IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 16, Issue 11 Ver. IV (Nov. 2017), PP 47-49
www.iosrjournals.org

Ambras Syndrome - A Rare Congenital Entity In Pediatric Population

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Abstract: *Ambras syndrome is a very rare type of congenital skin disease characterized by excessive hair growth on the entire body, with the exception of the palms, soles, and mucous membranes. Congenital generalized hypertrichosis associated with gingival hyperplasia are rare cases published in literature. The frequency incidence of generalized congenital hypertrichosis is about one to billions of people. Here, is a rare case report of three and half-year old girl who presented with generalized hypertrichosis with gingival fibromatosis and dysmorphic facial features is discussed.*

Keywords*: Ambras syndrome, dysmorphic face, gingival fibromatosis, hypertrichosis*

Date of Submission: 30 -09-2017 Date of acceptance: 30-09-2017

I. Introduction

Hypertrichosis is a clinical condition characterized by excessive hair growth without androgenic stimulation(1). If there is no underlying cause of the excessive hair growth, it is referred to as primary hypertrichosis, which is further classified as congenital or acquired, based on the age of onset and generalized or localized based on the distribution of body hair(2). Ambras syndrome (AS) is a rare form of congenital hypertrichosis associated with gingival hyperplasia and dysmorphic facial features in which the body is covered with long, fine, vellus-type hair. This very rare X-linked dominant condition has been responsible for the descriptions 'dog faced' and 'werewolf (3). Though benign in nature, there is considerable psychosocial trauma attached to this, owing to the cosmetic disfigurement it produces. Thus, a multidisciplinary approach involving a psychologist, a dentist apart from the dermatologist would be mandatory.

II. Case Report

A three and half year old girl was referred to the Department of Pedodontics with coarse hair present over the entire body since birth. This was the second child of her parents. Her 7 year old brother was normal. This child was born after normal delivery, with syndromic facies. She was born out of a non-consanguineous marriage with an uneventful antenatal and post-natal history, as described by the mother. There were no associated systemic complications. There was no history of photosensitivity. Intraorally thick and swollen gum pads were noticed at birth.

Patient had delayed milestones of development. But now she is engaged in normal activities with only abnormality being increased hair growth all over the body. She was playful like any other normal three and half year old and responded well to all commands. Patient was averagely built and nourished normal intelligence and other motor activity. The main reason for the consult was the issue of dysmorphic facial appearance and absence of teeth and generalised distribution of hair over the entire body, sparing the palms, soles and mucosal surfaces. Skin was normal with excessive hair growth on shoulders, hands, legs, and back region. Hair was longer and dark brown to black and soft and fine, increased intercanthal distance, and bushy, concrescent eye brows with long eye lashes. Depressed nasal bridge, bulbous nose, round nose tip, flared ala of nose with increased interalar distance. Upper and lower lips were thick, mentolabial sulcus was absent. (Fig.1, fig.2, fig.3)

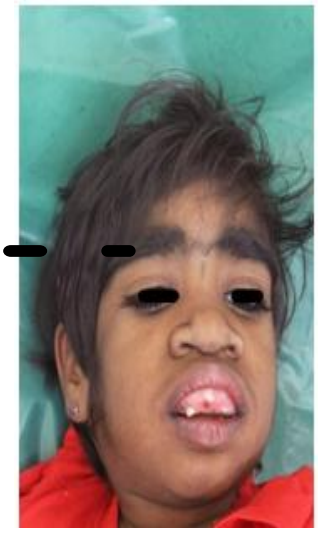

Figure 1: Dysmorphic face

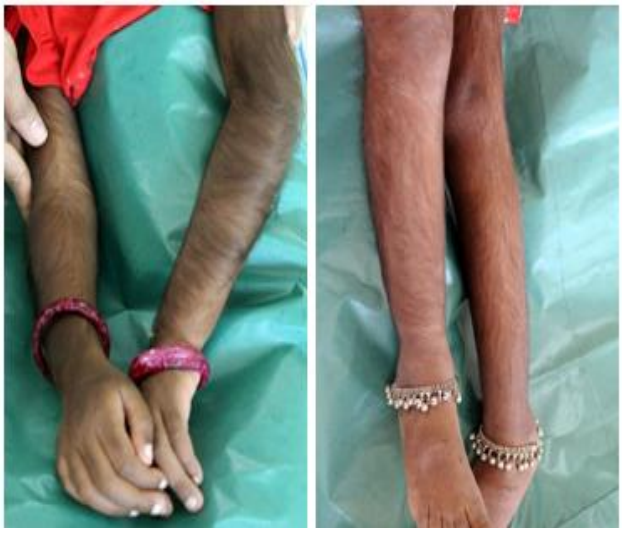

Figure 2,3: Hypertrichosis of hands and legs with bushy eyebrows and long eyelashes

Intraoral examination revealed generalized gingival enlargement which was pale pink in color, firm in consistency, and slight macroglossia was appreciated. In the maxillary arch 51, 61, 62, were seen clinically, 54 was covered with fibrotic gingiva (Fig.4). In the mandibular arch 73, 74, 81, 82, 83, 84, 85 were covered with fibrotic gingiva (Fig.5). Erupted teeth resembled features of the normal counterparts in par with the age and normal in appearance, and permanent tooth buds with normal follicular spaces, were seen in radiograph (Fig.6).

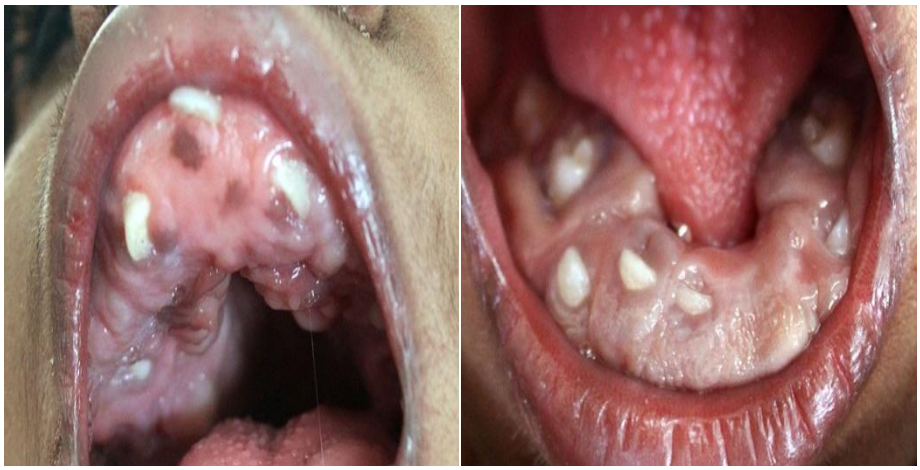

Figure 4, 5: Thick and fibrotic gingiva covering maxillary and mandibular teeth

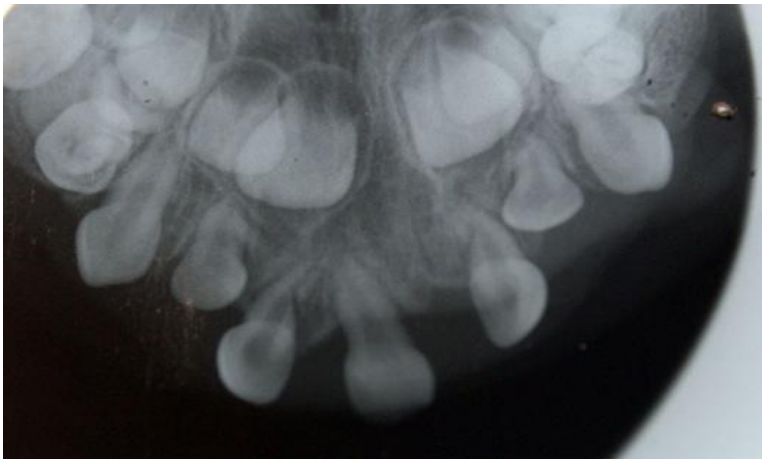

Figure 6: Maxillary occlusal view showing unerupted deciduous teeth

All these features were suggestive of a rare entity, AS, which consists of congenital hypertrichosis associated with gingival hyperplasia and dysmorphic facial features.

III. Discussion

AS was first described by Baumeister et al., in 1993 and believed to be that of Petrus Gonzales, as reported by Rashid M Rashid(4), followed by Balducci et al., who described the etiology as specific genetic alteration, that is, pericentric inversion (8) (p11.2; q22,) and can also be due to fetal alcohol syndrome and fetal hydentoin syndrome(5,6). It has also been noted that AS cases involve full-term births from non-consanguineous parents and no other family history of hypertrichosis. It is therefore of the greatest importance to confirm that drug is not the cause of the problem. In this case there was no history of drug intake by the mother during pregnancy period other than routine medications. Genetic mapping were not done due to financial constraints. Hirsutism due to adrenal hyperplasia or tumour is easily differentiated, for the distribution of coarse hair seen in these conditions is of the male sexual pattern. Congenital hypertrichosis lanuginosa also has to be excluded. This abnormality was first described by Thomas Bartholin in 1654. It is, however, un associated with gingival hyperplasia. Reported cases have been classified in two groups, both of which may be excessively hairy at birth. In the dog-face form the body, with the exception of the palms and soles, is covered with long silky hair from early childhood. The eyelashes are long and the eyebrows thick. This hair may grow to the length of 10 cm, and the hypertrichosis is permanent. Defects of the external ear and hypodontia or anodontia have been recorded. In some instances the condition is associated with mental retardation. In the monkey-face form hypertrichosis is always present at birth. Affected children usually die in infancy, but if they survive they develop a broad, flat, simian face with thick drooping lips and prognathism (Ebling and Rook, 1972)(7).

AS is characterized by increase in vellous type of hair growth on full body except for parts where normally no hair grows, like palm, soles of feet, mucosa, and dorsal terminal phalanges; the length of hair can reach several centimeters, which is silky and light in color; hair growth is specially seen on face, shoulders, and ear and uniformly covers eyelids, nose, cheeks, and periauricular region(4). A variety of dimorphisms like triangular coarse face, broad intercantha, broad palpebral fissures, long nose with round tip, wide interalar distance, anteverted nares, short integumental lower lip, and flat mentolabial sulcus, occasionally hexadactyly and supernumerary nipples are present(8). Other features include absence and delayed eruption of both the dentition, possibly due to gingival fibromatosis. The gingiva is usually of normal color, firm, and finely stippled in childhood, while later may acquire papillary projections. The grossly enlarged labial gingiva may prevent the closure of the lips and bilateral enlargement of the palatal mucosa may meet in the midline(7).

A variety of dental findings have been described in relation to congenital hypertrichosis lanuginose, including delayed eruption of permanent teeth, hypodontia, neonatal teeth, microdontia, and the presence of supernumerary teeth (9). But associated classical dysmorphic features characterizes AS. The patient requires periodic surgical excision of gingiva, regular home care oral hygiene measures, counseling, regular shaving of hairs for which different methods like mechanical shaving chemical depilatories, enzymatic depilatories, and permanent hair removal are available(10).

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*Layasha S. "Ambras Syndrome - A Rare Congenital Entity In Pediatric Population." IOSR Journal of Dental and Medical Sciences (IOSR-JDMS) 16.11 (2017): 47-49