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Desmoplastic Small Round Cell Tumor Of Thoracic Wall: A Case Report

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1. Abstract

1.1. Background:
Desmoplastic small round cell tumor is an uncommon and highly malignant neoplasm characterized by the presence of small round cells, which often lacks typical clinical and imaging manifestations. Its diagnosis relies on histopathological features, immunohistochemistry, and molecular examination. Predominantly affecting adolescents, this tumor primarily occurs in the abdominal and pelvic cavities, with only rare instances reported in the thoracic wall.

1.2. Case presentation:
In this article, we present a unique case of desmoplastic small round cell tumor originating from the thoracic wall that was admitted to our hospital. The patient, a 17-year-old female, sought medical attention due to chest trauma. Breast ultrasound revealed a heterogeneous echogenic mass located at the lower right margin of the breast. Subsequent chest computed tomography confirmed that the mass originated from soft tissue within the thoracic wall. A percutaneous biopsy was performed on the mass, revealing characteristic histopathological features consistent with desmoplastic small round cell tumor. Additionally, pleural effusion developed in this patient and cytological analysis demonstrated cancerous cells within it. Unfortunately, after being referred to a tertiary care center for further management, she was lost to follow-up.

1.3. Conclusion:
Desmoplastic small round cell tumor is an exceedingly rare and aggressive malignancy associated with an extremely poor prognosis. Accurate diagnosis heavily relies on careful evaluation of histopathological features along with immunohistochemical staining techniques and molecular testing methods.

2. Keywords:
Desmoplastic small round cell tumor, Thoracic wall, Pathological diagnosis

3. Background

Desmoplastic small round cell tumor (DSRCT) is a rare and highly malignant mesenchymal tumor characterized by the presence of epithelioid small round cells surrounded by sclerotic fibrous connective tissue. The first report on DSRCT was published in 1989 by Gerald et al, followed by subsequent literature reports both domestically and internationally [1]. DSRCT predominantly affects individuals between the ages of 5 and 30, with a higher incidence among males; however, females constitute a larger proportion of patients under the age of 20 [2-3]. While DSRCT typically originates in the abdominal or pelvic cavity, it can also occur extra-abdominally in locations such as the lung or thoracic wall [4]. In their study, Gerald WL et al found that only 6% of DSRCT cases occurred outside the abdomen [5]. This relatively unknown malignancy is considered aggressive with poor prognosis. Bulbul A. et al reported a median survival time ranging from 17 to 25 months for abdominal DSRCT cases, with a five-year survival rate of merely 15% [6]. Despite multimodal therapy approaches, studies have consistently shown that the five-year survival rate for DSRCT remains below 18% [7-9]. Although significant advancements have been made in medical technology, managing DSRCT still poses challenges. Herein, we present an exceptional case of thoracic wall DSRCT in a seventeen-year-old female to raise awareness about this disease and provide relevant clinical insights.

4. Case presentation

A 17-year-old female was admitted to our hospital for 1 month with right-side chest pain and shortness of breath. There is nothing special about her history, her family history, etc. Breast ultrasound (US) showed a mixed echoic mass at the lower limit of the right breast, with a large mass and blurred boundaries between the mass and the chest wall on ultrasound (Fig. 1a).

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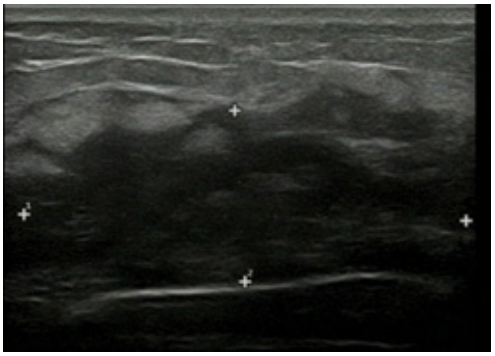

Fig.1: (A) US images of the patient:

Moreover, the color Doppler US showed a solid mass with abundant internal and peripheral blood flow (Fig. 1b).

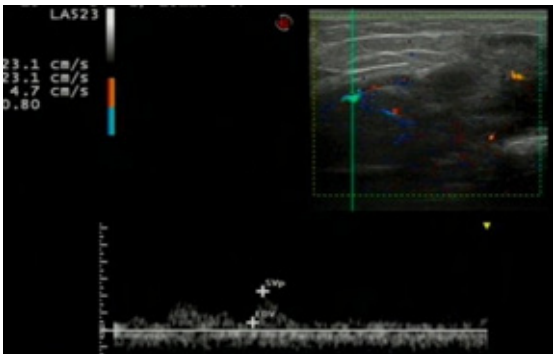

Fig.1: (B) US images of the patient:

At first, based on the breast US results, we thought the mass was of breast origin. Subsequently, her chest computed tomography (CT) examination revealed a soft tissue mass on the right chest wall, about 17.7*1.2cm in size, and the surrounding organs were compressed and displaced. A large amount of fluid was accumulated in the right thoracic cavity (Fig. 2).

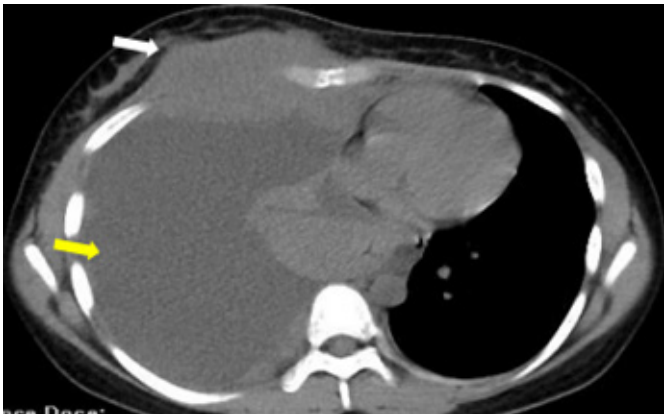

Fig.2: CT scan images of the patient:

The US-guided puncture biopsy of the right chest wall mass was performed in our department. Under the pathological microscope, the tumor cells were arranged in the shape of nests, sheets, and clusters with clear boundaries, and the fibrosarcoma of the tumor tissue was significantly hyperplasia. Immunohistochemistry showed that WT1, TTF-1, CD56, Syn, Desmin, and CgA were positive, and the proliferation index of Ki-67 was 30% (Fig. 3a-b).

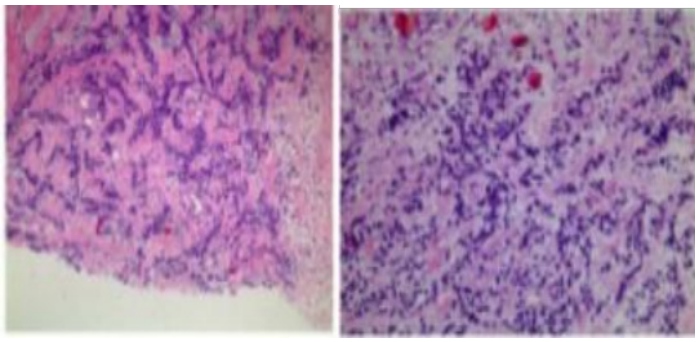

Fig.3: Pathologic image of puncture tissue obtained from right chest wall mass:

The pathological findings supported small round cell tumors and tended to promote the proliferation of connective tissue small round cell tumors. The diagnosis was a small round cell tumor of the right chest wall promoting connective tissue proliferation. CT examination of the patient‘s chest revealed a right pleural effusion, followed by a pleural puncture to extract the effusion and perform a liquid-based cytopathological examination, which showed that the deeply stained cells in the pleural effusion were consistent with cancer cells (Fig. 4).

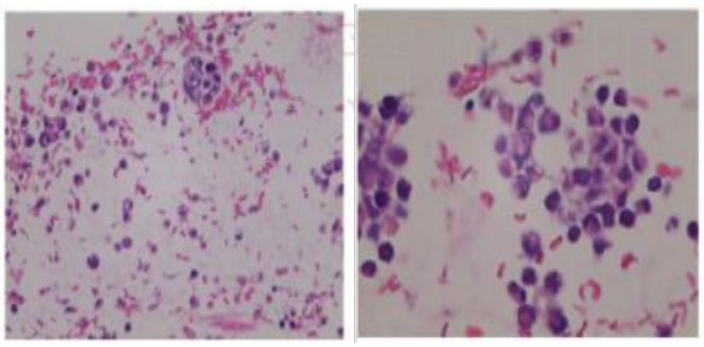

Fig.4: Liquid-based cytological pathological image obtained from right pleural effusion:

However, the patient‘s family requested transfer to a tertiary hospital for further treatment and discharge. Unfortunately, we lost contact with the patient and were unable to obtain follow-up information regarding her treatment and prognosis.

a. The mass is located posterior to the breast, exhibiting poorly defined boundaries and indistinct demarcation from the chest wall.
b. Abundant blood flow can be visualized within and surrounding the mass.

A soft tissue mass was identified on the right chest wall, extending backwards into the thoracic cavity measuring approximately 12.2*10.7cm in size, resulting in compression and displacement of adjacent organs.

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Significant fluid accumulation was observed within the right pleural cavity along with mediastinal shift towards left side and compression of right lung. Microscopically, cancer cells exhibited nest-like arrangement, sheet-like growth pattern with hyperchromatic nuclei, frequent mitotic figures, extensive interstitial proliferation of tumor tissues as well as abundant hyalinoid changes in collagen fibers. Immunohistochemical analysis revealed positive staining for Desmin (+), CD56 (+), TTF-1 (+), CgA (+), Syn (+), WT-1 (+), Vimentin (+); negative staining for CD20 (-), CK7 (-), CD99 (-), NSE (-); Ki-67 index was 30%. Microscopic examination showed cells with enlarged nuclei displaying deep staining characteristics. Immunohistochemical analysis demonstrated positive staining for TTF-1(+), CgA (+), Syn (+); negative staining for CK7(-) , Napsin A(-) , CA125(-) , CA19-9(-). Based on cytological morphology combined with immunohistochemical results, these deeply stained cells in pleural effusion were consistent with cancer cells.

5. Discussion

The incidence of small round cell tumors is relatively low, with a global occurrence rate of approximately 0.3 per million [10]. *** and has been clearly defined for nearly ten years [1]. The clinical manifestations of DSRCT lack specificity in imaging and are not typical. Symptoms vary depending on the primary site, with abdominal symptoms such as pain, distension, and constipation being the most common [4,11]. Compared to other sites, thoracic wall involvement in DSRCT is rare and has been infrequently reported in literature. In cases where the anterior thoracic wall is affected, DSRCT may present as a breast mass. In this study, initial misdiagnosis occurred during a breast ultrasound examination; however, subsequent chest CT confirmed that the mass originated from the thoracic wall. A puncture biopsy ultimately confirmed it to be DSRCT originating from the thoracic wall. Currently, there are no definitive diagnostic criteria both domestically and internationally; therefore, diagnosis still relies on pathological characteristics along with immunohistochemistry and molecular examinations. The presence of EWSR1-WT1 fusion gene resulting from characteristic chromosomal translocation t (11;22) (p13; q12) serves as a genetic feature indicating stable existence of DSRCT [12]. Characteristic immunohistochemical markers for DSRCT primarily include epithelial membrane antigen (EMA), keratin proteins (CKs), neuron-specific enolase (NSE), vimentin (VIM), desmin (DES), among others [13].

Although rare in this report‘s case involving the thoracic wall region, differentiation between breast tumors and other small round cell tumors including Ewing‘s sarcoma, rhabdomyosarcoma, and small cell carcinoma remains crucial. In our study, a mixed echo mass was identified on chest wall ultrasound examination which correlated with soft tissue mass findings suggested by chest CT. Furthermore, the histopathological characteristics of this case are consistent with those described in the literature. The pathological features include round cells of similar size, frequent mitotic images, and positive expression of TTF-1 (+), CgA (+), and Syn (+). DSRCT patients have a poor prognosis, and currently there is no standardized treatment plan; however, surgical resection combined with chemoradiotherapy, targeted therapy, and other therapeutic strategies remain the mainstay [2]. Surgical resection is considered the optimal treatment approach. Previous reports have indicated a 0% 3-year survival rate for patients with unresectable DSRCT [3]. In our case study, the patient presented with significant pleural effusion and dyspnea symptoms at diagnosis. Cytological examination confirmed that the pleural effusion was cancerous in origin. A series of studies by Monika Scheer et al., investigating multimodal treatments and risk factors in 60 DSRCT patients, identified pleural effusion as a potential risk factor associated with lower survival rates [14]. Some studies have demonstrated efficacy of small-molecule TKIs in treating advanced cases of DSRCT [15-16], providing an alternative treatment option. Due to our hospital being a grassroots facility rather than a specialized cancer center, upon diagnosis the patient opted to seek care at a higher-level cancer center; therefore, we lack follow-up information on her prognosis. Given its rarity and challenging detection through clinical or imaging findings alone, accurate diagnosis of DSRCT relies on histopathological features along with immunohistochemistry and molecular examinations. This report presents an exceptional case involving chest wall-derived DSRCT in a 17-year-old female patient. Further research is warranted to explore related pathogenesis as well as treatment options and prognostic factors for chest wall-derived DSRCT.

6. Conclusion

DSRCT is a rare malignancy, and its diagnosis relies on histopathological features, immunohistochemistry, and molecular examination, posing challenges in clinical and imaging detection. Herein, we present an exceptional case of chest wall DSRCT in a 17-year-old female. Further investigations are warranted to elucidate the underlying pathogenesis, treatment strategies, and prognosis associated with DSRCT originating from the thoracic wall. Radiologists should be cognizant of the characteristic imaging manifestations of this disease while considering potential differential diagnoses with breast lesions. Enhancing our comprehension of this disease can facilitate early detection and treatment interventions to improve patient survival rates.

7. Abbreviations

DSRCT: Desmoplastic small round cell tumor; US: Ultrasound; CT: Chest computed tomography.

8. Authors' contributions

JS participated in the puncture biopsy of the right chest wall mass as a diagnostic doctor, provided data and revised the manuscript. YY and QY collected patient data, conducted data analysis, and drafted the manuscript. RZ identified the case and contributed to diagnosis and puncture surgery. JH was involved in patient diagnosis and puncture surgery. All authors have reviewed and approved the final manuscript.

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9. Availability of data and materials

The data were presented in the main body of the manuscript.

10. Ethics approval and consent to participate

The ethics committee of Huizhou Hospital Affiliated to Guangzhou Medical University (Huizhou Third People's Hospital) approved the study.

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