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Case Report

A rare metachronous presentation of periductal stromal sarcoma in a case of recurrent rhabdomyosarcoma – Case report

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ABSTRACT

Introduction: Periductal stromal sarcoma (PSS) is a an extremely rare low-grade malignant neoplasm arising from breast periductal stroma. It usually occurs in peri and post-menopausal women. Herein, we report a rare case of a 14 year girl, known case of recurrent Rhabdomyosarcoma (RMS) presenting with periductal stromal sarcoma of breast. To the best of our knowledge, this is a first of its kind to be reported in literature.

Case Details: A 14year girl, known case of Recurrent Pleomorphic Rhabdomyosarcoma of retroperitoneum, presented with an abdominal mass and lump in right breast after 10 years. On chemotherapy, abdominal mass regressed and breast lump progressed in size. On further evaluation by histopathological examination and immunohistochemistry, the breast lump turned out to be a periductal stromal sarcoma.

Discussion: Periductal stromal tumors have histological similarities to phyllodes tumors, with the biphasic presence of hypercellular and variably atypical, mitotically active stroma hugging benign epithelium. There are very few case reports on periductal stromal tumors and thus a rare entity. PSS as a metachronous presentation with recurrent RMS is even more rare with the prevalence of multiple soft tissue sarcomas being 0.08%.

Conclusion: A clinically suspected metastatic lesion, if accessible for biopsy, warrants a meticulous histopathological examination to ascertain the cell of origin. The therapeutic strategy and prognosis of PSS is unclear, thus a longer follow up is imperative to determine its prognosis and clinical outcome.

Novelty: 2^{nd} case reported so far in younger individuals. 1st case of PSS in a Recurrent RMS.

1. Introduction

Periductal stromal sarcoma (PSS) is a an extremely rare low-grade malignant soft tissue neoplasm arising from breast periductal stroma.[1] It is a fibroepithelial tumor, characterized by its biphasic morphology of benign ductal elements and malignant stromal spindle cells displaying varying degrees of atypia and mitotic activity. It lacks the leafy architecture of phyllodes tumor but can progress to phyllodes tumor or other sarcomas. PSS occurs usually in peri and post-menopausal women. PSS occurring as a second primary in a case of Rhabdomyosarcoma is extremely rare. Herein, we report a rare case of a 14 year girl, known case of recurrent Rhabdomyosarcoma (RMS) presenting with periductal stromal sarcoma of the breast. To the best of our knowledge, this is a first of its kind to be reported in the literature.

2. Case Report

A 14year girl, known case of Recurrent Pleomorphic Rhabdomyosarcoma of retroperitoneum, (diagnosed

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in 2011 with recurrence in 2014 and 2020) presented with an abdominal mass and lump in her right breast after 10 years. The patient had no significant family history. In 2011, the patient had an abdominal mass for which she underwent Exploratory laparotomy and excision of left retroperitoneal tumor. It was diagnosed as Pleomorphic Rhabdomyosarcoma with diffuse positivity for desmin, myogenin and vimentin. She was treated with 6 cycles of VAC + IE (vincristine, actinomycin D, and cyclophosphamide /ifosfamide). In 2014, the patient had a recurrence in the paraspinal region with multiple bilateral inguinal lymph adenopathy. She was treated with chemoradiation (12 cycles of IRINOTECAN + CARBOPLATIN chemotherapy → Radiotherapy). On follow-up, the mass had reduced in size. All lab investigations were within normal limits. In 2020 December, the patient again had a recurrence. PET CT Scan showed 14 x 10 x 15.6 cm large ill-defined, solid, cystic mass lesion involving the left inferior perinephric space, adjacent para-colic with left iliac and gluteal soft tissue lesions, likely metastatic. Also seen is a 2.7 x 3.5cm mass in the right breast with an SUV of 3.8 which was suspected as metastasis and was treated with chemotherapy. A biopsy was taken from the retroperitoneal mass which was consistent with recurrent Pleomorphic Rhabdomyosarcoma with desmin, myogenin and vimentin positivity (Figure 1).

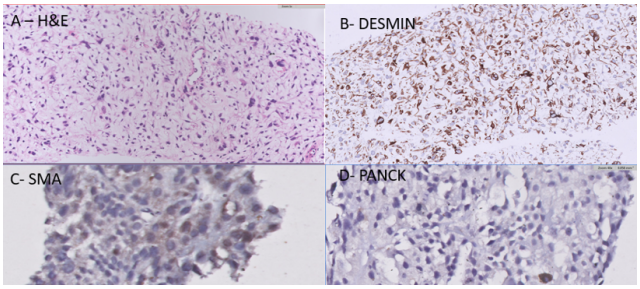

Fig. 1: A: H&E stain at 100x. **B, C, D:** IHC stain at 100x. Biopsy from the retroperitoneal mass showed pleomorphic cells (A) which were strongly positive for desmin (B), focally positive for SMA and negative for Pan cytokeratin.

On further follow up in April 2021, the retroperitoneal mass reduced in size (4.3 x 2.7 x 8.5. SUV Nil), whereas the breast mass increased in size (3 x 4.2cm) with an increase in SUV (5.6) (Figure 2).

3. Histopathology

Biopsy from the breast mass showed biphasic morphology with focal epithelial hyperplasia of the ducts and periductal proliferation of atypical stromal cells with nuclear pleomorphism and prominent nucleoli (Figure 3). On immunohistochemistry (Figure 3), these atypical cells were diffusely positive for CD34 and negative for desmin, myogenin and PanCK. Hence was diagnosed as Periductal

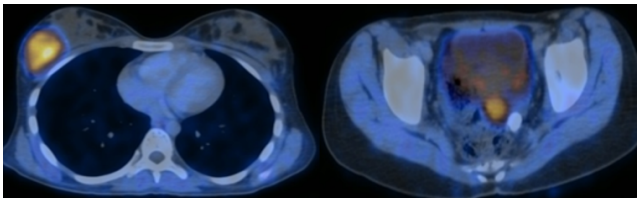

Fig. 2: PET-CT shows a hyperdense lesion in right breast with an SUVof 5.6

stromal Sarcoma of the breast in a known case of Recurrent Pleomorphic Rhabdomyosarcoma. The patient underwent excision of the Periductal stromal Sarcoma outside and continued chemotherapy for Rhabdomyosarcoma.

During a follow-up period of 15 months, the patient had not shown any symptoms or signs of local or distant recurrence of periductal stromal tumor with a significant reduction in the size of pleomorphic rhabdomyosarcoma.

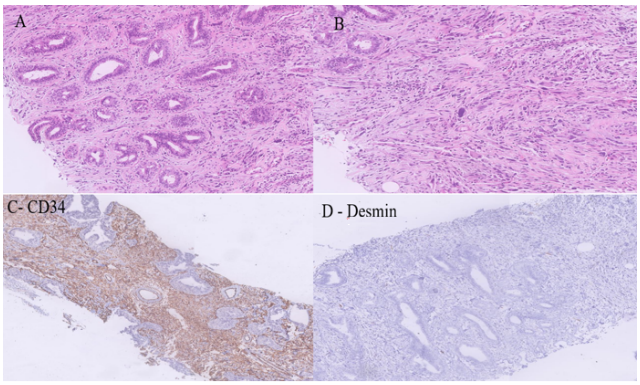

Fig. 3: A,B: H&E stain at 100x. **C,D:** IHC stain at 100x Histopathology shows epithelial hyperplasia of ducts with proliferation of atypical stromal cells. These atypical stromal cells show diffuse positivity for CD34 (A) and are negative for desmin (B).

4. Discussion

Periductal stromal tumours have histological similarities to phyllodes tumours, with the biphasic presence of hypercellular and variably atypical, mitotically active stroma hugging benign epithelium, but periductal stromal tumours do not show a distinct tumour outline or obvious fronded architecture. Instead, hypercellular stroma extends around benign epithelium of ducts and lobules in an irregular manner, between unaffected breast parenchyma. Although periductal stromal tumour lacks the fronded architecture of typical phyllodes tumours, there are finding of focal phyllodes features in recurrences and the morphological coexistence of periductal stromal tumour in some phyllodes tumours. Therefore, according to WHO classification, 5^{th} edition, periductal stromal tumour is

being regarded as a subtype of phyllodes tumour.[2]

It usually occurs in peri and post-menopausal women with a mean age of 55.3years,[1] however it has also been described in a 14year boy.[3] In our current case, age at presentation of PSS was also 14 years.

A case of synchronous presentation of PSS with bilateral breast carcinoma has been described,[4] whereas ours was a metachronous presentation of PSS with a soft tissue sarcoma. In a study on multiple soft tissue sarcomas in a single patient, the prevalence was found to be 0.08%, thereby ascertaining the rarity of our case.[5]

The therapeutic strategy for the PSS has not yet been determined as the number of studies available on periductal stromal sarcoma is currently limited. A close follow-up is required in these patients, though resection with adequate margins is considered sufficient.[6] In a case series of 10 women, one of them had a local recurrence and one woman experienced a progression to phyllodes tumor.[7] In our case, the patient didn't have a recurrence or progression of the disease for a period of 16 months. A genetic workup is also essential in patients with multiple soft tissue tumors.

Excess sarcoma risks after childhood cancer are well established with the most common second primary soft tissue sarcoma being fibrohistiocytic tumors, vascular tumors, nerve sheath tumors, leiomyosarcoma, liposarcoma and rhabdomyosarcoma.[8] Periductal stromal sarcoma as a second primary sarcoma has not been reported so far. In general, though secondary sarcomas are rare, there are predisposing factors that can substantially increase this risk in certain populations. Chemical exposure, certain viruses, cytotoxic and immunosuppressive agents, chronic edema, and radiation exposure are the predisposing environmental risk factors with the strongest association with sarcoma. In addition, the most common genetic disorders that carry a predisposition for sarcoma development include hereditary retinoblastoma (RB), Li-Fraumeni syndrome (LFS), neurofibromatosis type 1 (NF1), and DICER1 syndrome.[9] Our case had prior chemoradiotherapy owing to the etiological factor and necessitates the need for further genetic analysis.

Although treatment does not generally differ for primary versus second primary sarcomas, awareness of the predisposing factors can alter therapeutic strategies to minimize risk, aid prompt diagnosis by increasing clinical suspicion, and allow for appropriate surveillance and genetic counselling for those patients with cancer predisposition syndromes.

5. Conclusion

A high degree of vigilance is required in patients with a previous STS to detect both local recurrences and to identify new masses remote from the previous STS site. Acquiring an early histological diagnosis should be attempted. A clinically suspected metastatic lesion, if accessible for biopsy, warrants a meticulous histopathological examination to ascertain the cell of origin. The therapeutic strategy and prognosis of PSS are unclear, thus a longer follow-up is imperative to determine its prognosis and clinical outcome.

6. Source of Funding

None.

7. Conflict of Interest

None.

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