Cytological Diagnosis of Epidermal Inclusion Cyst of Breast: A Rare Case Series of Four Cases with Review of Literature

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Abstract: An epidermal inclusion cyst (EIC) arising from the breast is a rare, benign condition that may potentially be malignant. EICs of the breast often presents in the fifth decades of life and is primarily localized in the periareolar region. We report four cases of EIC of the breast, three women in their forties and one in twenties. The diagnostic accuracy needs correlation between sonographic, mammographic findings along with cytohistological features. Fine-needle aspiration cytology is essential for accurate preoperative diagnosis. Excision is usually recommended to confirm and avoid potential risks of infection and malignant transformation. Till now, less than 100 cases have been reported in English literature. The present study highlights not only its rare site of presentation, but also the role of FNAC in diagnosing this benign lesion.

Keywords: Breast lump, cytomorphology, epidermal inclusion cyst.

I. Introduction

An epidermal inclusion cyst (EIC) of the breast is a rare condition that develops due to the proliferation and implantation of epidermal elements within a circumscribed space in the dermis. This benign entity of breast is very rare with few case/case series is available in the English literature.

Most common location of the EICs are on the face, trunk, neck, extremities and scalp in decreasing order of frequency.¹¹ Rare cases of EICs reported in literature are of bone, breast and intracranial locations.⁵ EICs of the breast often seen in females in their fifth decade of life.³ It presents as breast lump mainly in periareolar region and needs to be differentiated from other neoplastic and non-neoplastic breast lesions.⁴ Diagnosis is straightforward for epidermal inclusion cysts that occurs in the breast subcutis as a small nodule, but enlarged cysts in the breast parenchyma requires to be differentiated from malignant or benign tumors of the breast.¹¹ Fine needle aspiration cytology (FNAC) is the best diagnostic tool for the evaluation of palpable breast lumps. Excision is usually recommended for definitive histopathological diagnosis.

II. Case Series

Table I: shows clinicopathological profile of all four cases. Cytological smear of all four cases revealed presence of numerous anucleate and nucleate squames in clusters as well as scattered singly.

Case number 1 – Ultrasound (USG) guided FNA was done and USG revealed a well defined lobulated oval heterogeneously hypoechoic lesion of size 54x37x19 mm at 8-10 o’clock position in paraareolar region with posterior acoustic enhancement. (fig. 1a) Mammography showed a well defined lobulated hyperdense lesion of size 52x33x22 mm seen in supero-lateral quadrant in mammary and premammary space reaching almost up to the skin surface, however overlying skin appears normal.(fig. 1b) Cytology revealed numerous anucleate squames.(fig. 1c)

Case number 2 and 4 also showed presence of numerous neutrophils and macrophages along with squames, hence reported as infected epidermal inclusion cyst. Case number 3 in addition to nucleate and anucleate squames, neutrophils, macrophages, fair number of foreign body giant cells is also seen, reported as infected ruptured epidermal inclusion cyst. (fig. 2) All four cases were confirmed on histopathology also.

III. Discussion

An epidermal inclusion cyst (EIC) is lined by stratified squamous epithelium that contains a distinct granular layer and lamellated keratin without calcification. Most frequent location of EICs are on the face, scalp, neck and trunk.¹¹ EICs of breast are rare benign entity.¹³,⁴,⁵,⁶,⁷ First case of EIC of breast on histopathology was reported in December 1900 at The Johns Hopkins Hospital at Baltimore, USA.⁶ It may develop at any age but most commonly found in the fourth and fifth decades of life.¹³

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Clinically, EICs developing in the skin of the head, neck and back areas presents as skin protrusions because of firm composition of subcutaneous tissues in those regions. On the other hand, breast EICs rarely presents as skin protrusion as the subcutis of breast contains flexible fat and mammary gland tissues that lack firmness.

Several hypothesis have been postulated regarding its development: (I) a genetic anomaly that arises from small nests of cells that left during the embryonic development. (II) implantation of the torn fragments of epidermis within the breast tissues which may results from trauma, reduction mammoplasty and needle biopsy. (III) squamous metaplasia of columnar cells of an ectatic duct in an area of fibroadenoma or fibrocystic changes or phyllodes tumour. (IV) may arises from obstructed hair follicles.

On mammography, EIC typically appears as a well circumscribed, noncalcified homogenous density and thus is indistinguishable from carcinoma breast having benign features. On ultrasonography, EICs have a solid, well circumscribed and complex or heterogenous appearance. The specific sonographic features of breast EIC is onion- ring appearance with alternating concentric hyperechoic and hypoechoic rings as described by Crystal and Shaco-Levy. These concentric rings corresponds to the multiple layers of the lamellated keratin. Denison et al reported specific sonographic features of breast EIC as its extension into the dermis. On Computed tomography scan, EIC appears as a lucent lobulated mass. MRI reveals an isointense lesion.

EICs may rupture and lead to serious complications. Inflammation is mediated by the horny material contained in the EICs and the material have chemotactic properties for polymorphs. Rupture may be associated with secondary foreign body reactions, granulomatous reactions, or abscess formation. The spontaneous rupture have been reported in ten cases.

Although EICs are benign, rarely they may undergo malignant transformation. In a study by Menville et al, 19% of the patients with EICs showed malignant squamous cell lining on histopathological examination. Cameron and Hilsinger reported malignant transformation in a very small number of cases (0.045%). The association between EICs of the breast and malignancy reported is 12%. EICs of the breast undergo malignant transformation more frequently as compared to other sites because of the fact that the pathogenesis behind the EICs of the breast related to the squamous metaplasia of the mammary duct epithelium. Furthermore, a significant correlation between size of the cyst and malignant transformation was studied.

Differential diagnoses considered for presence of cyst inside breast parenchyma are fibroadenoma, phyllodes tumor, fibrocystic disease with squamous metaplasia, and malignant breast neoplasm with benign features such as mucinous carcinoma. Asymptomatic small sized lesions (<2 cm) do not require any treatment but large and palpable lesions require surgical excision as as emphasized by Lam et al.

IV. Conclusion

Epidermal inclusion cyst is a rare benign lesion of breast. Clinically and radiologically these lesions are often mistaken as benign or malignant tumors. Preoperative FNAC is confirmatory in the presence of a typical pultaceous aspirate and cytomorphological features of EIC. Thus, FNAC plays a crucial role in its diagnosis and management. Symptomatic cases should be readily excised and need histological correlation to rule out any potential complications that can arise in these cysts.

References

Table I: Clinicopathological profile of the epidermal inclusion cyst in the breast

<table>
<thead>
<tr>
<th>Case Number</th>
<th>Age/Sex</th>
<th>Clinical Presentation</th>
<th>Imaging Findings</th>
<th>FNAC Diagnosis</th>
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</table>
| 1           | 50 Y/F   | Single, mobile, non-tender, well defined lump of size 6x6 cm in upper outer quadrant of right breast since 6 years | USG - well defined lobulated oval heterogeneously hypoechoic lesion of size 54x37x19 mm at 8-10 o’clock position in para-areolar region with posterior acoustic enhancement.  
Elastography - mosaic pattern of green, blue and red (Q value – 1.8)  
Mammography - well defined lobulated hyperdense lesion of size 52x 33x 22 mm seen in supero-lateral quadrant in mammary and premammary space reaching almost upto the skin surface  
Radiological Diagnosis – Fibroadenoma / Complicated cyst. | Epidermal inclusion cyst (EIC) |
| 2           | 47 Y/F   | Single, well defined, firm, mobile and painful lump of size 2 x 1cm in lower outer quadrant of right breast since 2 years. | USG - hypoechoic breast lesion with central necrosis of size 2x1cm at 6 o’clock position.  
Radiological Diagnosis – Benign breast lump. | Infected EIC |
| 3           | 40 Y/F   | Single well defined, firm, painless lump of size 1 x 1 cm in lower inner quadrant with presence of punctum on the overlying skin. | Mammography - well circumscribed, rounded soft tissue density lesion close to the skin surface at 3 o’clock position.  
Radiological Diagnosis – Sebaceous cyst | Ruptured EIC |
| 4           | 28 Y /F  | Single well defined, firm, mobile, non-tender, lump of size 2 x 2 cm in lower inner quadrant of right breast since 4 months. | USG - circumscribed, homogenous, oval hypoechoic lesion at 4 o’clock position. No increased vascularity was observed.  
Radiological Diagnosis - Fibroadenoma | Infected EIC |

Fig. 1a- USG showed a well defined lobulated oval heterogeneously hypoechoic lesion of size 54x37x19 mm at 8-10 o’clock position  
Fig. 1b – Digital Mammography craniocaudal view showing a well defined lobulated hyperdense lesion of size 52x 33x 22 mm seen in supero-lateral quadrant in mammary and premammary space  
Fig. 1c – Photomicrograph showing cluster of anucleate squames (MGG X 10)
Fig. 2 - Photomicrograph showing cluster of anucleate squames along with presence of numerous neutrophils.