Case Report

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An unusual case of huge hamartoma of breast in a 23 year old female: a case report

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ABSTRACT

Breast hamartoma is an underdiagnosed and poorly recognized rare benign lesion, accounting for approximately 4.8% of all benign breast lumps. There is lack of awareness of the characteristic clinical and histologic features of this entity in clinicians and pathologists. The pathogenesis of the development of breast hamartoma is still not fully understood. A 23-year-old female presented with a huge painless lump in right breast. The lump was present since last nine years. It was previously of the size of a bean, which rapidly increased to the present size during her pregnancy. FNAC revealed features of lipoma. However, on histopathological examination diagnosis of Hamartoma was rendered. A definitive diagnosis of hamartoma is hard to achieve on clinical examination, imaging studies and fine needle aspiration cytology. A correlation of histology, imaging findings with clinical impression is necessary.

Keywords: Breast, Hamartoma, Huge

INTRODUCTION

Breast hamartoma is an underdiagnosed and poorly recognized rare benign lesion, accounting for approximately 4.8% of all benign breast lumps. 1-2 However, with increasing awareness and widespread breast cancer screening, hamartomas are being diagnosed with greater frequency. The pathogenesis of hamartoma remains unclear and its diagnosis is underestimated by clinicians and pathologists. 3

Majority of affected patients are more than 35 years old, with a mean age of 45 years. Moreover, breast hamartomas range in size from 0.9 cm to 6.9cm. This benign lesion grows slowly and can become bigger if no intervention is done. In most of the cases, the breast hamartoma is excised within few months to years after its presentation, when it is not too big. However, the present case was seen in a young female with huge mass of a long history.

CASE REPORT

A 23-year-old female presented with a large lump in right breast. The lump was painless and present since 9 years with rapid increase in size during her pregnancy. There were no other associated complaints.

On local examination, the lump was located in upper inner and upper outer quadrant of right breast. The lump was freely mobile with smooth round contour. Approximate size on clinical examination was 20 cm in diameter. No skin ulceration or fixation was observed. Axillary lymph nodes were not palpable. Fine needle aspiration cytology (FNAC) examination revealed fragments of adipose tissue only. Ductal elements of breast were not seen in cytology smears. Hence a diagnosis of features suggestive of lipoma was given on FNAC. Successful excision of this lump was performed under general anesthesia and the lump was sent for histopathological examination.

Gross examination of the specimen revealed a single, well encapsulated yellowish ovoid mass measuring 21.5 x 18 x 4.8 cm and weighing 1100 grams. The mass had smooth rounded borders. Serial cut sections revealed greyish yellow homogenous smooth cut surface with interspersed grey white areas. There was no evidence of hemorrhage or necrosis on gross examination (Figure 1 and Figure 2). Extensive sampling was done and sections were stained with H and E stains.



Figure 1: Large ovoid well encapsulated greyish vellow mass.



Figure 2: Cut surface-solid and greyish yellow.

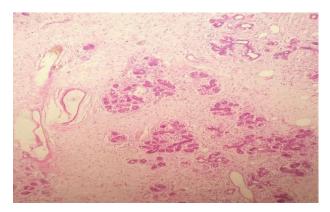


Figure 3: Microphotograph shows ductal elements-TDLU (terminal duct lobular unit) in a collagenised stroma (H and E-40X).

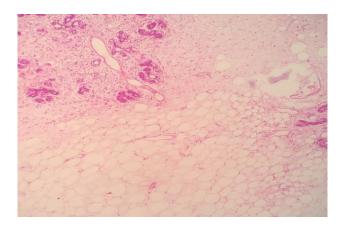


Figure 4: Microphotograph shows adipose tissue admixed with ductal elements (H and E -40X).

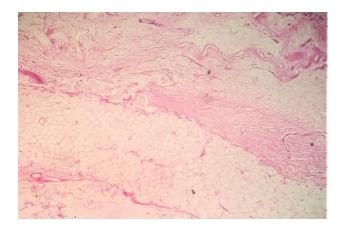


Figure 5: Microphotograph shows areas of fibrosis admixed with adipocytic element (H and E -40X).

Microscopy showed a well circumscribed lesion composed predominantly of lobules of adipose tissue admixed with terminal duct lobular units (TDLUs). Zones of fibrosis and hyalinization were seen interspersed in between. The ductal elements did not reveal any atypia (Figure 3, Figure 4 and Figure 5). Considering these features diagnosis of hamartoma of breast was given.

DISCUSSION

The word Hamartoma, Greek in origin meaning "Bodily defect", refers to the presence of a disorganized mixture of components which are endogenous to a particular site. Hamartoma is considered benign non-neoplastic lesion that may recur.

Breast hamartoma is a well circumscribed benign lesion composed of glandular tissue, epithelial elements, fibrous tissue and fat, which may be in normal or varying proportions. Hogeman and Osbterg were the first to describe this lesion back in 1968.⁴ Several cases were reported as adenolipomas and lipofibroadenomas.⁵ Then the actual term "hamartoma" was created in 1971 by Arrigoni et al.⁶ who concluded that breast hamartomas account for approximately 4.8% of all benign breast

lesions.¹ The exact pathogenesis of breast hamartoma development remains a mystery. However, the histological structure of breast hamartoma indicates that the tumor probably results from a dysgenetic factor rather than a neoplastic process, as recognized in the World Health Organisation in 1981.⁷ Although it is histologically benign, in situ and infiltrating carcinomas may arise within hamartomas.⁸⁻¹⁰

The lesion occurs predominantly in premenopausal women, mostly in their 40s with a wide range of teenage to women in their 80s. In our case, patient had this lesion from the age of 14 years, which is quite an earlier presentation. Patient observed this lesion when it was of the size of a bean. Lump increased in size slowly and gradually. Striking increase in size was observed during her pregnancy and the lesion achieved the present huge size of 21.5cm. This supported the assumption that both epithelial and stromal components of breast hamartoma express hormone receptors.¹¹ Hence this hormone responsive lesion proliferated tremendously during pregnancy.

Clinically, breast hamartomas are painless, mobile, soft to firm lumps, typically found in outer breast quadrants.¹² This usually misleads clinicians to diagnose them as fibroadenomas or lipomas. On ultrasound hamartomas appear to be well circumscribed with a smooth border and internally hypoechoic or of heterogenous echogenicity. They lack a retrotumor acoustic phenomenon which is one sonographic finding used to differentiate between benign and malignant lesions. 13 The appearance on mammography varies depending on degree of each of its constituents. Classically, they appear non homogenous with dense nodules consisting of fibrous tissue surrounded by a thin radio-opaque pseudocapsule formed from the displaced parenchyma of breast. Sometimes, it is difficult to distinguish it from fibroadenoma, when the lesion appears homogenously dense due to rich fibrous component. Though, fine needle aspiration cytology (FNAC) and needle core biopsy accurately diagnose most breast lesions, they are not very helpful in diagnosing hamartomas and differentiating them from other entities which mimic them.

In our case, clinical diagnosis of lipoma was considered. No ultrasonography or mammography was performed. Fine needle aspiration cytology (FNAC) of this lesion was advised. Cytological smears revealed only fragments of fibroadipose tissue and no ductal elements. Hence diagnosis of lipoma was given on cytology. Grossly, hamartomas are well circumscribed, round to oval, having smooth borders and measuring upto 20cm in size. Capsule may or may not be there. Sometimes a thin pseudocapsule is observed. Cut surface mostly resembles normal breast or fibroadenoma, depending on the constituents of lesion.

Histologically, hamartomas are mostly encapsulated, whether by a capsule or a surrounding rim of

pseudocapsule. The lesion is lobulated and exhibits mammary ducts, lobules, fibrous tissue and adipose tissue in varying proportions. There were attempts in the past to classify breast hamartomas into specific categories based on their histological appearance. Mc Guize et al, categorized them into fibrous, fatty and fibrofatty, whereas adenolipoma, fibroadenoma like, fibroadenoma with fibrous stroma and encapsulated fibrocystic changes were the categories given by Jones et al. ¹⁴ These classifications are not generally accepted by most authorities as they are not reproducible and have no clinical impact.

Recurrence rate of hamartomas is seen in around 8% cases. These cases most likely represent multifocal disease rather than true recurrence. Three months follow up of our case is uneventful. Surgical excision of breast hamartomas is considered to be curative. They have an excellent prognosis with or without surgical excision. As most of the hamartomas are circumscribed, often encapsulated with well delineated borders, they can be easily enucleated.¹⁵

CONCLUSION

Breast Hamartomas are uncommon benign lesions which possess certain distinguishing characteristic features on histology. A definite diagnosis is hard to achieve on clinical and radiological examination. Hamartomas are painless, histologically benign masses which could develop to a quite large size if local excision is not performed in time. As the ductal elements and stroma of this lesion are hormone responsive, a sudden increase in size of the lesion during pregnancy can occur.

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