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# Post Lactation Giant Breast Hamartoma: Challenges of Diagnosis and Surgical Management



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## Introduction

- Breast hamartomas are rare, benign breast tumors composed of varying amounts of fibrotic, fatty, and glandular elements.
- They are also named fibroadenolipoma, adenolipoma, and lipofibroadenoma depending on the predominant tissue composition.
- Rare subtypes include myoid, chondroid and psuedoangiomatous stroma.
- The incidence is 0.1 - 0.7%, but thought to be higher as 75% of breast hamartomas are asymptomatic and 66% are not palpable.
- Incidence is also likely higher because their histological composition, clinical presentation, and appearance on imaging are often similar to a fibroadenoma.
- Comprise 4.8% of all benign breast lesions and can present in women from 24 to 76 years old, but usually in women over 35
- It is important to accurately diagnose breast hamartomas because of its malignant potential.
- Most common symptoms include a painless, palpable, mobile mass that is circumscribed and often round to oval shaped grossly and on imaging.
- Usually slow growing, but can lead to significant breast asymmetry as in our patient.
- Exact cause is unknown, but it is thought to arise from dysgenesis and is not a true tumor.
- Can occur as a single tumor or in association with Cowden or Bannayan-Riley-Ruvalcaba syndrome, which can lead to the development of multiple hamartomas in various organ systems.
- Usually found incidentally on screening mammography, but often require tissue biopsy for confirmation.
- Liposarcomas should be considered on the imaging differential because they are malignant and have metastatic potential

## Case Description

Our patient is a 38 year old, premenopausal African American woman with a past medical history of bilateral fibroadenomas with subsequent surgical excisions. She delivered her first child and breastfed at the age of 35. Shortly after giving birth, the patient noticed an enlarging right breast lesion, prompting mammogram and US evaluation. Imaging studies at this time did not visualize an abnormality, but noted increased breast density consistent with breastfeeding. The patient was recommended additional imaging with contrast enhanced spectral mammography (CESM), but initially declined. Eight months later, the patient elected to proceed with CESM and was not breastfeeding at this time. The CESM findings demonstrated a large circumscribed fat density mass in the right lower outer breast measuring approximately 13.1 cm and consistent with a benign breast lipoma. The patient underwent a second bilateral breast ultrasound revealing a circumscribed oval isoechoic mass in the right lower outer quadrant measuring 3.9 x 10.1 cm, consistent with the sonographic appearance of a lipoma. Annual mammography was recommended.

**Imaging:** Follow-up mammographic imaging was now significant for a large encapsulated lucent mass encompassing the entire outer central right breast and consistent once again with a breast lipoma. The patient had no further workup at this time. She presented to the breast surgical oncology department nine months later after noting a rapidly enlarging mass in her right breast. She complained of right breast tenderness, denied erythema, thinning of skin, spontaneous nipple discharge, or nipple inversion. She also denied a history of high risk breast lesions. Age at first parity was 35, she breastfed for 12 months, and was taking oral progesterone for contraceptives. She denied a history of fertility treatments or hormone replacement therapy.

**Physical exam:** Her breasts were notably asymmetric. The right breast measured a 38DD and the left was a 34B with no evidence of skin retraction, erythema, scaling, or nipple inversion. Milky discharge was expressed from the right breast and a mass spanning the 6:00 to 10:00 position was palpated as approximately 12 x 12 cm. At the 6:00 and 8:00 position, the edges were notable for small nodules. The mass was firm, mobile, and not fixed to the underlying chest wall or tethered. There was no cervical, supraclavicular, infraclavicular, or axillary lymphadenopathy bilaterally.

**Follow up imaging:** A bilateral MRI of the breast with and without contrast confirmed presence of a circumscribed enhancing 12.3 x 6.9 x 8.7 cm mass in the upper outer and lower outer quadrant of the right breast, anterior, middle, and posterior thirds. This mass was thought to be most consistent with a hamartoma. There was also a 6mm enhancing mass in the right breast at the 3:00 position, 7 cm from the nipple that was recommended targeted US evaluation and biopsy. The left breast was unremarkable and there was no axillary or internal mammary lymphadenopathy.

A targeted breast ultrasound was performed of the right subareolar breast at the 3:00 position. The biopsy showed cores of benign adipose tissue and breast tissue, no malignancy or atypia was noted. The pathologist's histologic differential diagnosis included a hamartoma, lipoma, and non-lesional breast tissue.

**Histopathology:** Given the patient's asymmetry, pain and rapidly growing mass, she was recommended right breast surgical excision with immediate reconstruction with breast and plastic surgery. On final pathology, the specimen measured 15x13x4 cm, which contained benign, circumscribed, encapsulated, lobulated yellow adipose tissue throughout, no breast tissue or densities identified grossly. The final diagnosis was a "hamartoma-primarily composed of adipose tissue with scattered foci of benign breast tissue."

## Discussion

### Imaging

-Our patient's breast hamartoma was not detected with traditional imaging while lactating, making a somewhat difficult diagnosis more challenging. This led to the use of uncommon imaging modalities to detect the breast hamartoma when she presented after lactation cessation (CESM and MRI with and without contrast).

-The increased lobules and ducts during lactation increase breast density and can obscure masses. Ultrasound and mammography did not initially detect our patient's mass.

-CESM was the first imaging study to detect a mass in our patient and accurately identified the predominantly fatty tissue composition of the mass. CESM uses Full Field Digital Mammography with IV contrast, which is specifically useful in the diagnosis of this patient because it enhances the sensitivity for detecting breast cancer and suspicious masses, especially in patients with heterogeneously and extremely dense breasts. CESM may be considered in the diagnostic workup of patients who are lactating or have dense breasts, especially in women with inconclusive screening on mammogram and ultrasound, such as our patient.

-MRI with and without contrast was also helpful in leading to a non-invasive and accurate diagnosis of a breast hamartoma. MRI with and without contrast may also be considered in the evaluation of post-partum breast masses, especially if previous imaging is indeterminate.

### Surgical Challenges

-Our patient developed rapid growth of her right breast after cessation of lactation. Rapid and disfiguring growth is concerning for a malignant mass with possible metastatic potential and the mass created discomfort and visible asymmetry. Excisional biopsy is diagnostic and therapeutic, but was complicated by the need for plastics reconstruction because of the surgical challenges created by the asymmetry.

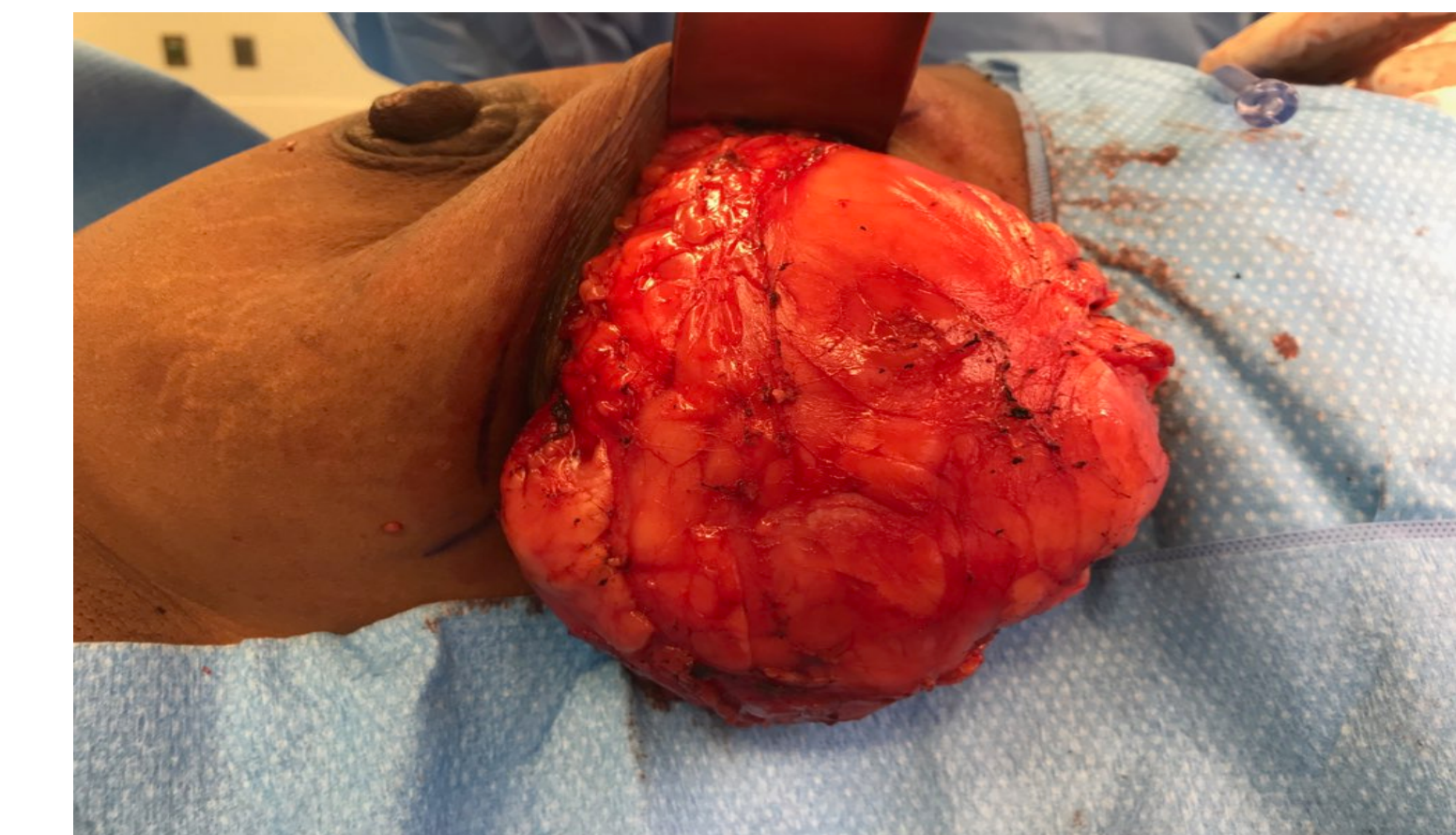
-Oncoplastic breast surgery was required to ensure adequate symmetry and margins. Multiple techniques can be used. Local tissue rearrangement has taken favor as it involves moving subcutaneous and glandular breast tissue to fill in the defect created by tumor excision.

### Hormonal Influences

-Lactation cessation may be implicated in the rapid growth of our patient's hamartoma because the majority of breast hamartomas are composed of epithelial and stromal cells, which have been shown to contain estrogen and progesterone receptors. It is known that breast hamartoma cells are influenced by estrogen and progesterone just as the epithelial cells comprising normal breast tissue. Cowden syndrome results from a PTEN mutation and a similar pathophysiological mechanism may be involved in the development of breast hamartomas as well. PTEN is a tumor suppressor gene that inhibits PI3K/AKT signaling, preventing estrogen dependent gene expression. Without PTEN suppression of PI3K/AKT, epithelial and endometrial cells reliant on estrogen receptor trophic signal can grow uncontrollably. This theory may also lead to consideration of alternative treatments to shrink or delay growth of rapidly enlarging breast hamartomas with medications such as oral contraceptives.

## Conclusion

Our goal is to inform clinicians about the challenges of diagnosing breast hamartomas in pregnant and lactating women, as well as women with dense breasts. It is appropriate to follow one's clinical suspicion with imaging and to consider CESM and MRI with and without contrast in lactating women and women with dense breasts as these imaging modalities allowed us to come to a more accurate diagnosis in this case. Although our patient did not get the recommended CESM until after she stopped breastfeeding, the literature on CESM suggest that it's sensitivity for detecting masses in heterogeneously and extremely dense breasts may have been able to detect this mass earlier, preventing rapid enlargement and the need for plastics reconstruction. Breast hamartomas should be on the differential for post-partum breast masses as pregnancy and lactation may affect the growth of hamartoma cells, which is the similar to normal breast tissue. It is known that the majority of breast hamartomas are benign, but obtaining a diagnosis with advanced imaging and histopathology in a timely manner is paramount. Consequences of delayed diagnosis include potential for malignant transformation, metastatic disease, and rapid growth involving significant breast asymmetry requiring plastic surgery reconstruction.



(FIGURE 1)  
Breast hamartoma before complete excision.



(FIGURE 2)  
Breast hamartoma removed from the right upper and lower quadrant of the breast.



(FIGURE 3)  
Gross specimen of breast hamartoma, primarily composed of adipose tissue with scattered foci of benign breast tissue.

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