was 16 years, she noticed multiple round swelling (nodules) over nose. The recurrence rate was increases when she exposed to sunlight. For that she visited to dermatology department, biopsy of nodules was done. After that she referred to our

department for further management of gingival enlargement.

On general physical examination found that her 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.

On extraoral examination we observed she had dysmorphic features including triangular facies, hypertelorism, incompetent lip, bulbous nose, a wide and prominent nasal root, a large interalar distance, round nasal tip, and anteverted nostrils, healing scar with pigmentation noted over tip of nose and bushy eyebrows, hypertrichosis over arms [Figure-1] and legs.

On intraoral examination, there was generalized bulbous gingival hyperplasia causing displacement of maxillary and mandibular anterior teeth. Gingival growth is pink in appearance and fibrous on palpation. High arch palate, Ellis class I fracture wrt 11,21, discoloration noted wrt 21,dental caries wrt 46,47.Over retained tooth wrt 73[Figure- 2].

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.

She had incisional biopsy report of the multiple nodules over nose which showed atrophic epidermis in the basal layer, number of melanocytes increased in basal cell layer. Multiple dilated blood vessels are present in the dermis, sebaceous gland present with few vellus hair follicles. Mild lymphocytic infiltrate, mast cells in dermis, increase collagen tissue in dermis with increased number of fibrocytes. Overall features are suggestive of mild fibroplasia [Figure- 3].

For diagnostic purposed we advised digital orthopantomograph which revealed generalized spacing in between teeth, sparse trabecular bone pattern in maxilla and mandible, widening of inferior alveolar canal on both side. Ill-defined occlusal coronal radiolucency involving pulp noted wrt 36,37suggestive of caries. Diffused radiolucency surrounding mesial and distal root of 36 suggestive of periapical abscess. IOPA of mandibular anterior region and maxillary occlusal radiograph reveals sparse trabecular pattern [Figure-4].

Case 1-Clinical Photographs



Figure 1-Dysmorphic Facial features, Bulbous nose, Hypertrichosis over hand

After that we planned full mouth gingivectomy for gingival hyperplasia.

Gingivectomy of maxillary and mandibular anterior soft tissue growth was done and

sent for histopathology examination. Histopathology report shows hyperplasia, orthokeratinized epithelium with infiltrating connective showing dense collagen fibers with mild chronic inflammatory infiltrate suggestive of fibroepithelial hyperplasia. [Figure5]. Root canal treatment wrt 21,36,37. Extraction wrt 73. After 3 months

follow up she got good result after gingivectomy [Figure 2].

After that patient refer to dermatology department for treatment of multiple nodules over nose. Radiofrequency therapy was given. After 15 days we observed, the size of the nodules were reduced.



Figure 2-Generalized gingival hyperplasia, Bulbous gingiva, high arch palate discolored tooth wrt 21. Post treatment clinical picture

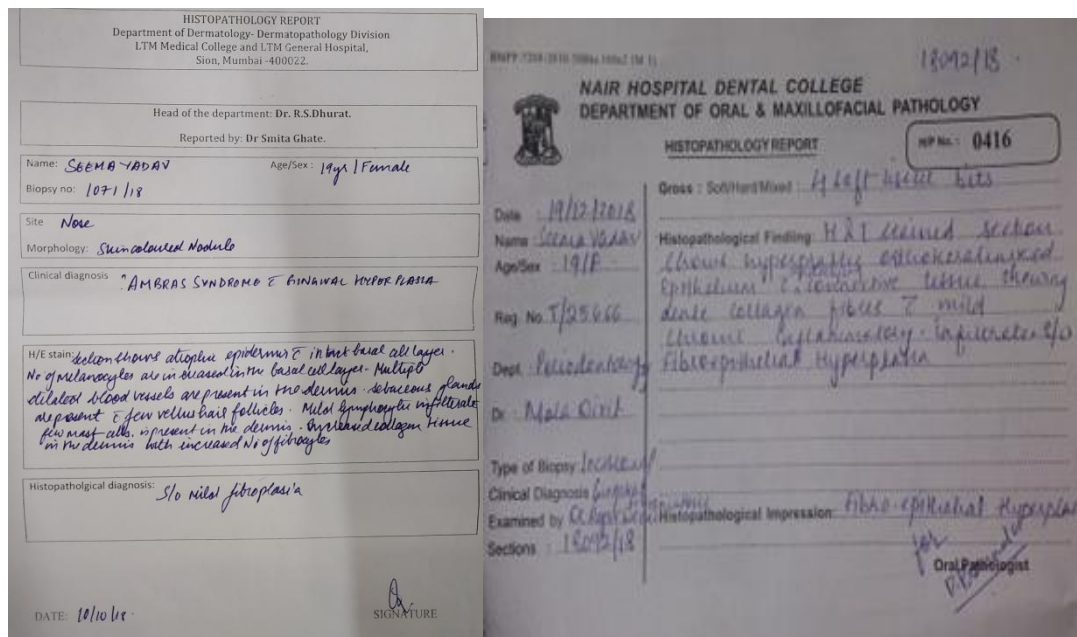


Figure 3- Report of soft tissue nodule Figure 5-Histopathological report of incised over nose gingiva of anterior region



Figure 4- IOPA & Maxillary Occlusal radiographs and panoramic radiograph shows sparse bony trabecular pattern, widened inferior alveolar nerve canal bilaterally

CASE REPORT 2

Patient father was called in the department of Oral and maxillofacial radiology. He was 45 years male. There was no significant family history noted. He had history of gingivectomy, at the age of 19 years .After that he was not taken any treatment.

On general physical examination found the entire body including the face, hands, back, and external ear were covered with fine, light colored hair except the palms, soles, and mucosa. He gives a history of daily removal of hair by shaving.

On extraoral examination, we observed he had dysmorphic facial features such as triangular facies, hypertelorism, bulbous nose ,a wide and prominent nasal root, a large interalar distance, round nasal tip, healed scar over nose and anteverted nostrils, hypertrichosis over arms and legs same as his daughter [Figure-6].

On intraoral examination there was generalized bulbous gingival hyperplasia, pinkish in appearance with smooth surface,

fibrous on palpation. Missing teeth wrt 11,12,21,22, 23, 24,41; 42. He had no history of removal of teeth. Displacement noted wrt13,31, high arch palate.[Figure-7]

Digital orthopantomograph and maxillary occlusal radiograph was taken which shows severe alveolar bone loss wrt 11,12,21,22,23 region, missing teeth wrt 41,42,43,37,38. Vertically impacted teeth wrt 18,28. Spacing in between teeth noted wrt 16,17,31,32,33. Well defined non-corticated smooth dome shaped radiopaque mass involving floor of maxillary sinus noted on right side suggestive of benign mucosal cyst.[Figure -8]

Complete blood and endocrinal evaluation was done, but not did not show any abnormality. Incisional biopsy of maxillary anterior gingival region was done and sent for histopathological evaluation. It shows hyperplastic stratified squamous epithelium with underlying connective tissue showing dense fibrous connective tissue. The features are suggestive of fibroepithelial hyperplasia.[Figure-9]

Case 2- Clinical Photographs



Figure 6- Dysmorphic Facial features, Bulbous nose,Hypertrichosis on the hand



Figure 7-Generalized gingival hyperplasia, Bulbous gingiva, high arch palate discolored teeth wrt 21, Post treatment clinical picture- 15 days follow up.

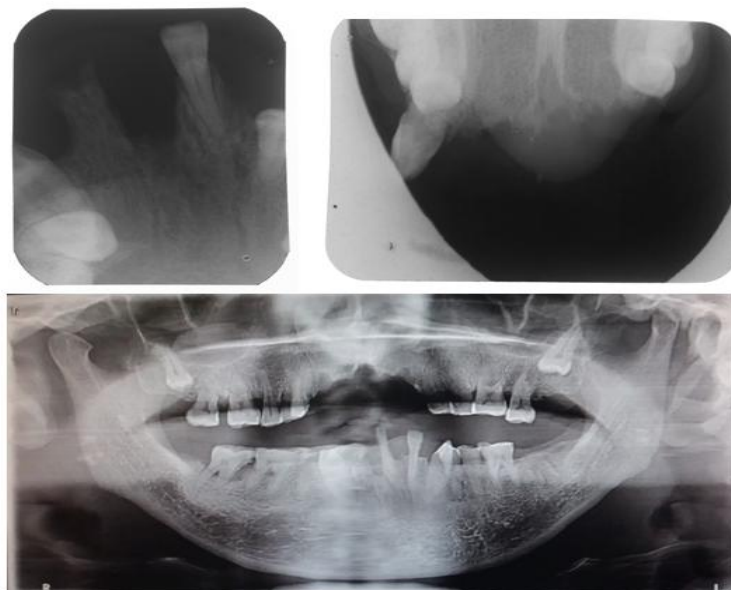


Figure 8- IOPA of mandibular anterior region, maxillary occlusal radiograph and OPG revealed severe alveolar bone loss wrt 11,12,21,22,23 region, Benign Mucosal cyst in right maxillary sinus, impacted teeth 18,28

16/3/18

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NAIR HOSPITAL DENTAL COLLEGE
DEPARTMENT OF ORAL & MAXILLOFACIAL PATHOLOGY

HISTOPATHOLOGY REPORT H/P No. : 0437

Date : 1/1/19
 Name : Mahendra Prabhu
 Age/Sex : 45/M
 Reg. No. : T/25698
 Dept. : Periodontology
 Dr. : Dr. Mala D.

Type of Biopsy : Incisional
 Clinical Diagnosis : Gingival Enlargement
 Examined by : Dr. Rajiv
 Sections : 16313/18

Gross : Soft/Hard/Mixed : Soft tissue, like

Histopathological Findings:
 The H&E stained slide section shows hyperplastic stratified squamous epithelium with underlying CT showing dense fibrous CT. The overall histopathological features suggestive of fibro-epithelial hyperplasia.

Histopathological Impression: Fibro-epithelial hyperplasia

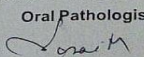
Oral Pathologist


Figure 9-Histopathology Report

DISCUSSION

Ambras syndrome was first reported in literature by Baumeister et al [1] in 1993 and the second case by Balducci et al. [2]. Ambras syndrome has been mapped to the short (q) arm of chromosome 8. It follows an autosomal dominant pattern of inheritance. The first recorded case of Ambras Syndrome was 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). [1, 3,4,5] This condition affects both the sexes without any racial and geographical distribution. Clinically it manifests 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.

In this two cases we found that long, fine, vellus type 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. Our findings are supported a case reported in literature by Vinay Kumar et al [6] in 2016 and Ishita A et al [7] in 2016.

Dental abnormalities such as anodontia, delayed primary and secondary dentition may occur and rarely associated with postaxial rudimentary hexadactyly and multiple exostosis. [4,5,6] In our case we found gingival hyperplasia in case both cases. Anodontia, impacted teeth are seen in case 2.

On radiological examination we observed severe alveolar bone loss in maxillary and mandibular anterior region, benign mucosal cyst of maxillary sinus on right side. Generalized sparse trabecular bone pattern [Figure -7]. Such radiographic findings are not reported in literature. The diagnosis of Ambras Syndrome is strictly based on clinical features.

In these two cases, full mouth gingivectomy using scalpel was planned for gingival hyperplasia. Gingivectomy using laser can also be executed. Another finding we observed that healing rate was delayed after gingivectomy in case 1 as well as case 2. Belengeanu et al. [8] described two siblings with reported Ambras syndrome

born to normal parents and propose that these patients might represent either an autosomal recessive pattern or germline mosaicism.

The main aim of therapy for Ambras Syndrome is to improve the esthetic appearance 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 removals are epilator methods which include 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.^[9,10]

CONCLUSION

The diagnosis of Ambras Syndrome is mainly on clinical features, characterized by a distinctive type of hypertrichosis at birth as described above, facial dysmorphism, gingival hyperplasia and familial pattern of inheritance. Goal of therapy is mainly to improve physical appearance.

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