Juvenile breast hypertrophy: report of two cases presenting with massive asymmetrical breast enlargement and palpable breast masses

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ABSTRACT

Background: Juvenile breast hypertrophy is a rare benign condition leading to rapid and massive enlargement of one or both breasts in young girls during peri-pubertal period. Patients encounter physical and psycho-social problems. Therefore, the condition warrants early appropriate management. Breast imaging studies (ultrasound and **magnetic resonance imaging**) are particularly important in order to exclude tumors and occult pathologies. First line treatment modality is reduction mammoplasty that significantly improves quality of life with lesser chances of recurrence.

Case Presentation: We report two post-menarchal girls with juvenile breast hypertrophy who presented as different clinical cases facing psychological and social embarrassment. Breast ultrasound showed right breast mass in both patients that were biopsied. The lesions turned out to be fibroadenomas on histopathological correlation. Patients were then referred to plastic surgery and endocrinology department.

Conclusion: Juvenile breast hypertrophy is definitely a disturbing cosmetic problem. Early investigations and appropriate management will surely leave a positive impact on the quality of life.

Keywords: Case report, breast, hypertrophy, peri-pubertal, ultrasonography, fibroadenomas, mammoplasty.

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Background

Juvenile breast hypertrophy also known as virginal hypertrophy or macromastia is a rare and relentless benign disease [1]. There is massive increase in size of one or both breasts during peri-pubertal period [2]. Patient often experiences initial rapid growth phase for 3-6 months followed by a longer slow growth phase that continues into childbearing years if the condition is left untreated [3]. It is a rare condition, only 65 cases were reported between 1910 and 2009 in the meta-analysis conducted by Hoppe et al. [4] and only additional nine cases were identified between 2010 and 2017 according to the study conducted by Hisham et al. [2]. Extreme breast enlargement causes physical and psychological problems in the form of back pain, shoulder pain, poor-fitting clothing, trouble in exercising, and lack of confidence [5]. The proposed underlying mechanisms include increased levels of gonadal hormones or end-organ hypersensitivity to normal levels of gonadal hormones [6]. However, these theories still need investigatory support. Disease management revolves around minimizing mass effect of the enlarged breast and improvement in quality of life. Effective therapies include

surgical management and hormonal treatment. However, chances of recurrence are there [5].

Case Presentation

Case # 1

A 13-year female patient presented to our breast clinic with the complaint of bilateral breast enlargement and palpable right sided breast lump for 1 year. There was enormous increase in size of both breasts in the last 6 months. The condition was causing her severe mastalgia and refraining her from schooling and social activities. She attained thelarche at 11 years of age and menarche at 12 years of age. Her past medical and family history was unremarkable. She has not received any medication yet.

On examination, she was a slim girl with a normal body mass index (BMI) of 20 kg/m²(weight = 48 kg, height = 1.55 m). Both breasts were pendulous and disproportionately enlarged; right breast larger than left one, and with widened areolas. Palpation revealed uniformly firm texture with a palpable mass on right side. There was no history of trauma, sores, nipple discharge, or itching. Bilateral breast ultrasound was performed which showed diffusely thickened stromal and glandular tissue bilaterally (Figure 1). In addition, a large well defined hypoechoic lesion having smooth margins was also noted occupying upper half of right breast measuring 9.8×5.2 cm, showing moderate vascularity on color Doppler evaluation (Figure 2). There was no skin thickening or nipple retraction. No significant axillary lymphadenopathy was found. Ultrasonography (USG) diagnosis of juvenile breast hypertrophy associated with a giant fibroadenoma in right breast was made.

Trucut biopsy of right breast lump was taken and sent for histopathology that showed benign fibroepithelial lesion with features favoring fibroadenoma.

Patient was referred to endocrinology and plastic surgery department for appropriate management.

Case # 2

Another 14-year female patient presented to our breast clinic with progressive massive bilateral breast enlargement for 8 months. In addition, patient complained of mastalgia, neck discomfort, and severe backache. She was facing psychological problems and social embarrassment as well. She attained thelarche at the age of 12 years and menarche at the age of 13 years. Her past medical and family history was unremarkable. She has not taken any medication yet.

On examination, she was a healthy patient with normal BMI of 22.2 kg/m² (weight = 45 kg, height = 1.50 m). Breasts were asymmetrically enlarged and pendulous with widened areolas; right breast larger than left one (Figure 3). There were two palpable mobile lumps on right side.

Pressure sores were also found in the infra-mammary folds. No nipple discharge, skin discoloration, or itching was seen. No history of trauma was given by the patient.

Bilateral breast ultrasound showed diffusely thickened edematous breast parenchymal tissue bilaterally. In addition, two well-defined hypoechoic lesions were noted in upper outer quadrant of right breast, one measuring 24 \times 15 mm and other measuring 21 \times 10 mm. USG diagnosis of juvenile breast hypertrophy associated with two fibroadenomas in right breast was made. No significant axillary lymphadenopathy was found on either side.

Trucut biopsy of right sided breast lumps followed by histopathology revealed these lesions to be benign fibroepithelial lesions with no evidence of atypia. Features favor fibroadenomas.

Patient was referred to endocrinology and plastic surgery department for further management.

Discussion

Juvenile breast hypertrophy is a rare and benign condition; however, its diagnosis is confirmed only after detailed patient history, thorough physical examination and exclusion of underlying conditions such as malignant or endocrine disorders. The condition is accompanied by physical and psychological consequences that negatively impact the quality of life so psychological monitoring plays a major role in disease management [1].

Several theories have been proposed regarding exact underlying etiology for juvenile breast hypertrophy. The popular theories include end-organ hypersensitivity to normal level of gonadal hormones, imbalanced endogenous hormone production, increased expression of



Figure 1. (A) (Right breast) and (B) (Left breast): ultrasonography shows diffusely thickened stromal and glandular tissue bilaterally.



Figure 2. Large well defined hypoehoic lesion occupying upper half of right breast having smooth margins showing moderate vascularity on color Doppler evaluation. Histopathology revealed it to be fibroadenoma.



Figure 3. Clinical photograph: there is massive asymmetric enlargement of both breasts which appear pendulous with widened areolas.

estrogen or progesterone receptors and excessive local estrogen production. Phosphatase and tensin homolog gene tumor suppressor gene mutation and deletion has also been postulated to have a link with precocious lobulo-alveolar development and excessive ductal branching [2]. Laboratory investigations including endocrinology profile rarely reveal abnormalities.

Breast imaging studies are particularly important in order to exclude tumors and occult pathologies. Mammography is difficult to interpret in young women owing to dense breast parenchyma and has a very limited role in this respect. However, ultrasound has a very important role in this age group, and it is essentially needed for detection as well as characterization of any masses; if present in the enormously enlarged breasts in cases of juvenile breast hypertrophy. Ultrasound is required to differentiate between cystic/solid and benign/malignant breast lesions. Ultrasound can also provide image guidance for percutaneous biopsy of focal lesions [7]. In both our reported cases ultrasound showed associated benign looking solid breast masses; one in first case and two in second one. These masses turned out to be fibroadenomas on histopathology. The association of juvenile breast hypertrophy and fibroadenomas has not been discussed in the previous case reports. Magnetic resonance imaging (MRI) may be more helpful in delineating breast architecture and occult pathologies in indeterminate cases.

Differential diagnosis of juvenile breast hypertrophy includes giant fibroadenoma, phylloides tumor, lymphoma, and sarcomas.

Spontaneous resolution is rare and timely management is the most challenging aspect. Treatment options include surgical management and medical therapy (preoperatively, postoperatively, or alone) [8]. First line option is reduction mammoplasty with free nipple graft or pedicle-based technique. This is followed by mastectomy and implant reconstruction in cases of recurrence. Of reduction mammoplasty techniques, free nipple graft technique has comparatively lesser chances of recurrence. But it must be delayed until full breast development has been achieved as surgery in active growth phase can lead to recurrence [6]. Free nipple grafting however has certain drawbacks including loss of lactation, graft failure, variable return of sensation and contractility of the nipple, and pigmentary changes of the nipple-areolar complex. Therefore, it is essential to counsel the patient and her family members about the limitations of this procedure [3].

Medical management include hormone modulators, for example; tamoxifen, dydrogesterone, medroxyprogesterone (Depo-provera), bromocriptine, and danazol. Most of the proposed theories regarding etiology of Juvenile breast hypertrophy (JBH) revolve around the estrogen imbalance. So tamoxifen, being an estrogen receptor modulator, can be a useful adjunct to surgical management despite normal endocrinology profile [1]. Use of tamoxifen in preoperative period followed by reduction surgery, gives stable post operative results in younger breasts JBH is prone to recurrence [9]. Some studies also show that a combination of reduction mammoplasty and "adjuvant" tamoxifen therapy can eliminate the need for repeated surgeries in young patients [1]. Furthermore, in the experienced clinical setups, tamoxifen therapy can become an alternative to surgical reconstruction during the brisk growth phase of the disease [10].

However, there are no evidence-based recommendations regarding the timing and dosage of these hormone modulators. Higher doses should be used with extreme caution. For example, side effects of tamoxifen include endometrial hyperplasia, venous thromboembolism, hot flashes, and bone density changes [2].

Conclusion

Juvenile breast hypertrophy is a disturbing cosmetic problem. Early diagnosis and timely management can improve physical and psychological outcomes. However, breast imaging studies including ultrasound and MRI result in non-specific imaging findings, and thus remain mostly non-conclusive. They are of value in detecting occult masses and image guided biopsies. A combination of reduction mammoplasty and tamoxifen therapy can eliminate the need for repeated surgeries in young girls.

What is new?

Juvenile breast hypertrophy is a rare and benign condition; however, its diagnosis is confirmed only after detailed patient history, thorough physical examination and exclusion of underlying conditions such as malignant or endocrine disorders. It is a rare condition, only 65 cases were reported.

List of Abbreviations

BMIBody mass indexMRIMagnetic resonance imaging

Conflict of interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

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Consent for publication

Written informed consent was obtained from the patients for publication of this case report and any accompanying images.

Ethical approval

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Summary of the case

1	Patients (gender, age)	Females (13 and 14 years old)
2	Final diagnosis	Juvenile breast hypertrophy
3	Symptoms	Case 1: Bilateral breast enlargement and palpable right sided breast lump for 1 year. Case 2: Progressive massive bilateral breast enlargement for 8 months.
4	Medications	No medication
5	Clinical procedure	Bilateral breast ultrasound core biopsies of breast masses
6	Specialty	Diagnostic radiology