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Occult Primary Breast Carcinoma: A Rare Case Report with Review of Literature

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Abstract: ***Occult primary breast carcinoma presenting as axillary lymphadenopathy is a very rare entity. Owing to the absence of primary tumor, diagnosis and treatment of such cases is a real challenge for oncologists. The histopathological examination and immunohistochemistry evaluation of the tissue plays a salient role in diagnosing and typing of such tumor. Here, we are reporting a case of a 38-year-old woman with occult primary breast carcinoma, discovered after initially presenting with neurological symptoms. Immunohistochemistry showed that the biopsied lymph node was positive for GATA3. Suggesting breast as the site of primary tumor.***

Keywords: Occult primary breast carcinoma; neurological symptoms, GATA3

1. Introduction

In 1907 Halsted described the Occult primary breast carcinoma for the first time. By definition it's the histologically proven breast carcinoma, which is discovered outside the breast in the absence of primary breast tumor. Occult primary breast cancer presenting as axillary metastasis is very rare, accounting for less than 1% of all breast cancers, having a peak incidence at the age of 55. Axillary metastasis without an obvious primary tumor remains a difficult diagnostic and therapeutic challenge. Here, we report a case of occult primary breast cancer with a unique initial presentation, and we review the currently available literature of this rare entity. [2], [3]

2. Case Report

A 38 year old female came with the complaints of generalized weakness and imbalance while walking since two months. Clinical examination revealed ataxia and dysdiadochokinesia, the deep tendon reflexes were preserved. Head to toe physical examination revealed a large right axillary palpable mass approximately measuring 6cm in diameter. Tru-cut biopsy was performed from the right axillary mass and was sent for histopathological examination. Biopsy from the axillary mass revealed a tumor mass arranged in sheets, cords and trabeculae. Individual tumor cells were round to oval with increased N:C ratio, vesicular nuclei and prominent nucleoli (Figure1).

A histopathological diagnosis of poorly differentiated epithelial malignancy was made and IHC was advised for further confirmation and typing.

Further, GATA 3 for confirming that the axillary tumor mass is originally a breast tissue, along with ER, PR and HER2neu, was performed on the tissue biopsied from the right axillary mass.The IHC results were as follows:

1) GATA-3: Positive (Figure 2)
2) ER: Negative
3) PR: Negative
4) HER2neu:Positive,3+(complete membrane staining)

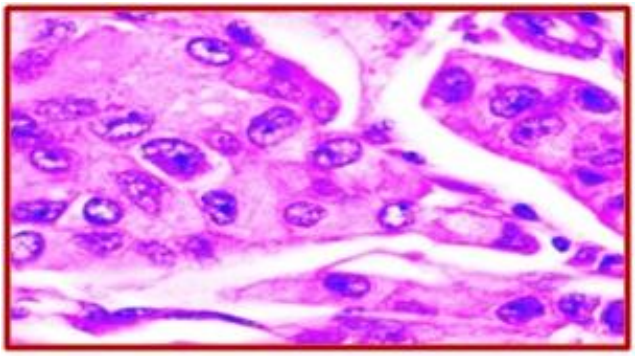

Figure 1: H&E 400x.

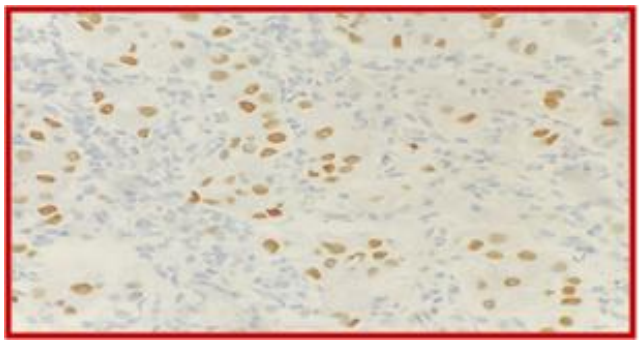

Figure 2: (IHC 400x) GATA3 showing nuclear positivity.

Then, PET scan was performed which revealed non FDG avid subtle enhancing soft tissue in the central quadrant of the right breast parenchyma measuring 1.7x1.3cm. Increased FDG uptake in the necrotic right axillary nodal mass measuring 3x3.3cm.

Cerebellar ataxia due to paraneoplastic syndrome with source being the malignant right axillary lymph node was taken into consideration. Clinically, there was no palpable breast lump. A right modified radical mastectomy was performed and the specimen was again sent for histopathological examination.

We received two labelled containers. Container1 labelled as "Right breast" consisted of right modified radical mastectomy specimen measuring 24x13x2.5cm. On serial sectioning, few grey-white, firm, and elastotic areas were seen. No definitive

tumor mass was identified in the mastectomy specimen. 15 lymph nodes were isolated, out of which largest lymph node measured 6cm and smallest measured 0.5cm in diameter respectively.

Container 2 labelled as "Axillary lymph nodes" consisted of multiple fibrofatty tissue altogether measuring 6x2x1cm. No lymph nodes were identified.

On microscopy sections from grey-white areas of the mastectomy specimen showed features of sclerosing adenosis.

Section studied from the largest axillary lymph node mass showed a malignant tumor with similar morphology as was seen in the axillary biopsy specimen.

Positive GATA3 & HER2neu 3+ on IHC favours the diagnosis of:

Occult poorly differentiated invasive Breast Carcinoma (No Special type) with Axillary lymph node Metastasis.

Modified RB Score (3+3+2=8) Grade III
Pathological Staging- pT0N1a

Henceforth, a Final diagnosis of Occult Poorly Differentiated Invasive Breast Cancer with Metastasis to Ipsilateral Axillary Lymph Node was made.

Significant neurological improvement was noted in the patient, after the mastectomy was done.

3. Discussion

Approximately <1 % of all breast cancers is occult, that is, patients present with regional or distant metastasis (usually axillary lymphadenopathy) which is histologically consistent with a primary breast cancer, but clinical and mammographic evaluation of the breast does not demonstrate a tumor. The other malignant neoplasm which are known to metastasize to the axillary nodes include melanomas and carcinomas of lung, thyroid, stomach, colon, rectum, pancreas and ovaries. However, these metastases are rarely the first signs of the disease.

A large proportion of patients (20–30%) with occult primary breast cancer have a positive family history of breast cancer, although there was no positive family history in our patient.

Positron Emission Tomography (PET) has been frequently used in the detection of occult breast carcinoma and is widely used in the diagnosis of the primary tumor in such challenging cases, especially in women with radio-dense breasts. It is also said that breast scinti-mammography can be a useful, cheap and practical diagnostic tool in the evaluation of the patient with occult breast cancer, but experience with this modality is still limited. [1]

Positive results of estrogen and progesterone are suggestive of breast cancer, and this occurs in approximately 50% of cases. Regardless, negative ER/PR staining does not exclude the diagnosis of breast cancer and it is important to know that other carcinomas like cancer of the colon, ovary, endometrium, kidney and melanoma can also show detectable ER/PR positivity.[1][2][3]

GATA3 is a member of the zinc finger transcription factor family. It is involved in development and differentiation of many tissues and cell types, and is reported to be a sensitive and specific marker for urothelial and breast carcinomas in tissue sections. A role for IHC detection of GATA3 act as a valuable aid in the identification of breast carcinoma. [4]

The GCDFP-15 and mammaglobin antibody is another sensitive marker of breast carcinomas. It has been reported recently that the sensitivity of mammaglobin is better than that of GCDFP-15, but it lacks the specificity of GCDFP-15. The expression of Mammaglobin is not altered at the metastatic lymph node site and can help, in combination with other markers, to establish the correct diagnosis of metastatic breast carcinoma.[5]

The treatment of occult primary breast cancer has been inconsistent and controversial since its initial description and due to its rarity, it has been difficult to standardize the management. Hence, by far there is no clear consensus concerning optimal treatment of occult breast carcinoma.

4. Conclusion

Occult primary breast carcinoma with axillary metastases is not only rare, but is a potentially challenging entity, for making a diagnosis as well. Due to the absence of primary tumor in the breast, it becomes difficult for the clinicians to label it as "breast carcinoma", immunohistochemistry plays a significant role in such cases and helps the pathologist along with the treating physicians to know the origin of the tumor. Markers such as GATA3, GCDFP-15 and mammaglobin are of great importance in establishing the diagnosis of occult primary breast cancer. Due to the rarity of the disease, its natural history has not been documented yet. The treatment remains very contentious and requires a collaborative approach. Axillary dissection with breast conservation and ipsilateral breast radiotherapy seems to be a good therapeutic choice and can be advocated to improve local control in addition to esthetic results.

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