

Primitive Neuroectodermal Tumor (Pnet) Presenting As A Breast Mass-Case Report

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Abstract

Primitive neuroectodermal tumors (PNET) are rare, malignant, small-round-cell tumors of the bone and soft tissue that usually occur in children and young adults¹ uncommonly found within the breast tissue. In this case report, we examined a 53-year-old patient, who developed a lump on her left breast and was later diagnosed with ES/PNET. Clinical presentation, age at development and radiological findings were of interest and were discussed. Diagnosis of the tumour was confirmed using various immunohistochemical studies.

Keywords: Breast, Primitive Neuroectermal Tumour

Introduction

Ewing's sarcoma/primitive neuroectodermal tumour (ES/PNET) is a member of the Ewing's sarcoma family of tumours (ESFT). Extraskelatal ES presenting as a breast mass is unusual. ES typically occurs in adolescents and young adults aged between 10 and 20 years²

The diagnosis of ES/PNET requires panels of immunohistochemical study and the presence of a t(11;22) translocation detected through fluorescent in situ hybridization (FISH). In This case report examines a rare case of ES/PNET observed in a 53-year-old woman, who

developed a painless and progressive tumour presenting as a breast mass. The patient's family provided their consent to the study.

Case Report

We report here a case of a 53-year-old woman presented a painless and progressive mass in her left breast for 6 month. She had loss of weight with lethargy and backache. On examination, the entire left breast was erythematous, indurated with multiple fixed axillary lumps. She was anaemic There were no other lesions arising from the trunk or limbs.

Bilateral mammography was performed and showed an ill-defined, isodense mass deep in the left upper inner quadrant, without evidence of microcalcifications . Sonography revealed oval, homogeneously low echoic masses of 12x10 cm size was identified with posterior acoustic enhancement.

CT scan of thorax, abdomen and pelvis revealed left breast tumour which was lobulated and heterogeneously enhancing, with poor plane of demarcation between the tumour, skin and anterior chest wall muscles. Multiple large mediastinal and axillary lymphadenopathies were seen. Small right lung nodules wash present with

thickening of pleura. No liver lesion seen. No other bone or suspicious soft tissue lesions were seen on the axial skeleton and trunk .

On cycological examination we founded singly scattered cells and poorly cohesive clusters with lymphocytes and mitotic figures in background. Cell having round to oval nuclei high N:C ratio and abundant vacuolated cytoplasm. (Fig-1)

Core biopsy sample, using trucut needle, was taken from the left breast. Histopathological interpretation from the breast lesion biopsy was reported as presence of malignant small round blue cells, arranged in solid sheet with area of necrosis. Cells were mildly pleomorphic, moderate eosinophilic cytoplasm, round nuclei and small inconspicuous nucleoli.(fig-2)

By immunohistochemical staining, the tumor cells were strongly positive for vimentin, CD99 (Fig. 1F), and FLI-1 but were negative for cytokeratin, leukocyte common antigen, synaptophysin, chromogranin, CD56, desmin, and myoD1.(Fig-3)

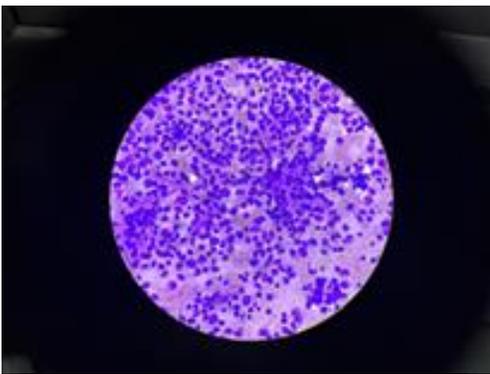


Figure 1: Cytology shows singly scattered cells and poorly cohesive clusters with lymphocytes and mitotic figures in background. Cell having round to oval nuclei high N:C ratio and abundant vacuolated cytoplasm.

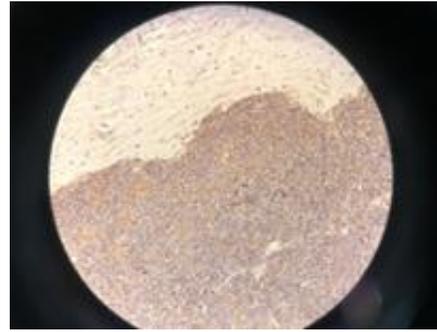


Figure 2: Microscopically, tumor is composed of small, round cells with inconspicuous nucleoli and moderate amount of cytoplasm, which are arranged in sheets or solid nests (Hematoxylin & Eosin staining, ×200).

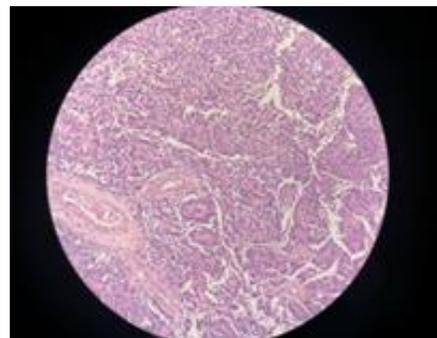


Figure 3: On immunohistochemical staining, tumor cells show strong membranous immunoreactivity for CD99.

Discussion

We report a rare case of extraskeletal ES/PNET, presenting with a rapidly growing palpable breast lump. Age at presentation, radiological findings and immunohistochemical findings were of interest and documented in this report.

ES and PNET are typically undifferentiated³ Translocation t(11;22)(q24;q12) resulting in EWS/FLI1 fusion, which can be identified in more than 90% of ES/PNET cases, is the genetic hallmark of ES/PNET⁴. In cases with classic morphology where other small round cell neoplasms have been excluded immunohistochemically, the expression of CD99 cell surface antigen (product of the MIC-2 gene) is required to support ES/PNET diagnosis.⁵⁻⁶

The majority of patients with ES/PNET are 10-20 years old², and other small studies of adult ES/PNET from the Royal Marsden, the Memorial Sloan-Kettering and the Dana-Faber Cancer Centers have reported a median age of 24-27 years^{2,7-8}. Popli et al reported a 14-year-old girl with unilateral localised primary Ewing's sarcoma, which to the best of our knowledge, is the youngest reported so far.⁹ However, our patient was 53 years of age at the time of diagnosis, which is unusual.

ES/PNET development within breast tissue was unlikely to be diagnosed upon first presentation. Findings from mammography and ultrasonography breast images may vary as they could be from a hypoechoic mass with posterior enhancement or heterogeneous mass with a necrotic area^{10,11}. In this patient, a well-circumscribed mass was documented, which possibly arose from the soft tissue beneath the breast. Precise identification of the tumour location (chest wall soft tissue) and recognition of variation in radiographic findings described in this tumour may be a clue for radiologists to avoid misinterpreting the tumour as a breast mass that might preclude the diagnosis of soft tissue tumours.

ES/PNET is an aggressive tumour with a high incidence of local recurrence and distant metastasis. The choice of treatment of PNET depends on the size of tumor, faraway metastasis, age and general conditions of the patients. A combination of multiple modalities, including surgery, chemotherapy and radiation therapy, was the most appropriate treatment for our patient². All members of the ESFT tend to share the propensity for metastatic spread. Overall the survival rate for 5 years was 38% whereas 5 year survival rate for localised disease was 49%. Consistent use of systemic chemotherapy to treat localised ESFT effectively improved the 5-year survival rate from 5 to 10% up to 65%, which is primarily due to the elimination of micrometastases⁸⁻¹²

Although the optimum combination chemotherapy has not yet been established, a regimen containing vincristine, adriamycin, cyclophosphamide and actinomycin D, was the standard first-line treatment for patients with localized disease¹².

Conclusion

we report a rare case of ES/PNET presenting as a breast mass. Clinical presentation mimicked invasive breast cancer. The tumour did not respond well to multimodality treatment and local and distant metastasis occurred less than 2 years after first diagnosis.

Management varies according to the presentation and experience of each centre as there are no established guidelines on primary Ewing's sarcoma/PNET of the breast. The guiding principles are derived from treatment approach and chemotherapy regimens targeted on soft tissue sarcomas.

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