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Hamartoma breast: A rare case report highlighting the limitations of aspiration cytology

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Abstract

A 26 year female presented to FNAC clinic with a lump right breast. It was firm, measuring 3x3 cm in central quadrant, FNAC yielded moderately cellular smear, showing presence of antler horn clusters, monolayered sheets and blunt tipped clusters of monomorphic ductal epithelial cells with interspersed myoepithelial cells and bare bipolar nuclei. Scant stromal fragment were seen. Cytological diagnosis of fibroadenoma was given.

Subsequent USG, both breasts revealed normal fibro-glandular tissues with predominantly fatty tissues interspersed within, with no definite abnormality.

The lesion was excised and we received a specimen size measuring 7x4x3cm, showing circumscribed non-encapsulated lesion. On cut surface, fatty areas admixed with grey white firm areas were seen. Sections showed disorganised TDLU with interspersed adipose tissue and scant area of dense hyalinized stroma. Few ducts were cystically dilated with focal apocrine metaplasia. Lesion was diagnosed as Hamartoma breast. A case is being discussed for overlapping cytological feature with fibroadenoma.

Keywords: Disorganised TDLU, fibroadenoma, adipose tissue

Introduction

Breast hamartomas are rare, benign, and slow-growing breast lesions, accounting for approximately 4.8% of all benign breast masses ^[1, 2]. They are most commonly encountered in middle-aged women, particularly during the perimenopausal period ^[2]. Clinically, breast hamartomas typically present as painless, mobile, and firm-to-soft palpable masses, often located in the outer quadrants of the breast, and may occasionally manifest as anisomastia ^[2]. Histologically, these lesions are well-circumscribed, comprising a mixture of glandular tissue, epithelial elements, fibrous stroma, and adipose tissue in varying proportions ^[1]. The definitive diagnosis of breast hamartomas relies on a triple assessment, integrating clinical examination, radiological findings, and histopathological evaluation ^[1]. Although generally benign, rare cases have reported an association with breast malignancies, with the incidence of carcinoma arising within hamartomas being as low as 0.1% ^[1].

The histologic architecture of breast hamartomas suggests a dysgenetic origin rather than a true neoplastic process. Their association with dysgenetic syndromes, such as Cowden syndrome linked to PTEN gene mutations, further supports this hypothesis ^[3]. However, cytological diagnosis remains challenging due to the heterogeneous tissue composition and overlapping features with other benign breast lesions

Case presentation

A 26-year-old female presented to the Fine needle aspiration cytology (FNAC) clinic with a firm, mobile lump measuring 3 × 3 cm in the central quadrant of the right breast.

No definite abnormality was reported on USG.

FNAC smears were moderately cellular, showing *antler-horn clusters*, monolayered sheets, and blunt-tipped clusters of monomorphic ductal epithelial cells. Scattered myoepithelial cells and bare bipolar nuclei were also noted. A cytological diagnosis of fibroadenoma was suggested.

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Histopathology

Gross examination revealed an excised specimen measuring $7 \times 4 \times 3$ cm, irregular, demarcated, but not-encapsulated lesion. On cut showed yellowish area admixed with greyish white area. Multiple sections taken in different plane.

Microscopy showed disorganized terminal duct lobular units (TDLUs) interspersed with adipose tissue and areas of dense hyalinized stroma. Few ducts were cystically dilated with focal apocrine metaplasia and epitheliosis. Based on these features, a final diagnosis of breast hamartoma was made.

Discussion

Pryn first identified breast hamartoma as “mastoma” in 1928. Breast hamartoma is a rare and benign disease of the breast. Hogeman and Osbtang were the first to describe this lesion in 1968. Subsequently, the disease was named breast hamartoma by Arrigoni *et al.* [4].

Breast hamartomas are rare, benign, slow-growing lesions, constituting approximately 4.8% of all benign breast masses. They typically present as painless, mobile, firm-to-soft

masses, commonly located in the outer quadrants of the breast. [2] Histologically, they consist of glandular, fibrous, and adipose tissues in varying proportions. Radiological imaging plays a crucial role in the diagnosis of breast hamartomas.

Findings include

- **Mammography:** Well-defined lesions with mixed density, showing fatty and fibrous elements. A thin radiopaque capsule is often seen. Dense hamartomas rich in fibrous tissue can mimic fibroadenomas.
- **Ultrasound (USG):** Well-circumscribed lesions with smooth borders, showing heterogeneous or hyperechoic internal echoes. Some cases may be missed on USG despite clinical findings.
- **MRI:** Shows heterogeneous intensity on T1- and T2-weighted images with a thin capsule surrounding the lesion. Detection rates: Mammography 30%, USG 18%, MRI 68%. [1,2]

Table 1: Imaging Features of Breast Hamartoma

Imaging Modality	Findings	Detection Rate	Differential Challenges
Mammography	Mixed density, encapsulated, well-defined	30%	Dense lesions mimic fibroadenoma
Ultrasound	Well-circumscribed, heterogeneous, hyperechoic	18%	Occasionally missed
MRI	Heterogeneous intensity, thin capsule	68%	Best for accurate diagnosis

Cytological diagnosis of breast hamartomas is challenging due to its heterologous composition and overlapping features with other benign breast diseases. FNAC often yields nondiagnostic or limited samples, showing sparse ductal epithelial cells, scant myoepithelial cells, and variable amounts of adipose tissue.

Histologically, breast hamartomas are composed of a

mixture of breast ducts, lobules, fibrous stroma, and adipose tissue in varying proportions. Three histological subtypes have been identified: [4, 5]

- **Fibroadenolipoma:** Predominance of fibrous and adipose tissue
- **Myoid hamartoma:** With smooth muscle proliferation
- **Chondrolipoma:** Presence of cartilaginous components.

Table 2: Differential Diagnosis of Breast Hamartoma

Lesion	Key Features	Differentiating Points
Fibroadenoma	Well-defined, uniform stroma	Lacks mixed adipose and glandular tissue
Phyllodes Tumor	Hypercellular stroma, leaf-like projections	Increased mitotic activity
Lipoma	Homogeneous fat density	No glandular or ductal elements

Ductal hyperplasia, apocrine metaplasia, calcification and adenosis may occur within the hamartoma, with rarer instances of lobular or ductal intraepithelial neoplasms. Although hamartoma is usually benign, a malignant transformation is possible [6].

Diagnosis is based on a triple assessment: clinical examination, imaging, and histopathology. Core or fine-

needle biopsy is often insufficient; excisional biopsy is usually required.

Surgical excision is the treatment of choice and is considered curative [2].

The risk of malignancy within a hamartoma is very low (~0.1%) [1].

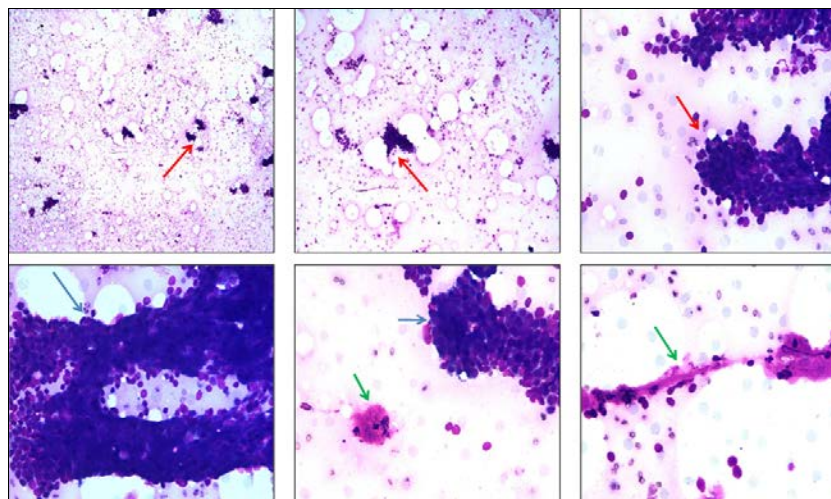


Fig 1: Picture collage of FNAC showing moderately cellular smear with antler horn clusters (Red arrow), monolayered Sheets, blunt tipped monomorphic ductal epithelial cell clusters (blue arrow), interspersed myoepithelial cells. Bipolar naked nuclei (black arrow) and fibrous stromal fragment (Green arrow).



Fig 2: Legends inbox

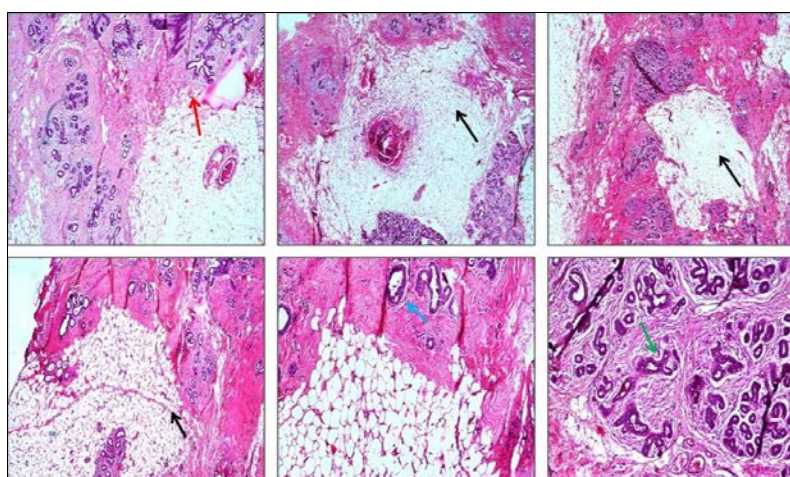


Fig 3: Picture collage of histopathology sections showing, disorganised TDLU (red arrow), interspersed adipose tissue (black arrow), Few cystically dilated ducts showing focally apocrine metaplasia (blue arrow) and epitheliosis (green arrow).

Conclusion

Breast hamartomas are uncommon benign lesions with variable imaging and histological features, posing diagnostic challenges. Accurate diagnosis requires a combination of clinical, radiological, and histopathological assessment. Excision is both diagnostic and curative. Prognosis is excellent.

Conflict of Interest

Not available

Financial Support

Not available

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