

A 12-year-old girl with congenital hypertrichosis, hypertrophy of gum and breasts: A rare case report

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Abstract

We reported a 12-year girl who presented with congenital hypertrichosis, gingival hypertrophy and massively pendulous enlargement of breasts with normal biochemical and hormonal reports. Reduction mammoplasty, long pulsed Nd: YAG laser were done to reduce the breasts' size and hypertrichosis respectively. The histopathology of the resected breast tissue was compatible with virginal macromastia. This patient presented with a rare combination of several unusual features and requires multidisciplinary approach to improve the quality of her life.

Keywords: Congenital hypertrichosis, Gum hypertrophy, Virginal macromastia

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Introduction

Congenital generalized hypertrichosis (CGH) represents a heterogeneous group of conditions that are phenotypically and genetically distinct; it is characterized by excessive universal hair growth as the hallmark feature that is disproportionate when compared to normal individuals of that age, sex, and race and it is not dependent on androgenic hormones [1]. It has informally been called werewolf syndrome because the appearance is similar to that of a werewolf. Various genetic syndromes have been associated with hypertrichosis condition such as Ambras syndrome, Barber Say syndrome, and Cantu syndrome.

Gingival overgrowth is a common gingival disease which occurs due to inflammation; drugs such as phenytoin, nifedipine, or cyclosporine; leukemic infiltration; malignancies; and false enlargements [2]. Gingival fibromatosis with hypertrichosis (GFH) syndrome is an extremely rare condition of genetic origin characterized by the excessive growth of

gingival tissue with abnormal facial and body hairs. Gingival diseases of genetic origin are uncommon conditions and their most common clinically evident form is known as hereditary gingival fibromatosis (HGF). It may occur singly or in association with other inherited syndromes [3].

Breast hypertrophy is a rare medical condition of the breast connective tissues in which the breasts become excessively large. Hypertrophy of the breast tissues may be caused by increased histologic sensitivity to certain hormones such as female sex hormones, prolactin, and growth factors [4]. When macromastia occurs in young women during puberty, the medical condition is known as virginal breast hypertrophy. A diagnosis is made when an adolescent's breasts grow rapidly and achieve great weight, usually soon after her first menstrual period. Some doctors suggest that the rapid breast development occurs before the onset of menstruation [5]. We described a syndrome of congenital hypertrichosis, gum hypertrophy and virginal macromastia.

Case Report

A 12 years old Bangladeshi girl presented with excess growth of hair on whole body since birth, gradual swelling of gums for 5 years and progressive enlargement of both breasts without galactorrhoea for 1 year. She had yet not attained menarche. Her mother's antenatal, natal and postnatal periods were uneventful. She was delivered at full term by normal vaginal delivery. Her school performance and developmental milestones were normal. She was from a non-consanguineous marriage and all her siblings were healthy. She had short stature with normal body proportions [height was 134 cm (<5th percentile for age), weight 35 kg, BMI-19.49 kg/m², upper segment to lower segment ratio 0.94, arm span 130 cm]. There was firm, painless gingival hypertrophy. Her breasts were massively enlarged and pendulous reaching the inguinal regions (right

40 cm, left 37 cm from the sternal notch to nipple areola complex). Long, dark terminal hair covered the entire body sparing the palms, soles, axilla and anterior chest. There were no other dysmorphic features. Haematological, biochemical and hormonal assays were normal.

Ultrasonogram of both breasts showed huge enlargement with proliferation of all components of the breasts tissue and mild ductal dilatation. Ultrasonogram of whole abdomen revealed normal. ECG revealed low voltage ECG. Echocardiogram was normal 2D & M-mode study with good biventricular function. X-ray chest P/A view and X-ray dorsal spine were normal. X-ray OPG was normal. Reduction mammoplasty, long pulsed Nd:YAG laser were done. Histopathological examination of breast tissue was compatible with virginal macromastia.



Figure 1 : Front and side profile of the girl showing hypertrichosis and gum hypertrophy

Discussion

Hypertrophy of the breast is a rare benign disorder characterized by excessive and rapid growth of one or both breasts. The cases with virginal breast hypertrophy (VBH) reported in the literature are between the ages of 10-24 years [6]. The exact etiology is unknown, but several estrogen-related theories have been suggested. The most popular of these theories is end-organ hypersensitivity despite normal estrogen levels [7]. Our patient had congenital hypertrichosis, macromastia and gum hyperplasia with normal hormonal levels. A similar case has been reported in India, a 13 year old girl had all three of these features along with mild insulin resistance [8]. Another patient, reported by Li et al., was very similar to those of the present patient, except that our patient did not have any

cardiovascular anomaly, nor anomaly of palate [9].

The patient also lacked features of polycystic ovarian syndrome. The serum levels of the various androgens were normal. Because of association of increased body hair with insulin resistance, we studied the insulin levels in our patient which was $7.9 \mu\text{U/ml}$ ($2-25 \mu\text{U/ml}$) and IR index was 1.0. HOMA I.R index = 1 was considered normal. Although there is no medical treatment, reduction mammoplasty for macromastia and laser therapy for hypertrichosis was done for this patient to improve quality of her life. Removal of about 9 kg breast tissue from each breast after reduction mammoplasty surgery she improved dramatically and can walk normally without her back pain. After laser therapy her facial hair significantly improved.



Figure 2 : Breast enlargement

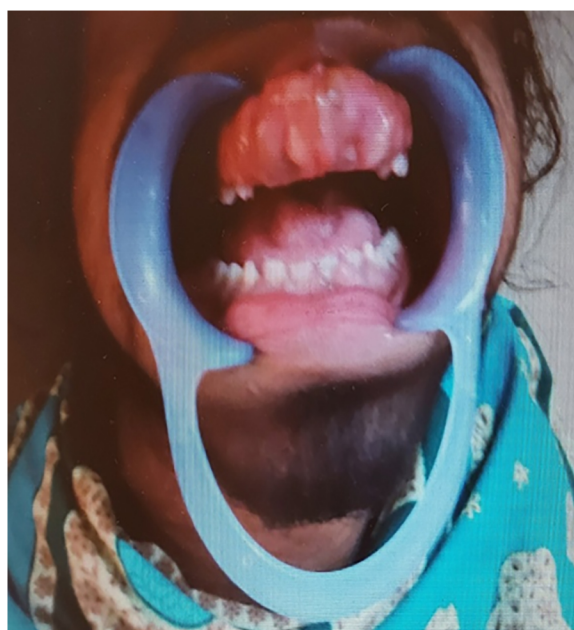


Figure 3 : Gum hypertrophy

Conclusion

Virginal breast hypertrophy is seldom found in our clinical practice, especially associated with congenital hypertrichosis and gum hypertrophy with normal hormonal levels. All these findings are excessive growth of different tissues. Although the exact mechanism is not known, it could be related to altered receptor sensitivity. It requires a multidisciplinary team with an empathetic approach for optimum outcome.

Acknowledgment

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Disclosure

The authors have no multiplicity of interest to disclose.

Table 1 : Hormonal Profile of the Patient

	Patient's value	Normal range
Serum follicle-stimulating hormone (FSH), mIU/ml	7.07	3-20
Serum luteinizing hormone (LH), mIU/ml	7.92	5-15
Serum free testosterone (pg/ml)	1.58	0.29-3.18
Serum dehydroepiandrosterone sulphate (DHEA-S), µg/dl	14.30	5-40
Serum estrogen (pg/ml)	71.6	<75
Serum progesterone (ng/ml)	0.18	1.13 - 10.2
Serum 17-hydroxy progesterone (ng/ml)	0.45	0.07-1.70
Serum prolactin (ng/ml)	15.7	3.2-20
Serum thyroid stimulating hormone (TSH), µIU/ ml	2.60	0.35-5.50
Serum free thyroxine (FT4), ng/dl	1.32	0.8-1.8
Basal cortisol (nmol/L)	264.3	138-690
Plasma adrenocorticotrophic hormone (ACTH), pg/ml	25.8	Undetectable to 46
Basal growth hormone (ng/ml)	0.082	0.06-5

Table 2 : Biochemical Profile of the Patient

Parameter	Patient's value	Normal range
Fasting blood glucose (FBG), mmol/L	4.5	<5.6
Plasma glucose 2 hour after 75 gm OGTT (mmol/L)	5.5	<7.8
HbA1C	4.6%	<5.7%
Fasting plasma insulin (µIU/ ml)	7.9	2-25
HOMA I.R index	1	—

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