Case report

Desmoplastic Small Round Cell Tumor: A Potpourri of Cases Highlighting Diagnostic and Therapeutic challenges

Abstracts

Desmoplastic small round cell tumour (DSRCT) is a rare with an incidence of 0.3-0.7 cases/million worldwide, aggressive neoplasm characterized by small round cells in a desmoplastic stroma, commonly linked to the EWSR1-WT1 gene fusion. This malignancy primarily affects children and young adults, with a higher incidence in males. DSRCT presents significant diagnostic and therapeutic challenges due to its rarity and poor prognosis, which includes a low 5-year survival rate of 15%–30%.

This case series details three instances of DSRCT, each highlighting distinct diagnostic challenges and therapeutic approaches. The first case involved a young woman with bilateral lower limb edema and a confirmed diagnosis of high-grade rhabdomyosarcoma, leading to rapid deterioration and death shortly after diagnosis. The second case described a 62-year-old woman with a poorly differentiated tumour in the breast, necessitating aggressive neoadjuvant chemotherapy. The third case featured a 62-year-old man with a mediastinal mass causing airway obstruction, requiring urgent intervention and chemotherapy.

DSRCT presents formidable challenges in oncology due to its aggressive nature and poor prognosis. Emphasizing early diagnosis and a multidisciplinary approach is critical to improving patient outcomes in this rare malignancy. Further research into novel therapeutic strategies is necessary.

Introduction

Desmoplastic small round cell tumour (DSRCT) is a rare and aggressive mesenchymal neoplasm characterized by small round tumour cells embedded in a desmoplastic stroma. This malignancy often harbours the EWSR1-WT1 gene fusion, a critical marker that aids in its diagnosis. [1] DSRCT shares histological features with other small round cell tumours, such as Ewing sarcoma and rhabdomyosarcoma, both known for their aggressive behaviour, early metastasis, and high mortality rates. [2,3]

Primarily affecting children and young adults, DSRCT exhibits a significant male predominance, with peak incidence during the third decade. With an estimated incidence of 0.2–0.74 cases per million individuals annually, DSRCT poses a significant challenge due to its rarity and poor prognosis, with a 5-year survival rate ranging from 15%–30%.[2,3]

Given the limited data on treatment outcomes, this case series presents three rare occurrences of DSRCT, focusing on the diagnostic challenges, therapeutic interventions, and clinical outcomes in the context of this formidable disease.

Case presentation

Case 1

A 23-year-old woman presented with progressively worsening bilateral lower limb edema, resulting in difficulty ambulating. Imaging revealed extensive peritoneal nodules along with a mass of unknown origin, raising concerns for malignancy.

A biopsy was performed on a lesion located in the right perineum, and the histopathological examination ultimately confirmed a diagnosis of high-grade rhabdomyosarcoma, a particularly aggressive subtype of small round blue cell tumours. This diagnosis was met with a treatment plan that included aggressive multimodal therapy.

Despite aggressive multimodal therapy, the patient's condition rapidly deteriorated, leading to multiorgan failure. She tragically succumbed to her illness two weeks after diagnosis, underscoring the aggressive nature of small round blue cell tumours and the limitations of current therapeutic strategies.

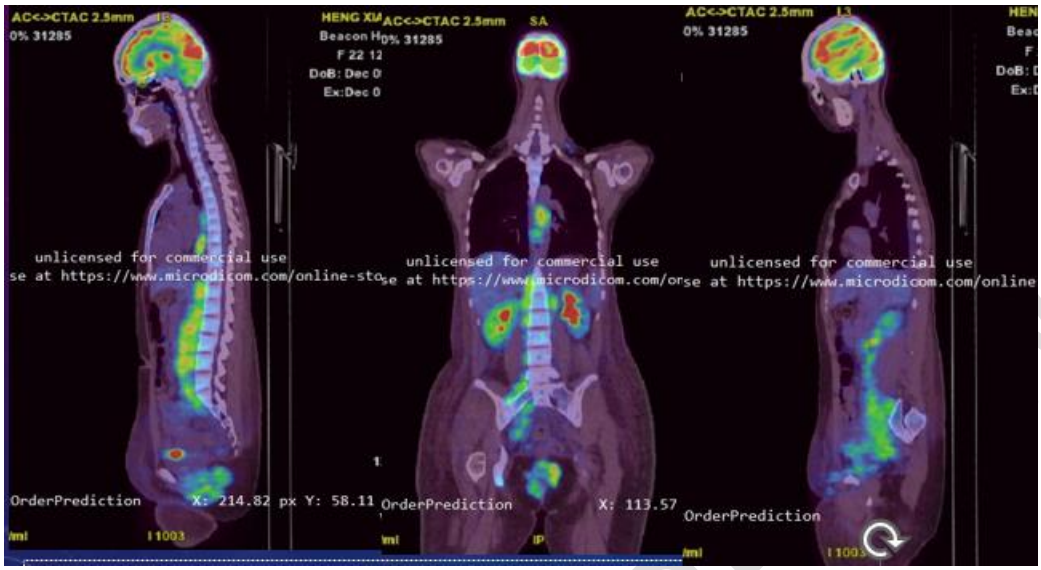
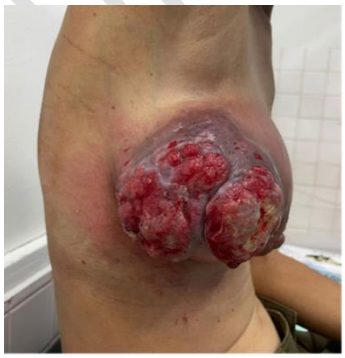


Figure 1: PET-CT scan showed increased FDG uptake mass at the perineum extending to the gluteal cleft measuring 6.9x5.6x6.2cm with multiple matted intra-abdominal LN

Case 2

A 62-year-old woman presented with a rapidly enlarging fungating lesion on her right breast. Imaging studies and biopsy confirmed a poorly differentiated tumour consistent with a small round blue cell tumour. A contrast-enhanced computed tomography (CECT) scan revealed a large right breast tumour with multiple enlarged axillary lymph nodes, while a positron emission tomography (PET) scan demonstrated increased fluorodeoxyglucose (FDG) uptake in both the right breast mass and the axillary lymph nodes.

Histological analysis confirmed a myogenic origin, with malignant cells showed strong and diffuse positivity for Vimentin, Desmin, and myogenin. Given the extent of the disease, the patient was initiated on neoadjuvant chemotherapy to reduce the tumour burden before the surgery. Despite the aggressive therapeutic approach, the extensive regional involvement presented significant clinical challenges.



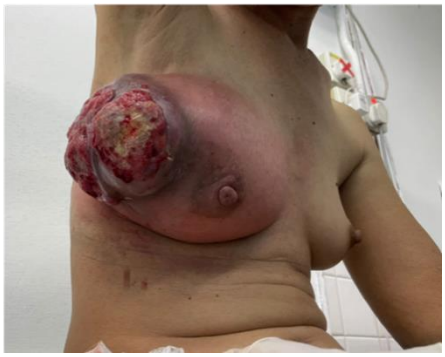


Figure 2: Photos showed the rapidly enlarging fungating right breast lesion.

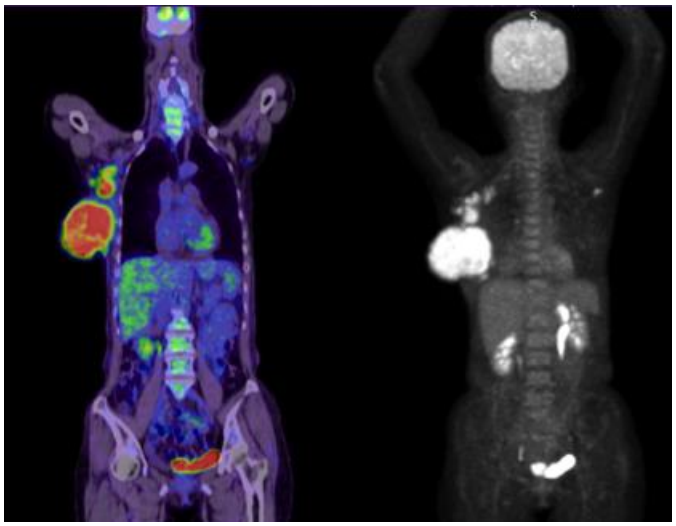


Figure 3: CECT scan showed a huge right breast tumour with multiple enlarged axillary LN . PET CT Scan showed increased FDG uptake of right breast lump and axillary LN

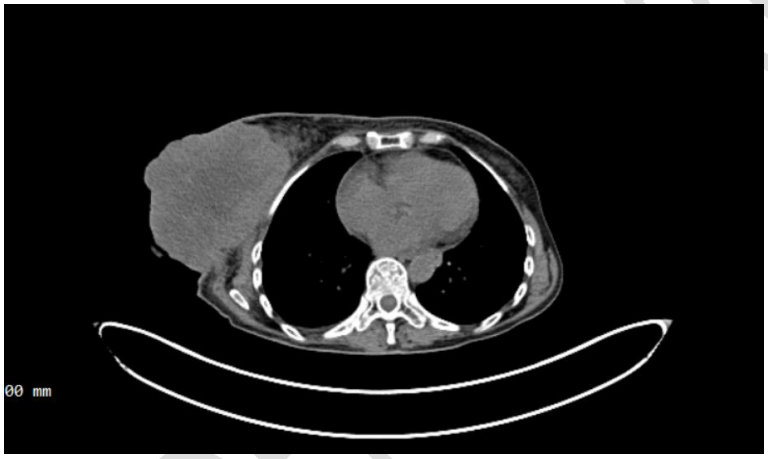


Figure 4: CECT Thorax showed enlarging right breast lesion.

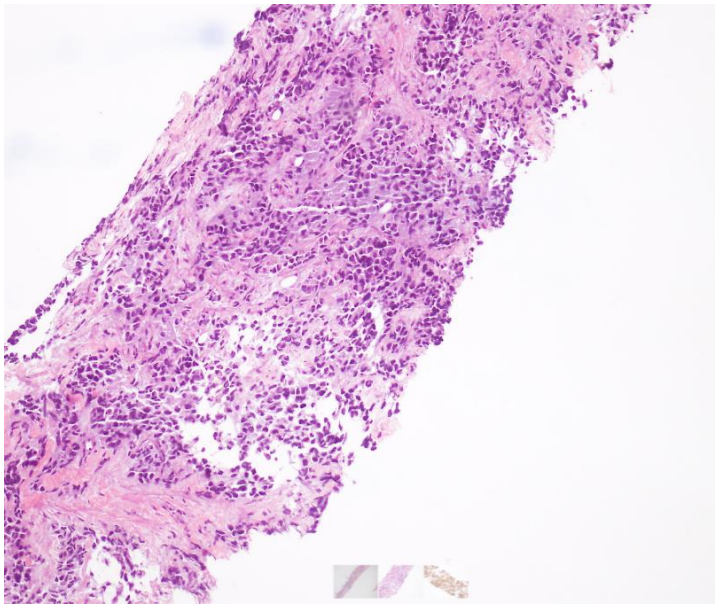


Figure 5: The malignant cells are strongly and diffusely positive for Vimentin, Desmin and myogenin.

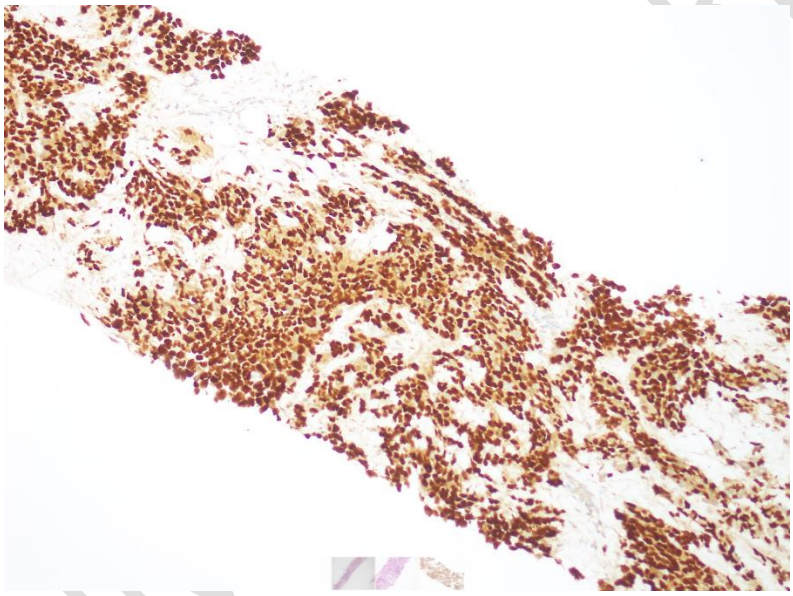


Figure 6: Strong desmin reactivity characteristic of desmoplastic small round cell tumor

Case 3

A 62-year-old gentleman presented with sudden breathlessness. A CT scan revealed a superior mediastinal mass infiltrating the thyroid gland and causing compression of the trachea, raising concerns for airway obstruction. Histopathology identified a small round blue cell tumor with neuroendocrine features.

Urgent airway stenting was performed to alleviate the obstruction, and chemotherapy was initiated to manage the aggressive malignancy. This case underscores the life-threatening nature of

mediastinal DSRCT and highlights the importance of prompt airway management in similar presentations.

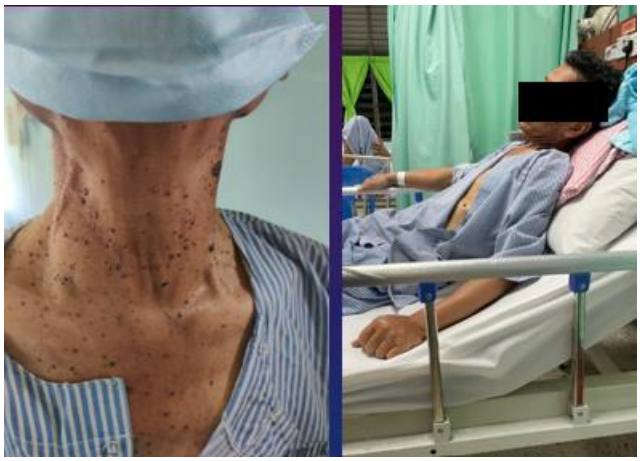


Figure 7: He unable to lie flat due to enlarging intra-thoracic mass.

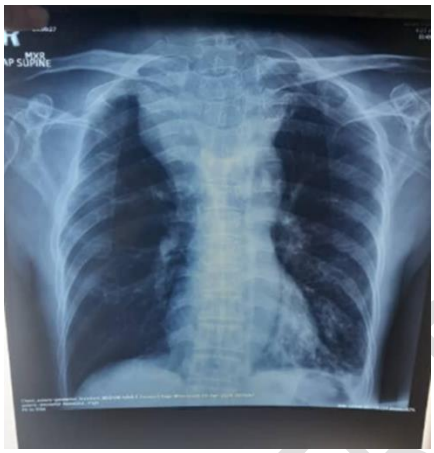


Figure 8: Chest X-ray showed enlarging mediasternal mass.

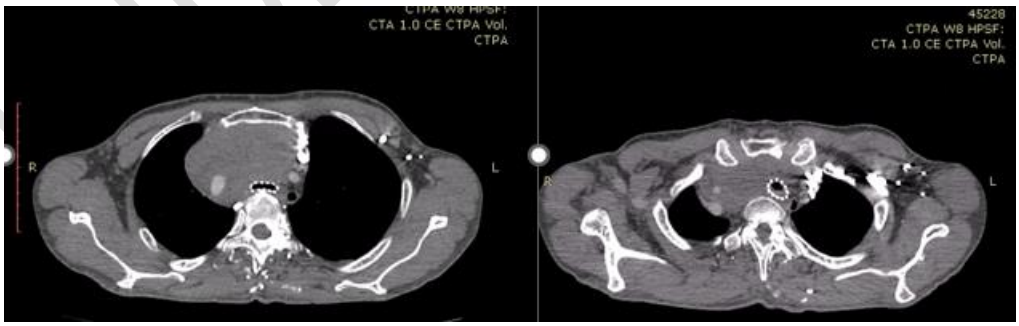


Figure 9: Enlarged mediastinum and CECT thorax showed mass causing airway compression

Discussion

“The desmoplastic small round cell tumors (DSRBCT) is a rare and aggressive tumor with an incidence of 0.3-0.7 cases/million worldwide, with extremely high mortality rate. Patients usually

have an average life expectancy of less than 3 years at diagnosis, and less than 15% of them are given a 5-year life expectancy".[1,2,11] "The vast majority of patients with desmoplastic small round cell tumors (DSRCT) develop tumors primarily in the abdominal cavity, where multiple serosal implants are commonly observed. Clinical presentations outside the abdominal cavity are rare, typically confined to the thoracic cavity and paratesticular region. Occasionally, these tumors can manifest in the limbs, head and neck, kidneys, and brain, underscoring their unpredictable nature. [1,3] A recurrent translocation, t(11;22)(p13;q12), leads to the formation of the EWSR1-WT1 fusion gene, resulting in a chimeric protein with significant transcriptional regulatory activity"[2]

Clinical symptoms often reflect the primary site of the tumor, presenting as pain, palpable masses, abdominal distension, obstruction, or ascites. Alarmingly, at the time of initial diagnosis, approximately 90% of patients exhibit multifocal disease, which can include nodular formations, diffuse peritoneal involvement, omental disease, or a combination of these conditions. Notably, a significant number of patients demonstrate diaphragmatic involvement.

"Tissue sampling remains the gold standard for a definitive diagnosis. Diagnostic techniques such as fluorescence in situ hybridization (FISH) to detect EWSR1 rearrangements and reverse transcription polymerase chain reaction (RT-PCR) to identify EWSR1-WT1 fusion transcripts are routinely employed".[8] "Histologically, the tumor shows considerable variability, with small, round, oval, or spindled cells arranged in nests, cords, sheets, or trabeculae. These cells have hyperchromatic nuclei, scant cytoplasm, a high mitotic rate, and well-defined solid areas in a desmoplastic stroma. Less common patterns include papillary, glandular, and cribriform forms, with cells that are clear, fusiform, pleomorphic, rhabdoid, or basaloid, often forming rosettes or pseudorosettes".[11,12] "These complementary methods enhance diagnostic accuracy, confirming cases that may be ambiguous when assessed by either technique alone".[4,6,7]

"Imaging studies provide crucial insights into tumor characteristics. On ultrasound, DSRCTs appear as lobulated, hypoechoic, and heterogeneous masses with increased vascularity. Unenhanced CT scans reveal hypoattenuating masses affecting the peritoneal surfaces, omentum, and mesentery, while MRI typically shows heterogeneous isointense to hypointense masses on T-weighted images, sometimes with foci of calcification. PET/CT imaging often reveals intense FDG hypermetabolic activity within the soft tissue masses". [1,4,5]

"Despite aggressive multimodal treatment approaches, DSRCT is associated with dismal outcomes. Approximately 60–70% of patients succumb to disease progression, usually within three years of diagnosis.[3,6,7] Median overall survival ranges from 28 to 60 months, with median disease-free survival between 10 and 15 months. Systemic chemotherapy and surgical intervention have been shown to reduce mortality rates, though the presence of residual macroscopic disease post-resection correlates with an increased risk of mortality" . [8,9]

"Currently, there is no validated staging system for DSRCT, but the extensive nature of peritoneal disease often leads to the use of the Peritoneal Cancer Index for assessment. Management typically involves a combination of chemotherapy, radiation, aggressive cytoreductive surgery, and intraperitoneal hyperthermic chemotherapy (HIPEC). Estimated median overall survival rates vary significantly based on treatment: those not undergoing surgery or HIPEC have a 3-year survival rate of approximately 26%, while patients who undergo HIPEC and surgery show an impressive 71%, and those receiving only debulking surgery have a 62% survival rate"[2,3]

"Small round blue cell tumors are notorious for their aggressive behavior, with over 40% of patients presenting with distant metastasis at the time of diagnosis. Although multimodal treatment remains the cornerstone of management, survival rates after intensive therapy remain disappointingly low. Current studies indicate a 3-year overall survival rate of 44%, with a 5-year survival rate hovering around 15%".[1] A study conducted by Syed Hamza et al. in the USA in 2022 found that the

incidence of DSRBCT has been steadily increasing since 2000. The study also highlighted that patients with this disease have better survival outcomes when surgery is combined with chemoradiotherapy.[12]

Conclusion

Desmoplastic small round cell tumours present a significant challenge in oncology due to their aggressive nature and poor prognosis. Early diagnosis, multimodal treatment, and ongoing research into novel therapies are critical in improving outcomes for patients facing this rare malignancy. This case series highlights the diagnostic and therapeutic complexities of DSRCT and underscores the need for a multidisciplinary approach to managing these patients.

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Ethical Approval

As per international standards or university standards written ethical approval has been collected and preserved by the authors.

Consent

As per international standards or university standards, patients written consent has been collected and preserved by the authors

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