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Intra-Abdominal Desmoplastic Small Round Cell Tumor: A Case Report and Review of Literature

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Abstract **Case Report**

Introduction: Desmoplastic small round cell tumor is an uncommon and aggressive sarcoma affecting young males, its symptomatology is non-specific. Abdominal and pelvic CT scans help to orient, while anatomopathological and cytogenetic studies provide a definitive diagnosis. ***Case Report***: We report a case of a 21 years old male who complains of left hypochondral pain, physical examination revealed a painful mass on left hypochondrium. Abdominal computed tomography revealed intersplenorenal mass and one on left iliac fossa. He underwent a laparoscopic exploration and the resultant biopsy revealed a desmoplastic small round cell tumor. He underwent a neoadjuvant chemotherapy followed by surgery and consolidative radiochemotherapy with no recurrence during the 6 months follow up. ***Discussion***: Desmoplastic small round cell is very rare mesenchymal tumor, it is described as a result from the chromosomal translocation t (11; 22) (p13; q12) by Gerald and Rosai in 1991, it affects usually young male, patients consult for abdominal pain or mass, Computed abdominal and pelvic tomography is the first examination to request, it allows diagnostic orientation and classification, while exploratory laparoscopy and biopsy establish diagnosis with certainty showing Well-defined islands of small, round cells, embedded in abundant desmoplastic stroma with the expression of epidermal markers (cytokeratin, Epithelial Membrane Antigen), mesenchymal markers (desmin, vimentin) and neural markers (neuron specific enolase reactivity). The current treatment is based on neoadjuvant chemotherapy followed by surgery and consolidative radiochemotherapy, but the prognosis is poor. ***Conclusion***: Desmoplastic round cell tumor is an uncommon tumor of the young subject, it must be evoked in the presence of adequate imaging, the multimodal treatment is the current standard, but the prognosis remains poor. Future genetic therapies focused on developing targeted immunotherapy, might promise more optimistic results.

Keywords: Case report, Desmoplastic small round cell tumor, intraabdominal.

I. INTRODUCTION

Desmoplastic small round cell tumor is an uncommon and aggressive sarcoma that affects adolescent and young adult Caucasian males [1], it has been described for the first time by Gerald and Rosai in 1991 [4]. Symptomatology is non-specific, presenting as abdominal masses that may or may not be painful, most often located in the peritoneal cavity [3]. Imaging, mainly abdominal and pelvic CT scans, helps to orient the diagnosis [13], which can be confirmed by anatomopathological study, revealing well-defined islands of small, round cells, embedded in abundant desmoplastic stroma that shows immunohistochemical evidence of epithelial, mesenchymental and neural differentiation [5, 6].

Given the rarity of this type of tumor, there is no consensus on how to manage it. Treatment is currently multimodal, essentially chemotherapy, surgery followed by radiochemotherapy [1-11], but the prognosis remains poor [14].

We herein describe a case of 21 years old patient by analyzing the clinical, pathological features,

immunophenotypic profile of this rare disease, and by reviewing the treatment outcomes.

II. CASE REPORT

1. Clinical History

A patient of 21 years old with no particular medical history and no toxic habits, presented to the emergency department of CHU IBN ROCHD with left hypochondral pain that had been present for 2 months and had worsened. Physical examination revealed no abnormalities except for a painful mass on left hypochondrium with slight tenderness to palpation, the rest of the abdomen is supple.

2. Paraclinical Investigations

The patient underwent an abdominopelvic Computerized-Tomography (CT) scan, which revealed intersplenorenal mass which measures 13 x11x 14 cm **(Figure 1)** and one hypogastric mass that measures 14 x 11 x 10 cm **(Figure 2)**.

Biological check-up carried out a normal income and thoracic CT-Scan was normal.

The patient has undergone a laparoscopic exploration that reveals a mass in the left hypochondrium adherent to the left colonic angle and the greater omentum, and a hypogastric mass.

Pathological examination revealed a tumoral process organized into irregularly rounded basophilic structures of variable size, presenting cytonuclear atypia with images of mitosis, evolving within a well individualized, dense, desmoplastic fibrous connective tissue **(Figure 3)**.

On immunohistochemical study, WT-1 labelling was negative, pan-cytokeratin (AE1/AE3), desmin, anti-CD99, anti-Ki-67 were positive, concluding that the anatomopathological and immunohistochemical appearance was in favor of a desmoplastic small round cell tumor **(Figure 4)**.

3. Treatment and Evaluation

a) Chemotherapy

The patient received 6 cycles of VAC-IE chemotherapy (J1-J21) with granulocyte-colony stimulating factor (G-CSF); the protocol includes:
J1: Vincristine 2mg total dose + Doxorubicin 75mg/m² + cyclophosphamide 1200mg/m² + MESNA 1200mg/m²
J21: Ifosfamide 1800 mg/m² + MESNA 1800 mg/m² + Etoposide 100mg/m² during 5 days.

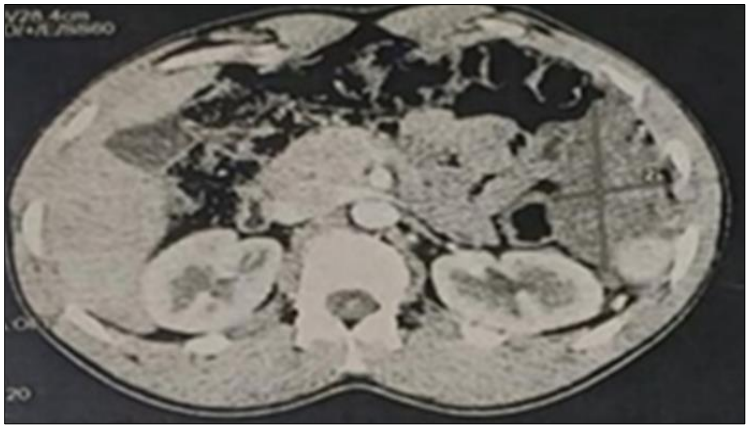

Figure 1: Axial abdominal CT-scan showing the intersplenorenal mass

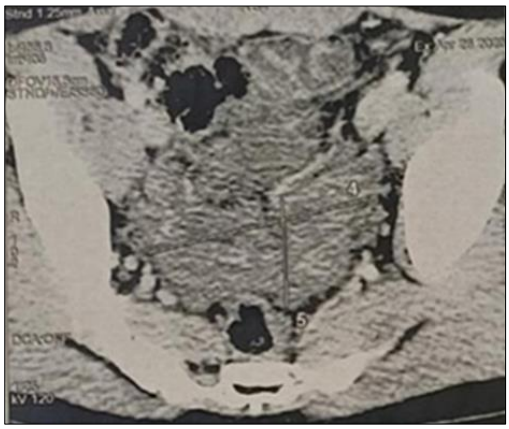

Figure 2: Axial pelvic CT-scan showing the hypogastric mass

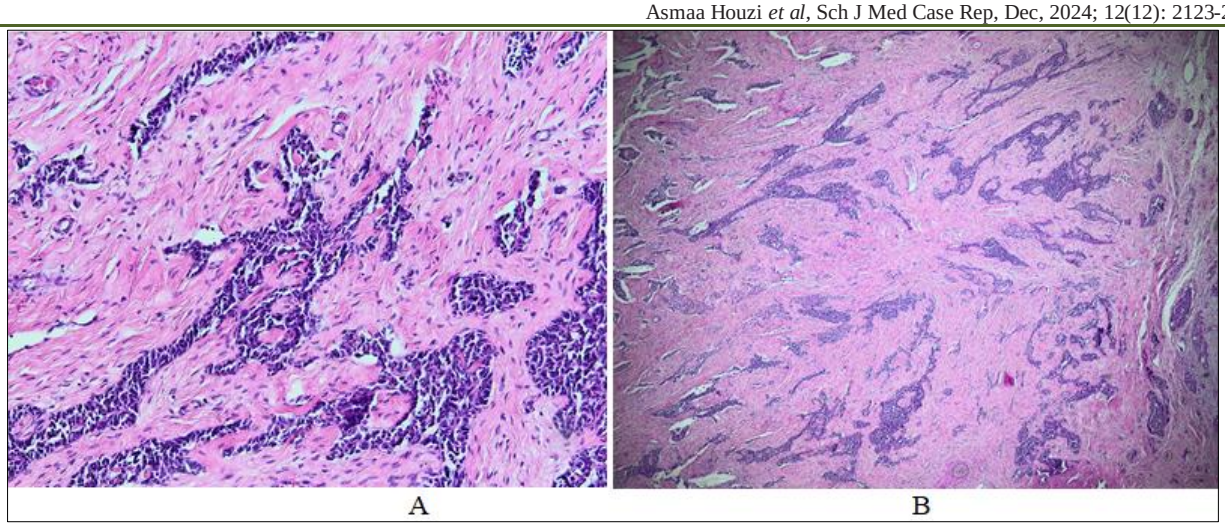

Figure 3: Histological images of a desmoplastic small round cell tumor. Image A (HE, x40) shows a poorly differentiated proliferation organized in anastomotic cords on a prominent desmoplastic stroma. Image B (HE, x200) shows a proliferation of small, round cells with reduced cytoplasm and hyperchromatic nuclei

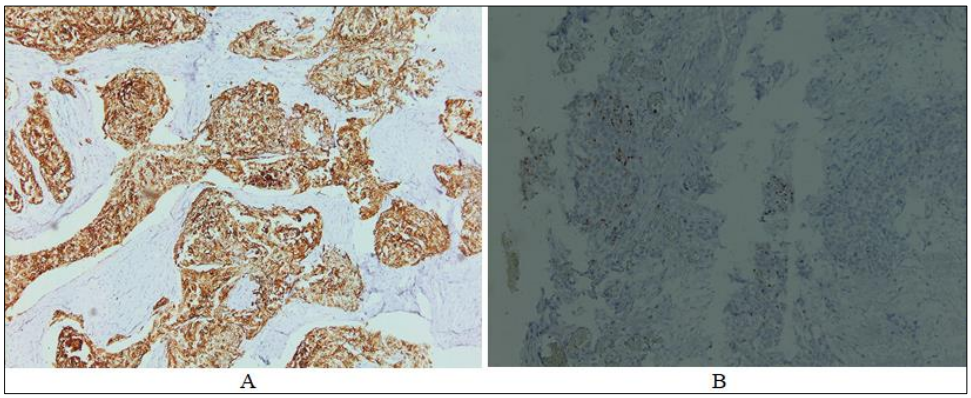

Figure 4: Immunohistochemical profile of desmoplastic small round cell tumor. Image a shows strong and diffuse expression of cytokeratin AE1/ AE3. Image B shows focal expression of desmine

b) Surgical Treatment

The patient underwent laparotomy with tumor resection of two masses a mass in the left colonic angle and a hypogastric mass with cytoreduction of carcinosis's nodules.

Surgical exploration showed the absence of effusion, the absence of liver metastases, and the presence of nodules of carcinosis with a PCI score of 22, the presence of a 12 cm mass adherent to the left colonic angle and the greater omentum, coming into contact with the spleen, the stomach and the tail of the pancreas without invading them **(Figure 5)**, and the presence of a 7 cm mass between the bladder and the rectum **(Figure 6)**.

c) Radiochemotherapy

Consolidative whole abdominopelvic radiochemotherapy was administered after chemotherapy and surgical resection, the patient underwent computed tomography (CT) based simulation in the supine position with custom vac-loc cradles for immobilization. The isocenter was typically marked at midabdomen and midseparation with the assistance of in-room lasers. Serial, noncontrasted CT images were obtained from midthorax to midfemur, and these images were transferred to the treatment-planning system for CT-based treatment planning.

For target volumes delineation, the clinical target volume (CTV) included the peritoneal cavity and retroperitoneal areas excluding liver and kidneys, the CTV was expanded by 10 mm for creation of the planning target volume (PTV) to account for setup variability and motion. Organs at risk were contoured including the liver, kidneys, lungs, heart, bladder and rectum **(Figure 7)**.

Radiation was delivered to the whole abdomen and pelvis to 30 Gy in 20 fractions, 1.5 Gy per fraction, administered once daily via IMRT with 6-MV photons. A representative dose volume histogram for this patient is illustrated **(Figure 8)**.

The patient had no severe complications after the radiochemotherapy, except a grade 1 diarrhea relieved by symptomatic treatments.

The patient was seen in consultation three months later, he was in a very good general condition, with no functional signs and, on physical examination, a supple abdomen with no palpable mass or abdominal tenderness.

An abdominopelvic CT-scan has been realized showing a disappearance of the two masses intersplenorenal and hypogatric **(Figure 9)**.

III. DISCUSSION

Desmoplastic small round cell tumor (DSRCT) is an uncommon and aggressive sarcoma that affects adolescent and young adult Caucasian males [1], it has been described for the first time by Gerald and Rosai in 1991 as a result from the chromosomal translocation t (11; 22) (p13; q12) which leads to the fusion of the Ewing's sarcoma gene's (EWS) with the Wilms tumor suppressor gene's (WT1) leading to the formation of the EWSR1-WT1 gene which is an oncogene protein that acts as a transcriptional regulator, modifying gene expression and ultimately allowing tumor growth [4-10].

The DSRCT is a very rare mesenchymal tumor, it is clinically characterized by male predominance (>5:1); the age of predilection is between 18 and 26 years old [4-7].

This type of tumor mainly develops in the peritoneal cavity, but also in other sites such as: ovaries, liver, kidneys, pancreas, bones, heart, ethmoidal sinuses, scalp, hand, paratesticular region, pleura, chest, and posterior cranial fossa [2, 3]. Metastases occur most commonly to the liver, lung, bone marrow, and lymph nodes [2].

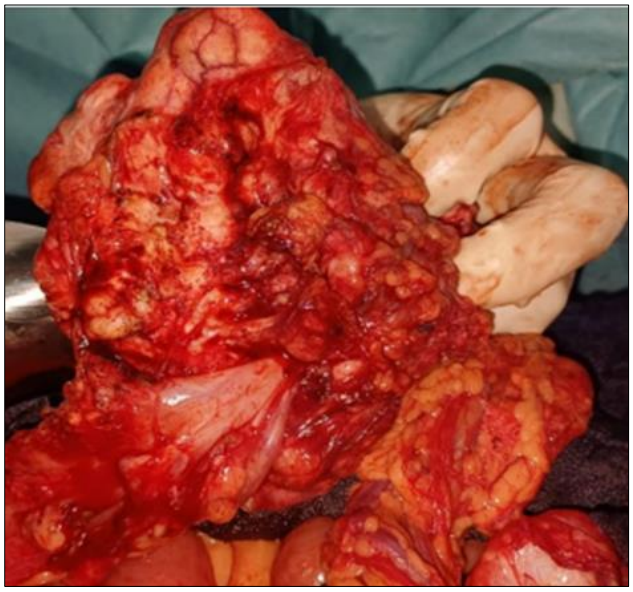

Figure 5: The mass of left hypchondrium adherent to the left colonic angle and great omentum

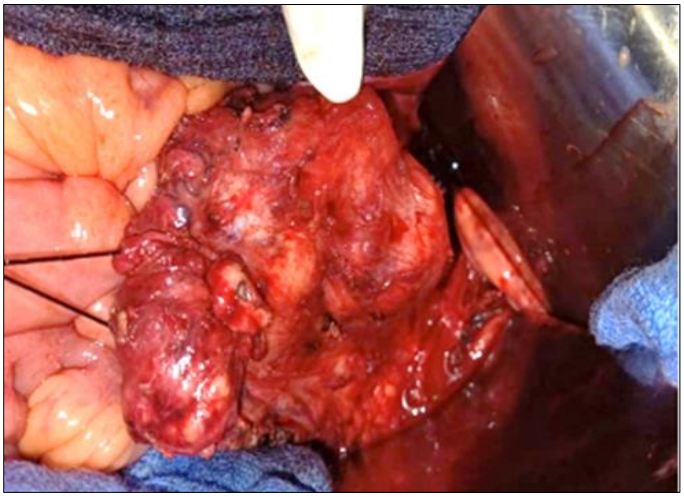

Figure 6: The hypogastric mass between the bladder and the rectum

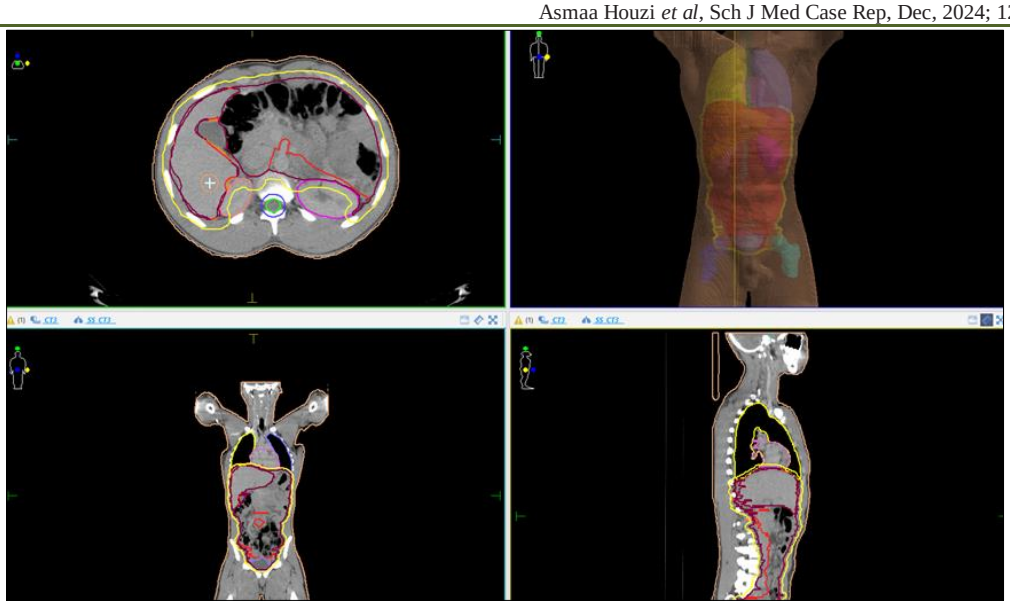

Figure 7: Whole-abdominopelvic intensity-modulated radiation therapy plans for desmoplastic small round cell tumor. Yellow lines indicate prescribed 30-Gy dose area

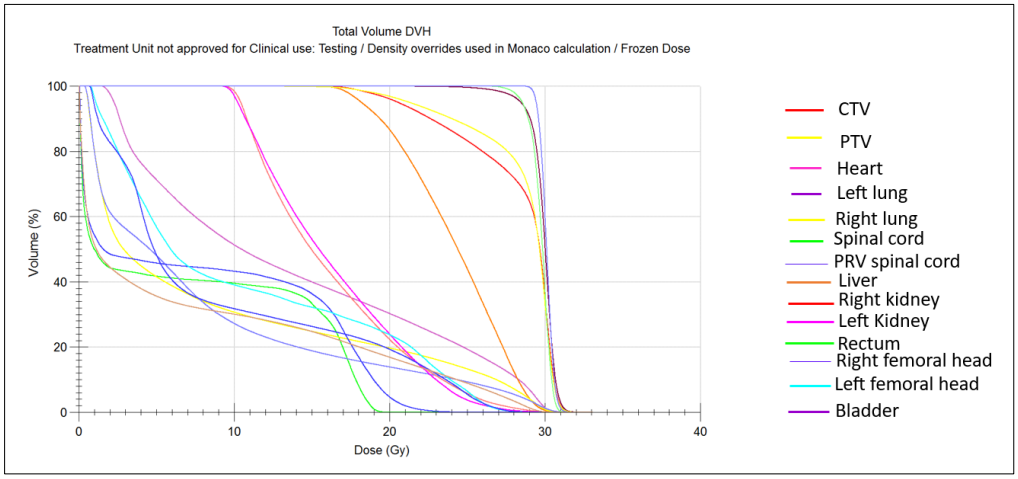

Figure 8: Dose volume histogram for intensity modulation radiation therapy for desmoplastic small round cell tumor

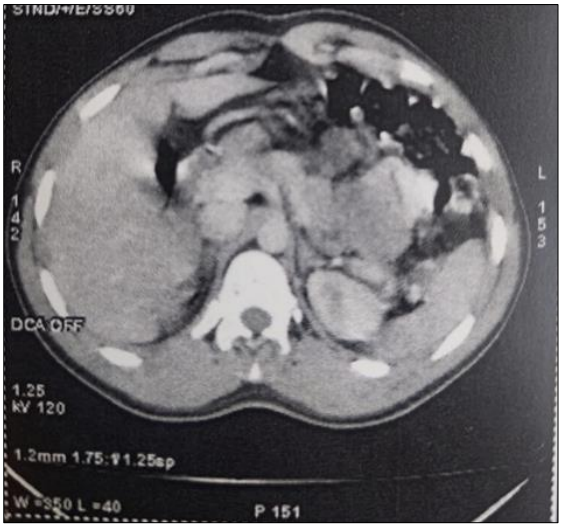

Figure 9A: Abdominal axial CT-scan after multimodal treatment showing disappearance of intersplenorenal mass

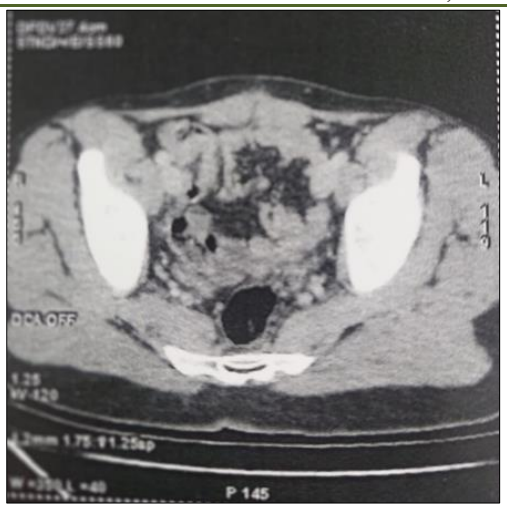

Figure 9B: Plevic axial CT-scan after multimodal treatment showing disappearance of hypogastric mass

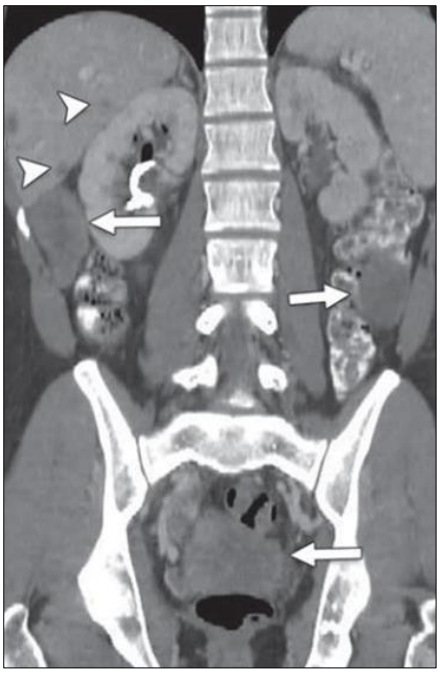

Figure 10: Coronal contrast-enhanced CT image of abdomen and pelvis shows multiple attenuated soft-tissue masses throughout peritoneal cavity and liver metastases [13]

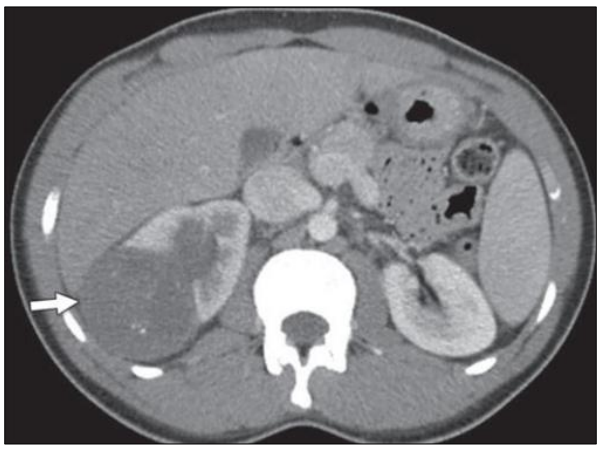

Figure 11: Axial contrast-enhanced CT image of abdomen obtained at presentation shows hypoattenuated right renal mass containing punctate calcifications [13]

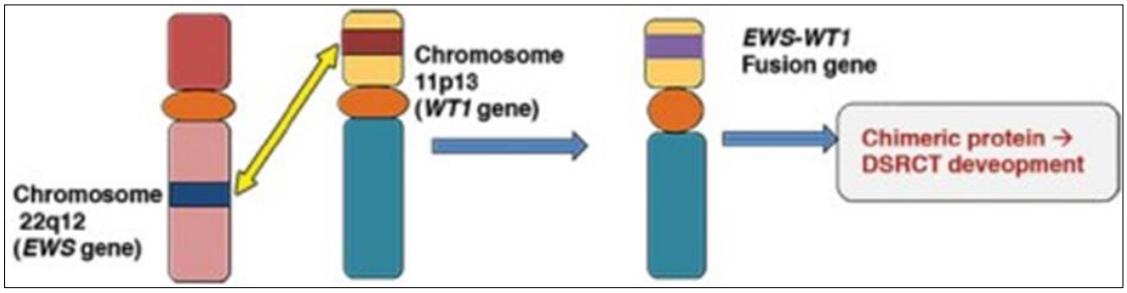

Figure 12: Schematic illustrates t (11;22) (p13;q12) reciprocal translocation that regulates transcriptional activity and downstream pathways resulting in DSRCT [13]

Clinical symptoms are not specific, in most cases, patients present with abdominal pain, abdominal distension or a palpable abdominal mass; we can also observe other symptoms like ascite, urinary disorders, fever, hepatomegaly and peripherical adenopathies. Clinical signs associated with the tumor's mass effect include intestinal obstruction, hydronephrosis and erectile or urinary dysfunction. DSRCT can spread to the peritoneum, omentum and hematogenous metastases mainly to the liver, lungs and bone [3].

Abdominal-pelvic Computed tomography (CT) is the first-line paraclinical examination used when DSRCT is suspected, it shows multiple heterogenous soft-tissue masses in the peritoneal cavity with hypodense areas **(Figure 10)**, it is also rare to find a solitary peritoneal mass **(Figure 11)** [13].

On abdominal-pelvic MRI, DSRCT appears as heterogenous to isointense to hypointense on T1-weighted images and hyperintense on T2-weighted images. 18-FDG PET-scan is indicated of doubt on abdomino-pelvic CT or MRI [13].

Several classifications have been proposed to classify desmoplastic round cell tumors. The most widely recognized is Gilly's classification, which is based on the number and size of lesions **(Table I)** [3].

Table I: Gilly's Classification for desmoplastic small round cellules tumor

Gilly 1	Neoplasic granulations less than 5mm located in a half abdominal space
Gilly 2	Neoplasic granulations less than 5mm located all over the peritoneal space
Gilly 3	Neoplasic granulations with diameter between 5 and 20 mm
Gilly 4	Neoplasic granulations more than 20mm

The diagnosis of DSRCT is based on pathologic, immunohistochemical, and molecular analysis; the tumor has the same characteristics as Ewing sarcomas, primitive neuroectodermal tumor, rhabdomyosarcoma, small cell mesothelioma, neuroblastoma, lymphoma, wilm's tumor; all these tumors are considered differential diagnoses. Pathology reveals Well-defined islands of small, round cells, embedded in abundant desmoplastic stroma, areas of necrosis, cystic degeneration and hemorrhage may be observed, associated hyperplastic and prominent blood vessels may be seen [5-13]. Immunohistochemical reveals the expression of epidermal markers (cytokeratin, Epithelial Membrane Antigen), mesenchymal markers (desmin, vimentin) and neural markers (neuron specific enolase reactivity) [6].

Genetic study by PCR provides the most specific diagnosis, it shows reciprocal chromosomal translocation t (11;22) (p13; q11 or q12), that results from fusion of exon 7 of the Ewing sarcoma (EWS) gene on chromosome 22 with exon 8 of the Wilms tumor suppressor gene (WT1) on chromosome 11, the resultant EWSWT1 chimetric transcript EWS/WTI is diagnostic of this tumor and codes for a protein that acts as a transcriptional activator that fails to suppress tumor growth **(Figure 12)** [5,13].

Due to the rarity of this type of tumor, treatment remains challenging, it includes neoadjuvant chemotherapy followed by debulking surgery and adjuvant radiotherapy for consolidation [8].

Surgery is the first-line treatment, it increases survival rate, with three modalities: Partial tumor resection: resection of >50% of all tumor masses, macroscopic total resection which is resection of all macroscopically visible masses and cytoreduction surgery resection of more than 90% of masses, but with a macroscopic residue [7].

Hyperthermic intraperitoneal chemotherapy (HIPEC) involves applying chemotherapy heated to a high temperature inside the peritoneal cavity, it can be used by used by some teams, the most widely used molecule is cisplatin as 100mg/m2 (90min, 41°C).

HIPEC has been shown to significantly increase overall patient survival in some series however, it causes significant morbidity, ranging from 12% to 52% in adults

but treatment is better tolerated in children, but there is no standardization for using it in DSCRT [11].

DSCRT responds well to chemotherapy, it can be used in a neoadjuvant or adjuvant situation. Neoadjuvant chemotherapy is necessary to reduce tumor volume, there are many regimens adopted but with no particular preference, we can use P6 protocol which combines cyclophosphamide, doxorubicin and vincristine, alternating with ifosfamide and etoposide, other regimens that have been described involve vincristine, ifosfamide, doxorubicin and etoposide (VIDE); vincristine, actinomycin-D ifosfamide and adriamycin (VAIA); a cyclophosphamide, pirarubicin, etoposide and cisplatin (the modified PAVEP regimen). Adjuvant chemotherapy is used by some teams, based on anthracyclines or low dose cyclophosphamide and weekly intravenous vinorelbine, but its use isn't validated yet [11]. Exclusive chemotherapy is reserved for metastatic patients [14].

Whole abdominal radiotherapy (WART) with IMRT is used after induction chemotherapy and surgery, it targets areas of residual disease while sparing normal tissues [9], for delineation volumes, the clinical target volume (CTV) corresponds to entire peritoneal and involved retroperitoneal areas, excluding the uninvolved kidneys and liver, the planning target volume (PTV) corresponds to CTV with a 10mm margin, organs at risk that should be contoured are : liver, kidneys, lung, heart, bladder and rectum. The recommended dose is 30 Gy in 20 sessions with conventional fractionation of 1.5 Gy with 6MV photons, with or without a focal boost of 6-10 Gy on residual disease. Using radiosensitizing chemotherapy is often necessary. The most commonly used molecules are Temozolomide, irinotecan, cyclophosphamide, vinorelbine and bevacizumab. The toxicities reported after radiotherapy were mainly digestive (nausea, vomiting, diarrhea grade 1 and 2) and hematological such as anemia and thrombocytopenia [1-11].

DSRCT is a tumor with a poor prognosis, the 3-years overall survival (OS) is 55% after a complete multimodal treatment versus 27 % if one of modalities is missing [14], survival rate at 5 years is less than 15% [13]. There are some factors that increases the 3-OS like: the absence of metastasis and complete surgery [14].

Multimodal treatment remains inadequate in desmoplastic round cell tumors, given the low 5-year survival rate. In vivo, studies have shown the role of the cdk4/6 inhibitor palbociclib, explained by the fact that the EWSR1/WT1 protein plays a dominant role in transcription by binding to the cyclin D1 (CCDN) promoter and stimulating DSRCT growth via the cyclin D-CDK4/6-RB axis, so inhibition of this axis by palbociclib will reduce tumor growth in DSRCT [12].

Further investigations and prospective studies are required in order to define the optimal therapeutic strategy for this disease.

IV. CONCLUSION

Desmoplastic round cell tumor is an uncommon tumor of the young subject, it must be evoked in the presence of adequate imaging, the proposed treatment is multimodal chemotherapy, surgery followed by radiochemotherapy, but despite this, the prognosis remains poor and the survival rate very low. Future genetic therapies focused on developing targeted immunotherapy, might promise more optimistic results. Until then, more than one medical specialities should collaborate in order to face the challenge and treat such patients.

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