Case Report

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Pathology Section

Navigating the Diagnostic Pitfalls of Myoid Hamartoma: A Case Report

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ABSTRACT

Breast is the site for many benign and malignant conditions. Beside the common diseases of the breast, some of the rare conditions of the breast can present with overlapping clinical and radiological features posing a diagnostic challenge. Breast hamartomas are benign lesions with varying clinical, radiological and histological features. They are generally asymptomatic, but sometimes can grow into large sizes causing asymmetry of the breast. Clinical features may also vary depending upon the size and duration of the lesion. This case report details a rare case of Myoid hamartoma of the breast in a 55-year-old woman presenting with a painless gradually progressive breast lump for seven years. Initial clinical evaluation and imaging studies suggested the diagnosis of Phyllodes tumour. Histopathological and immunohistochemistry examination of the excised lesion helped in establishing the definitive diagnosis of myoid hamartoma. Due to the rarity of this lesion, diagnosis can be delayed due to its overlapping features with other lesions of the breast. Excision of the lesion is considered curative and histopathological examination can prevent misdiagnosis and over treatment. Thus, this case underscores the diagnostic challenge posed by lesions that exhibit suspicious radiological features but benign pathological origins. Documenting this case reinforces the necessity of multidisciplinary approach in diagnosing rare entities.

Keywords: Benign, Breast lump, Smooth muscle

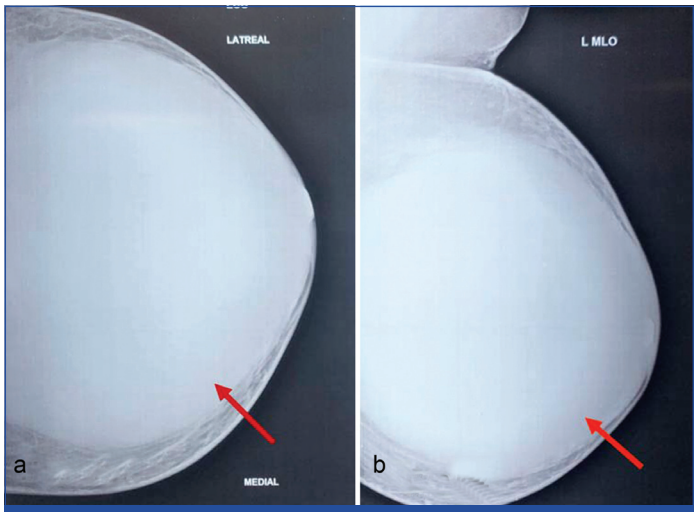
CASE REPORT

A 55-year-old woman presented to the Outpatient Department (OPD) with complaints of gradually progressing lump in the left breast over a period of seven years not associated with pain with no history of swelling in the axilla or neck. There was no history of trauma to left breast, nipple discharge or ulcers, no history of lump in the contralateral breast, and no known co-morbid conditions. She had no history of loss of weight and appetite. She is a multiparous woman with four full-term normal vaginal delivery. She attained menopause 10 years back. There was no history of hormone replacement therapy. She had no prior history of breast disease or family history of breast cancer.

General examination was normal. On local examination, nipple areolar complex and infra mammary crease appeared to be lower. Breast asymmetry was noted with the left breast appearing enlarged. On palpation, predominantly a firm mass measuring approximately around 10x8 cm was noted occupying all quadrants of the left breast. Skin was free and not fixed to underlying muscle, chest wall. No nipple discharge was present. Contralateral breast examination was normal and bilateral axilla was free.

Mammogram showed a well-defined mass measuring 15x10 cm with breast in breast appearance occupying all the four quadrants of the left breast with solid cystic components [Table/Fig-1a,b]. Imaging differentials were Phyllodes tumour, Breast Imaging-Reporting and Data System (BI-RADS) III probably benign. Based on clinical and radiological findings, clinical diagnosis of Phyllodes tumour was made.

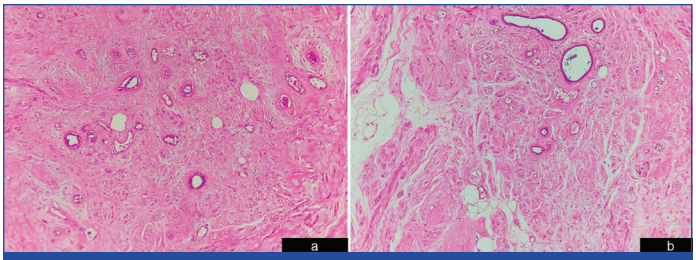
Following this, core needle biopsy was done and was suggestive of stromal predominant lesion. The patient underwent a left simple mastectomy with level I nodal sampling. Grossly, a single grey white, firm circumscribed lesion measuring 14.5x10.0x7.5 cm was seen occupying the central and inner quadrants of the left breast with grey red haemorrhagic areas [Table/Fig-2]. Microscopy examination revealed a well circumscribed lesion composed of ducts, lobules and adipose tissue in varying proportions with



[Table/Fig-1]: Mammogram of left breast: a) Cranio-caudal view; and b) Medio-lateral oblique view showing a huge well defined mass occupying all four quadrants of left breast measuring 15x10 cm (arrow).

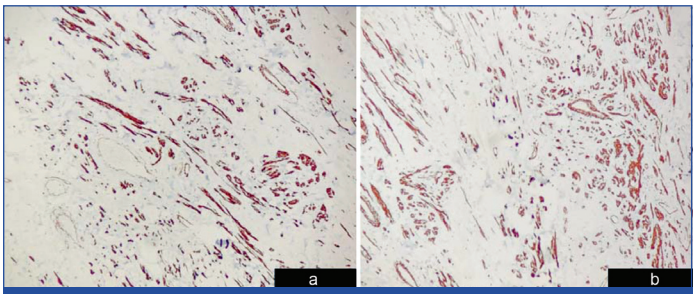
[Table/Fig-2]: Gross image depicting cut surface of left mastectomy showing a circumscribed lesion with haemorrhagic areas (arrow).

prominent stromal components [Table/Fig-3a,b]. Adjacent breast parenchyma appeared unremarkable. No evidence of increased mitosis, cytological atypia and areas of necrosis was noted. Fifteen lymph nodes examined showed reactive hyperplasia. Based on histopathological findings, differential diagnosis of hamartoma was considered and immunohistochemistry was carried to confirm the diagnosis and also to rule out the possibility of phyllodes tumour with lipomatous component.



[Table/Fig-3]: a) Showing predominantly smooth muscle components with entrapped ducts (H&E, 40×); b) Showing ducts surrounded predominantly by smooth muscle component and few adipocytes (H&E, 100×).

In immunohistochemistry, the lesional cells were strongly positive for desmin [Table/Fig-4a] and Smooth Muscle Actin (SMA) [Table/Fig-4b] in the smooth muscle fibers. Ki-67 labelling index was 2% and beta catenin was negative. CD34 highlighted the blood vessels. Histomorphological features combined with immunohistochemistry examination showing SMA and Desmin positivity, low Ki-67 index and CD34 highlighting only blood vessels supported the diagnosis of myoid hamartoma. Furthermore, the lesion lacked cytological atypia, infiltrative borders and significant mitotic activity, thereby reducing the likelihood of malignant spindle cell lesions. Definitive diagnosis of myoid hamartoma was established with the above histopathological and immunohistochemistry features. Postoperative period was uneventful. No recurrence was reported in the two years period following surgery.



[Table/Fig-4]: a) Showing smooth muscle cells with positive staining for Desmin (IHC,100×) b) Showing Smooth muscle cells with positive staining for Smooth Muscle Actin (SMA) (IHC,100×).

DISCUSSION

Hamartomas are masses of disorganised tissue composed of cells pertaining to their site of origin. The term 'Hamartoma' was first used by Eugen Albrecht in 1904 to describe a tumour like lesion. Hamartoma is derived from the Greek word 'hamartia' which denotes 'fault' or 'defect'. They can virtually occur in all organ systems starting from head to toe. Syndrome associations like Cowden syndrome have been established in few cases, especially when they involve bilateral breasts and occur at multiple sites [1].

Breast hamartomas account for a very small number of benign breast lesions and were first described in the year 1968 by Hogeman KE and Ostberg G [2]. The specific term 'Hamartoma' was used by Arrigoni MG et al., in 1971 [3]. Incidence of mammary hamartoma ranges from 0.7 to 4.8% [4]. Myoid hamartomas show wide variations in age, occurring any time after puberty. Hamartomas are reported to occur in the postmenopausal age group and persist for a few years similar to the present case. The condition predominantly affects women in their fifth decade of life [4], aligning with the typical age range for most benign breast lesions, thereby causing delay in diagnosis. They are also known to occur in teenagers and also above the age of 60 years. Very rarely myoid hamartomas were reported in male and are usually diagnosed in men over 35 years of age [5]. To date, there is only one documented clinical case involving a 13-year-old boy, with an unusual intrathoracic extension [6].

Clinically, they can present as a slow growing painless lump involving predominantly the outer quadrant of the breast. The prolonged seven years clinical history with progressive growth in the present case highlights slow growing nature of myoid hamartoma. Smaller size lesions can be asymptomatic and are detected on routine examination. Most of the reported cases of breast hamartomas is usually smaller, ranging in size between 2-5 cm [4]. Lesions measuring more than 10 cm are less common and the present lesion measured approximately 14.5 cm on gross examination, causing breast asymmetry and inferior displacement of nipple areolar complex on clinical examination [7,8]. Based on the available literature, this appears to be one of the largest reported myoid hamartomas of the breast.

Imaging features of hamartomas are variable due to the mixture of fibrous, fatty, and glandular elements. On imaging they can appear as a well circumscribed round or oval masses with a surrounding thin capsule on mammograms. The classic term used on imaging for breast hamartoma is 'breast within a breast' appearance. When the stromal component predominates, it can mask the characteristic "breast-within-a-breast" appearance of a hamartoma, leading to a misdiagnosis. Differential diagnoses for such cases include fibroadenoma and phyllodes tumour distinguished by their lack of internal fat, as well as lipoma which is characterised by a homogeneous fatty composition without significant soft-tissue elements [9]. The radiological impression of phyllodes tumour in the present case underscores the diagnostic pitfall in large size myoid hamartoma. Although a partial "breast within a breast" was noted, the classic imaging feature of myoid hamartoma was not fully appreciable due to marked stromal predominance and large size of the lesion.

Adequate core biopsy volumes are essential for accurate diagnosis of myoid hamartoma, as its spindle and epithelial components can closely resemble phyllodes tumour on limited sampling. However, definitive diagnosis on core biopsy is challenging due to its overlapping features with other breast lesions [10]. This case underscores the challenges of relying on core needle biopsy on diagnosing rare lesions of the breast. Initial core needle biopsy showed a stromal predominant lesion pointing towards fibroepithelial lesion and diagnosis of myoid hamartoma was established only after histopathological examination of resection specimen. Because small biopsy can miss the co-existence of other components like epithelial, smooth muscle and adipose tissue and diagnosis on small biopsy also depends on the sampled region. There are several variants of mammary hamartoma depending on the composition of glandular and fibroadipose elements. Some of the variants are adenolipoma, fibroadenolipoma, and myoid hamartoma [11].

Myoid hamartoma is a rare subgroup of hamartoma arising from the breast with predominance of smooth muscle components. Their origin is still debated with few proposed theories. The prominent spindle cell component reflects leiomyomatous metaplasia of myoepithelial cells, typically occurring within a background of adenosis [12]. On the contrary, another theory suggests that myoid hamartoma probably arises from multipotent stromal mesenchymal cells which differentiate into smooth muscle, adipocytes or even cartilage [13]. HMGA2 rearrangement, common in other benign mesenchymal tumours, supports a shared pathogenesis potentially from mutated mesenchymal stem cells [14].

On histopathological examination, they are composed of differentiated glandular, stromal and fatty tissue with areas of smooth muscle cells predominance from which its name originates. Myoid hamartoma can present with classical benign circumscribed lesions similar to

fibroadenoma to irregular lesions suspicious of malignancy. Myoid hamartoma with focal myxoid changes, cartilaginous differentiation and pseudoangiomatous stromal hyperplasia are also documented in the literature [15]. Immunohistochemical markers show positive staining for SMA, desmin and vimentin and also help to confirm the diagnosis. As outlined by World Health Organisation (WHO) classification of breast tumours, essential criteria to diagnose hamartoma include the presence of components of normal breast tissue along with clinical and imaging features [16].

Differential diagnosis includes fibroadenoma (with smooth muscle metaplasia), leiomyoma, adenomyoepithelioma, fibromatosis, granular cell tumour, and malignancies like leiomyosarcoma or metaplastic carcinoma [17]. Definitive distinction relies on histopathological examination showing disorganised glandular, fat and prominent smooth muscle component without pericanalicular patterns, plus absence of atypia or mitoses.

Till date no documented cases exist of infiltrating carcinoma specifically arising within myoid hamartoma of the breast. A few breast hamartoma cases (not specifically myoid) have co-existed with malignancies like invasive ductal carcinoma or ductal carcinoma in situ, but these appear incidental rather than transformative [18,19]. Hamartomas are benign and treatment depends on the size and clinical symptoms. Surgical excision remains the standard treatment for myoid hamartoma of the breast, ensuring complete removal [13]. Recurrence in myoid hamartoma of the breast is extremely rare, with only a handful of documented cases worldwide Literature identifies two cases with recurrence before 2026 [20,21].

CONCLUSION(S)

The present case highlights that in spite of indolent biological nature of myoid hamartoma, it can present as a large breast lesion with longstanding history, clinically and radiologically simulating other fibropeithelial lesion causing diagnostic challenge. Given the rarity of myoid hamartoma, awareness of this entity is important for clinicians, radiologists and pathologists to avoid any misdiagnosis. Histopathological examination plays a crucial role in diagnostically challenging cases and helps to avoid misinterpretation.

This case report suggests that owing to the complex and overlapping features in clinical and radiological presentation of rare lesions of the breast, systematic approach combining clinical features, radiological findings followed by histopathological examination with immunohistochemistry is essential to arrive at the definitive diagnosis.

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