# Desmoplastic Small Round Cell Tumor: Case Report

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## *Tumor Desmoplásico de Pequenas Células Redondas: Relato de Caso*

## Tumor Desmoplásico de Células Redondas y Pequeñas: Informe de Caso

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ABSTRACT

**Introduction:** Desmoplastic small round cell tumor (DSRCT) is an extremely rare and aggressive sarcoma of mesenchymal origin that mainly affects the pelvic and abdominal area of the body. DSRCT is rare and occurs more frequently in young white men between the second and third decades of life. Some reports state that, since its discovery, only about 200 cases of DSRCT have been recorded since 1989. **Case report:** A 20-year-old young adult male presented with abdominal discomfort associated with loss of appetite and weakness for four months, with a weight loss of 10 kg during this period. Findings from complementary examinations included significant hepatosplenomegaly and a pelvic abdominal mass with hepatic and pulmonary nodules. The anatomopathological report of a neoplasm of small round blue cells, associated with the immunohistochemistry available at the service, the epidemiology, and clinical presentation, led to the definition of a presumptive diagnosis of DSRCT and the initiation of immediate systemic therapy. **Conclusion:** The DSRCT, despite the existing scientific knowledge, and due to its rarity and the limited information on diagnosis and therapy, remains a medical challenge.

**Key words:** Desmoplastic Small Round Cell Tumor; Sarcoma; Neoplasms/epidemiology; Adult; Case Reports.

RESUMO

**Introdução:** O tumor desmoplásico de pequenas células redondas (TDPCR) é um sarcoma extremamente raro e agressivo de origem mesenquimal que acomete, principalmente, a área pélvica e abdominal do corpo. O TDPCR é raro e acomete com mais frequência homens brancos jovens, entre a segunda e a terceira décadas de vida. Alguns relatos dizem que, desde sua descoberta, apenas cerca de 200 casos de TDPCR foram registrados desde 1989. **Relato do caso:** Homem adulto, jovem, com 20 anos de idade, apresentou desconforto abdominal associado à inapetência e fraqueza há quatro meses, com uma perda ponderal de 10 kg no período. Os achados dos exames complementares incluíam hepatoesplenomegalia importante e massa abdominal pélvica com nódulos hepáticos e pulmonares. O laudo anatomopatológico de neoplasia de pequenas células redondas e azuis associado à imuno-histoquímica disponível no serviço, a epidemiologia e a clínica do paciente permitiram estabelecer o diagnóstico presuntivo de TDPCR e iniciar a terapêutica sistêmica imediata. **Conclusão:** O TDPCR, apesar do conhecimento científico-literário, e por conta de sua raridade e da informação sobre diagnóstico e terapêutica ser limitada, ainda continua como um desafio dentro da área médica.

**Palavras-chave:** Tumor Desmoplásico de Pequenas Células Redondas; Sarcoma; Neoplasias/epidemiologia; Adulto; Relato de Caso.

RESUMEN

**Introducción:** El tumor desmoplásico de células pequeñas y redondas (TDCPR) es un sarcoma extremadamente raro y agresivo de origen mesenquimatoso que afecta principalmente la región pélvica y abdominal del cuerpo. El TDCPR es raro y afecta con mayor frecuencia a hombres blancos jóvenes, entre la segunda y tercera década de vida. Algunos informes señalan que, desde su descubrimiento, solo se han registrado alrededor de 200 casos de TDCPR desde 1989. **Informe del caso:** Hombre adulto joven, 20 años presentó malestar abdominal asociado con inapetencia y debilidad durante cuatro meses, con una pérdida de peso de 10 kg en ese período. Los hallazgos de los exámenes complementarios incluían hepatoesplenomegalia importante y masa abdominal pélvica con nódulos hepáticos y pulmonares. El informe anatomopatológico de neoplasia de células pequeñas, redondas y azules, asociado con la inmunohistoquímica disponible en el servicio, la epidemiología y la clínica del paciente permitieron establecer un diagnóstico presuntivo de TDCPR y el inicio de la terapia sistémica inmediata. **Conclusión:** El TDCPR, a pesar del conocimiento científico y literario existente, y debido a su rareza y la información limitada sobre diagnóstico y terapia, sigue siendo un desafío dentro del campo médico.

**Palabras clave:** Tumor Desmoplásico de Células Pequeñas y Redondas; Sarcoma; Neoplasias/epidemiologia; Adulto; Informes de Casos.

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## INTRODUCTION

Desmoplastic small round cell tumor (DSRCT) is a rare malignant mesenchymal neoplasm composed of small, round tumor cells of scarce and basophilic cytoplasm immersed in abundant fibrous stroma that predominantly affects young men between the second and third decade of life[1,2].

Its physiopathology involves the translocation t(11;22)(p13;q12) which creates the EWSR1-WT1 fusion gene, and produces a chimeric protein that acts as a transcriptional regulating activity[3,4]. Most of the tumors develops in the abdomen with local dissemination and multiple serous implants, being uncommon extraperitoneal presentations[5-7].

Clinically, it manifests with pain or abdominal distension, palpable mass, inappetence, weight loss and ascites. The integration of histopathological, immunohistochemical and molecular findings[8-12] supported the evaluation. Histologically, it characterizes by nests of small round blue cells immersed in desmoplastic stroma. The immunohistochemistry revealed multilinear expressions, including epithelial (cytokeratins), muscular (dot-like pattern desmin) markers, and occasionally neuronal[10,11].

The diagnosis is defined by the detection of the translocation t(11;22)(p13;q12), that creates the EWSR1–WT1 fusion gene, being identified by techniques such as FISH or RT-PCR[11,12]. In the absence of molecular confirmation, the diagnosis can be considered presumptive or highly suggestive when there is strong correlation among the clinical histopathological or immunohistochemical findings, except relevant differential diagnoses[10,12].

The prognosis is reserved and nearly 60% to 70% of the patients die in three years even with multimodal treatment[13,14].

This report followed the recommendations of CARE Case Report Guidelines for clinical case reporting[15] and approved by the Ethics Committee of "*Universidade Federal de Santa Catarina*", report number 7,104,186 (CAAE (submission for ethical review): 82574924.0.0000.0121) in compliance with Directive 466/2012[16] of the National Health Council (CNS).

## CASE REPORT

Three weeks prior to admission, the patient was submitted to an abdominal ultrasound (US) which revealed moderately enlarged and heterogeneous liver, with diffuse fine dotted infiltrate and three mixed texture hepatic cysts with hyperechogenic predominance suggestive of chronic hepatopathy.

At day 1 of admission (January 27, 2023) the 20-year old male patient, smoker, living in the city of Palhoça (SC) was admitted to "*Hospital Universitário Polydoro Ernani de São Thiago*" in Florianópolis (SC) due to abdominal mass. Reported asthenia for four months, inappetence, right hypochondriac bulge and 10 kg-loss. Denied fever, abdominal pain, myalgia, pruritus and gastrointestinal alterations. There was no relevant prior personal history, his mother had breast neoplasm.

At the examination, was thin, lucid and oriented, with painless lymph node in left supraclavicular fossa. Tachycardic, reduced right vesicular murmur and distended abdomen with hepatomegaly and firm painless mass in right iliac fossa (Figure 1).

Lab tests revealed important elevation of AST/ALT (170/403 U/L) and GGT/FA (300/485 U/L), compatible with mixed pattern of cholestatic hepatocellular injury, indicative of acute liver lesion combined with tumor infiltration. AFP (alpha-fetoprotein) at normal levels (3.5 ng/mL), excluding hepatoid component or hepatocarcinoma. Preserved complete blood count (CBC), liver, renal functions and metabolic tests.

In day 2 from admission, kept mixed injury pattern with preserved bilirubin, albumin and INR. Investigation for infections (EBV, hepatitis B and C, HIV) and autoimmune was negative. The iron status profile revealed low sideremia, reduced saturation of transferrin and ferritin discreetly elevated, compatible with chronic disease anemia.

A new CBC showed mild normocytic anemia with presence of poikilocytosis and ovalocytes without leukocytosis or thrombocytopenia. Pro-BNP was mildly elevated without clinical repercussions of cardiac failure. Renal, electrolytic and lipid functions were normal.

Abdomen and pelvis computed tomography revealed voluminous mass of heterogeneous lymph nodes conglomerates, hypoattenuation

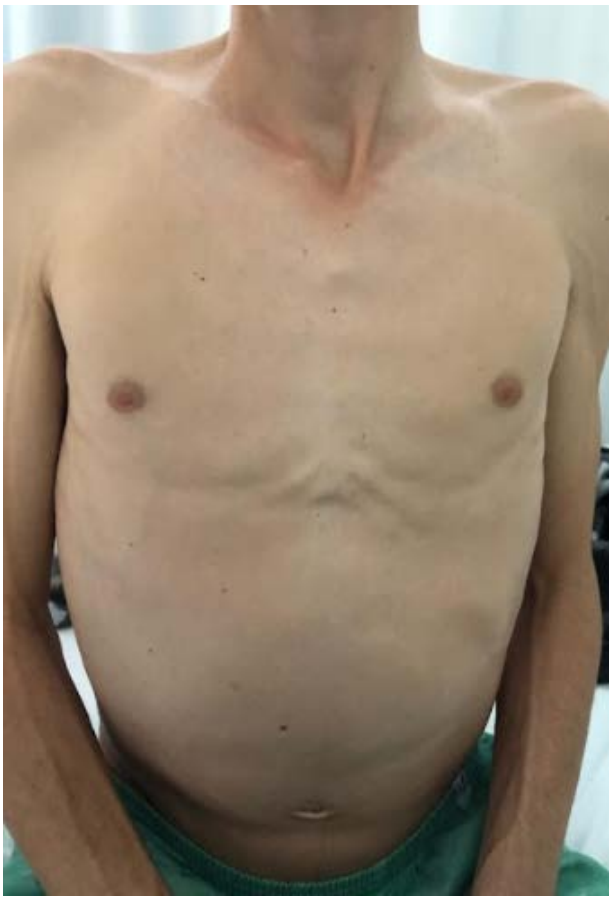

**Figure 1.** Patient with significant hepatosplenomegaly, fixed abdominal mass and left supraclavicular lymphadenomegaly

and peripheral enhancement, filling the abdominal cavity and retroperitoneum. Important hepatosplenomegaly was observed in addition to multiple secondary implants, moderate ascites, and lung metastases compatible with disseminated metastatic disease (Figure 2).

On day 3 from admission, diagnostic thoracocentesis revealed citrine exudative pattern and presence of atypical, isolate and grouped cells, of irregular chromatin and basophilic cytoplasm compatible with metastatic dissemination.

Biochemistry glucose of 110 mg/dL, elevated LDH, total protein of 3 g/dL and albumin of 2.4 g/dL, with a serum-pleural albumin gradient of <1.1 g/dL, confirming exudate by Light's criteria. Bacterioscopy and BAAR were negative, the hematological function and biochemistry were kept stable.

Immunophenotyping by flow cytometry revealed predominance of lymphocytes T CD3+/CD5+, with TCD4+/TCD8+ of 2.7. 6.4% of mature B lymphocytes were identified as well (CD19+, CD20+, CD45++), relation Kappa/Lambda of 1.5, in addition to significant population of monocytes/macrophages (CD14+, CD45++) in 38% of the sample. Remarkably, 7.7% of non-hematopoietic cells (CD45-) with positive strong expression of pancytokeratin and Ber-EP4 (EPCAM) were compatible with glandular epithelial cellular infiltration. The immunophenotypic profile confirmed metastatic epithelial infiltration without evidence of lymphoma.

In the same day, US-guided percutaneous liver biopsy was performed for histopathological analysis.

In day 9 from admission, the anatomopathology showed proliferation of small round blue malignant cells infiltrating the hepatic parenchyma. Immunohistochemistry was positive for cytokeratin (diffuse granular pattern and perinuclear dot), in addition to expression of low molecular weight cytokeratin (CAM 5.2). Synaptophysin, chromogranin A and CD45 were negative, excluding neuroendocrine and hematolymphoid lineage.

All the findings led to the presumptive diagnosis of DSRCT although without definitive confirmation due to the absence of evaluation of WT-1 (C-terminal domain) and molecular tests unavailable at the service.

In day 10 from admission evolved to tachypnea associated with moderate right pleural effusion and bilateral diffuse nodular infiltrate. Was submitted to relief thoracocentesis with immediate clinical improvement (Figure 3).

The patient was emotionally stable during the visits supported by his aunt, his main caregiver, and the palliative care multiprofessional team.

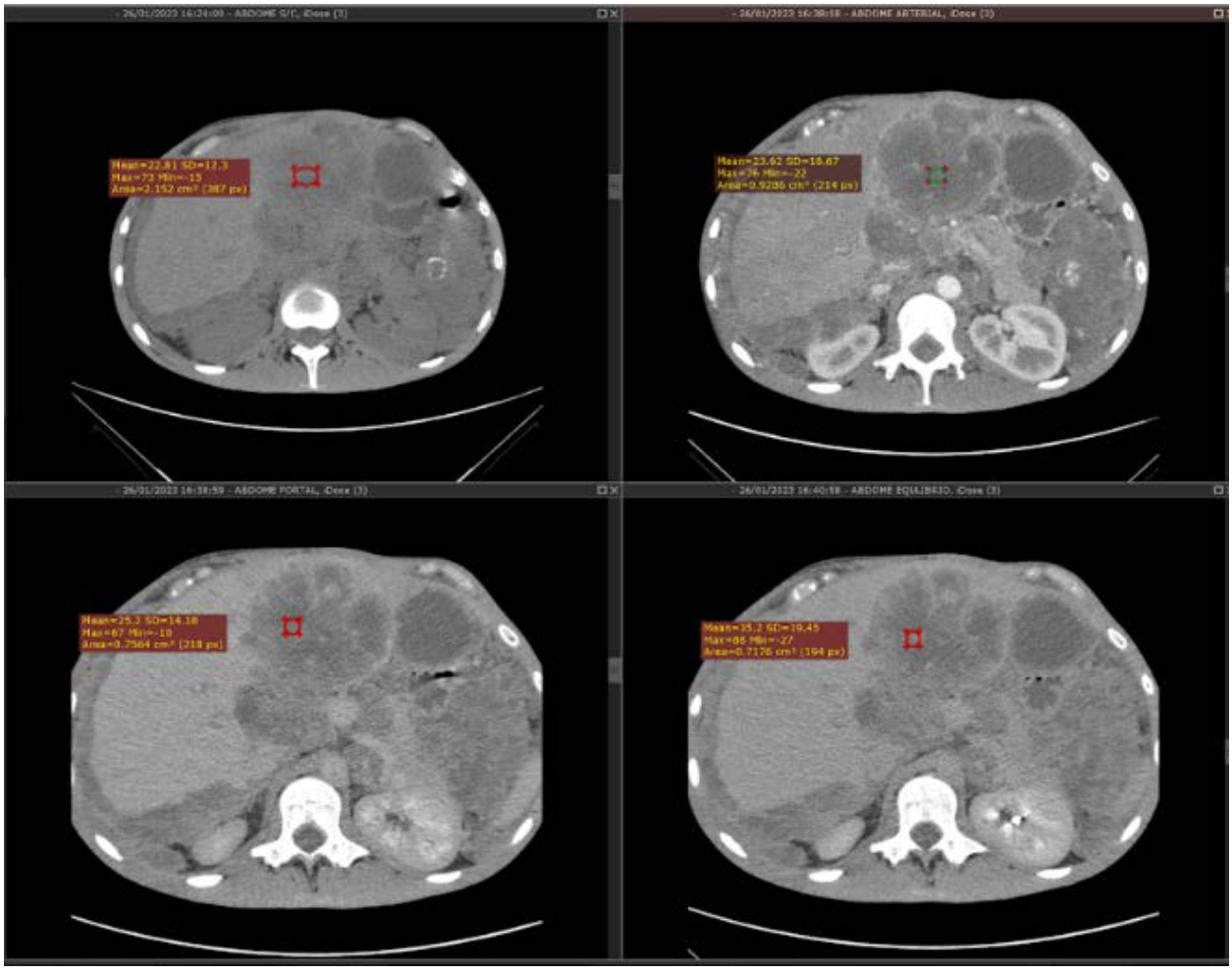

**Figure 2.** Voluminous lesion involving abdominal, retroperitoneal and pelvic organs with hepatosplenomegaly, multiple secondary implants and moderate ascites

In day 11 from the admission, upon evaluation of the oncologic team and due to diagnostic and therapeutic limitations of the service, it was decided to transfer the patient to a reference oncologic unit, the "*Centro de Pesquisas Oncológicas (CEPON)*" for molecular confirmation and systemic treatment. Still as an inpatient at the university hospital, was submitted to the first chemotherapy cycle. He continued hospitalized awaiting transference in clinical surveillance.

In face of stage IV presumptive diagnosis of DSRCT and disseminated metastatic disease and considering the possibility of confirmation of WT1 (C-terminal domain), FISH or RT-PCR either at the university hospital or at CEPON, it was decided to apply immediate systemic chemotherapy. The patient understood his condition and received psychological support which was given to his family too. The first chemotherapy cycle was applied at the university hospital based on protocols for aggressive sarcomas (doxorubicin, cyclophosphamide, mesna (uroprotection) and diphenhydramine).

After clinical stabilization, the patient was transferred to CEPON for specialized oncologic treatment where he received six cycles of the protocol VAC (vincristine, actinomycin D and cyclophosphamide), in addition to the seventh cycle of VAC without doxorubicin followed by an alternate cycle with etoposide and ifosfamide mesna (VP-16 + IFO/mesna) in March 2023.

Despite the intensive chemotherapy, there was no significant clinical or radiological response keeping hepatic and pulmonary lesions. Upon conclusion of the initial cycle at CEPON, the patient was discharged for oncologic outpatient follow up. He progressed to clinical deterioration with weight loss, voluminous ascites, refractory thoracic pain, edema and important dyspnea.

In August 2024, after oncologic revaluation, his functional performance was compromised (ECOG 3-4), and presented severe liver dysfunction and active progression of the disease. Due to poor clinical conditions to continue the chemotherapy and irreversibility of the status, it was decided to discontinue the oncologic treatment and referral to exclusive palliative care, providing the patient guidance about the expected evolution of the disease, symptoms control and family support.

The patient received symptomatic control, relief paracentesis (interrupted by pain) and oral morphine was prescribed in addition to detailed considerations on household management and warning signs. Progressed to death on August 22, 2024, nine days after the transition to exclusive palliative care with global survival of 19 months post diagnosis.

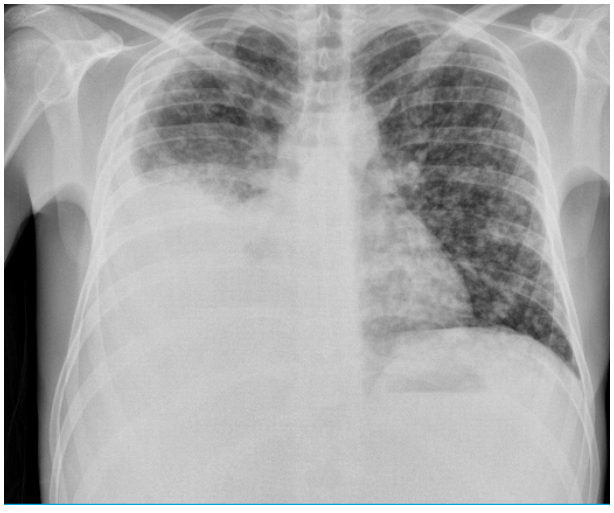

**Figure 3.** Multiple small nodular formations randomly distributed in the lower part of the lungs with right pleural effusion of 4.1 cm, causing passive atelectasis of the adjacent lung parenchyma

## DISCUSSION

The DSRCT is a rare and aggressive malignant neoplasia predominantly affecting young men mostly in the abdomen. Described by Gerald and Rosai in 1989, it is histologically characterized by nests of small, round blue cells with multilinear immunohistochemical expression, expanding the differential diagnosis with lymphoma, germinative tumors, neuroendocrine carcinomas and other sarcomas[5].

In the present case, the positivity for vimentin suggests mesenchymal origin while desmin marker indicates myogenic differentiation, favoring the hypothesis of sarcoma. The positivity for cytokeratin excludes seminoma, while the negativity for synaptophysin, chromogranin A and CD45 made improbable neuroendocrine neoplasia and hematolymphoid disease respectively. The absence of CD99 excluded Ewing sarcoma.

The exclusion of relevant differential diagnoses associated with histomorphological pattern of small, round, blue cells infiltrating the hepatic parenchyma allowed to establish a presumptive diagnosis of DSRCT.

The immunohistochemical evaluation for WT1 (C-terminal domain) as well as the molecular confirmation of translocation t(11;22)(p13;q12) involving the rearrangement EWSR1–WT1 could not be performed either in the university hospital or in CEPON due to technical unavailability. In an ideal scenario, these tests would constitute the subsequent phase for definitive confirmation of the diagnosis. Even though, the literature reports that in scenarios of diagnostic limitation, the strong clinicopathological correlation and systematic exclusion of the main differential diagnoses allow the presumptive diagnostic classification with high degree of plausibility[2,3,6,8].

Despite the diagnostic advances, DSRCT remains a therapeutic challenge with reserved prognosis and survival often below three years even with multimodal treatment. The rarity of the neoplasm and the absence of specific guidelines limits the therapeutic standardization and development of more effective targeted strategies[2,13].

This report described a typical case of DSRCT with predominant liver involvement in a young man then healthy, and uncommon presentation in face of the peritoneal or retroperitoneal pattern[6]. The silent evolution with unspecified symptoms and late

diagnosis reinforces the necessity of considering rare neoplasms in young patients with persistent constitutional symptoms.

The case contributes to the literature while emphasizing: (1) atypical clinical presentations predominantly in the liver; (2) relevance of the biopsy associated with immunohistochemical panel and, ideally, molecular tests for confirmation of EWSR1–WT1 rearrangement; (3) therapeutic and diagnostic challenges in limited access settings such as the National Health System (SUS); and (4) the importance of early integration of palliative care due to the aggressiveness of the disease.

In a scenario of paucity of reports, especially in Latin America, this case reinforces the necessity of early recognition of atypical presentations and timely referral to specialized centers.

## CONCLUSION

This report exposes the diagnostic complexity and aggressiveness of DSRCT, most of all in atypical presentations with predominant involvement of the liver. In the absence of molecular confirmation and evaluation of WT1 (C-terminal domain), the diagnosis was determined as presumptive based on the strong clinicopathological correlation and exclusion of relevant differential diagnoses.

The importance of the integration of clinical, radiological, histopathological and immunohistochemistry data was instrumental for the proper diagnostic definition, especially in a context of limited resources and when the clinical status does not allow invasive approaches. Early detection and agile referral to specialized centers are critical.

In advanced diseases, early introduction of palliative care is essential to ensure the quality-of-life and dignity of the patient. The case reinforces the necessity of considering DSRCT in differential diagnosis of hepatic neoplasms in young patients and underpins the central role of image-guided biopsy and immunohistochemistry.

The patient and her main caregiver understood the severity and irreversible progression of the disease, concurring with the therapeutic proposal and transition to exclusive palliative care prioritizing comfort and dignity.

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### CONTRIBUTIONS

Geraldo Lucas Lopes Costa and Leonardo Andrighetti contributed substantially to the study concept and design, acquisition, analysis and interpretation of the data, writing and critical review. Daniella Serafin Couto Vieira contributed substantially to the study conception and design, analysis and interpretation of the data and critical review. All the authors approved the final version to be published.

### DECLARATION OF CONFLICT OF INTEREST

There is no conflict of interest to declare.

### DATA AVAILABILITY STATEMENT

All the content underlying the text is contained in the article.

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None.

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