



# Desmoplastic Small Round Cell Tumor. A Case Report and Literature Review

Tumor desmoplásico de células pequeñas y redondas. Presentación de caso y revisión de la literatura

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## Palabras clave (DeCS)

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## Summary

Desmoplastic small round cell tumor (DSRCT) is a rare malignant neoplasm with an aggressive clinical course and high mortality. The case of a 21-year-old man is presented, who consulted for abdominal pain of moderate intensity radiating to the right flank, fever and weight loss. Contrast abdominal tomography was performed, documenting a large intraperitoneal mass with areas of central necrosis and extension to the pelvis, in addition to secondary neoplastic liver lesions. The diagnosis was confirmed by ultrasound-guided percutaneous biopsy, which reported extensive infiltration by malignant tumor, consisting of cells with vesicular nuclei of clear chromatin, eosinophilic cytoplasm and immunohistochemistry compatible with said tumor. This case report is compared with the findings described in other series published in the literature and a clinical review of the subject is made.

## Resumen

El tumor desmoplásico de células pequeñas y redondas (TDCPR) es una neoplasia maligna rara, con curso clínico agresivo y mortalidad elevada. Se presenta el caso de un hombre de 21 años de edad, quien consultó por dolor abdominal de intensidad moderada, irradiado al flanco derecho, fiebre y pérdida de peso. En tomografía abdominal con medio de contraste se documentó una gran masa intraperitoneal con áreas de necrosis central y extensión a la pelvis, además de lesiones hepáticas de aspecto neoplásico secundario. El diagnóstico se confirmó mediante biopsia percutánea guiada por ultrasonido, que mostró extensa infiltración por tumor maligno, constituido por células con núcleos vesiculosos de cromatina clara, citoplasma eosinófilo e inmunohistoquímica compatible con dicho tumor. En este artículo se hace una confrontación del caso con los hallazgos descritos en otras series publicadas en la literatura y una revisión clínica del tema.

## Introduction

Desmoplastic small round cell tumor (DSRCT) is a rare malignant neoplasm, which characteristically occurs in white children, adolescents and young adults (1). DSRCT was first first described as a pathologic entity by Gerald et al. in 1991, and few cases have been reported in the medical literature (2). The most common imaging finding is significant peritoneal involvement, usually multiple soft tissue masses dependent on the mesentery or omentum with no apparent origin in another solid organ. There are also usually enlarged lymph nodes (>1.5 cm) and low-signal adenopathy with loss of fatty hilum(3); liver metastases are found in up to one-third of patients and can be seen as low-signal nodules of variable size -0.8-12.7 cm and number - single or

multiple (4). Histologically, DSRCT is characterized by well-defined nests of small, round tumor cells embedded in an abundant desmoplastic stroma (5). It has an aggressive clinical course, with a mortality rate of 70%, three years after diagnosis (6). We present the case of a 21-year-old man with DSRCT.

## Presentation of the case

A 21-year-old male patient, with no relevant history, consulted for abdominal pain in the right flank, associated with fever and weight loss. Ultrasonography revealed a large abdominal-pelvic mass of heterogeneous echogenicity, with peripheral solid component, partially defined contours and central anechoic areas due to probable necrosis, measuring

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12 × 12 cm (AP×T); likewise, multiple focal hepatic lesions were identified, rounded, isoechoic with respect to the hepatic parenchyma, with central hypoechoic areas (Figures 1 and 2). Computed tomography (CT) of the abdomen with contrast medium confirmed an intraperitoneal mass measuring 14 × 12 cm (AP×T) with epicenter in the mesentery of the right hypochondrium, without involvement of adjacent organs, with heterogeneous density and solid peripheral areas that enhanced after contrast administration and significant central areas of low attenuation due to necrosis; the mass displaced the thin intestinal loops and mesenteric vessels without infiltrating them (Figure 3). In addition, a second mass of pelvic location was found, bilobulated, with a predominantly solid component that enhanced with contrast medium and central areas of necrosis, which displaced the posterior wall of the bladder and the anterior wall of the rectum without infiltrating them (figure 4). The hepatic lesions were rounded, hypoechoic, with central necrosis and involved both hepatic lobes (figure 5). An ultrasound-guided biopsy of one of the hepatic lesions with pathology was performed, which showed extensive infiltration by a malignant tumor arranged in sheets, consisting of cells with vesicular nuclei of clear chromatin, eosinophilic cytoplasm; immunoperoxidase studies were performed which confirmed the diagnosis (figure 6). The patient was treated with two cycles of chemotherapy, as well as bilateral nephrostomies due to obstructive renal lesions. Despite the management, the patient died one month after being admitted to the hospital.

## Discussion

Desmoplastic small round cell tumor (DSRCT) is a rare malignant neoplasm belonging to the histological descriptive category called small round and blue cell tumors, which also includes neuroblastoma, Wilms' tumor, rhabdomyosarcoma, Ewing's sarcoma, primitive neuroectodermal tumor, osteosarcoma, poorly differentiated synovial cell sarcoma and lymphomas (7). Since DSRCT show mesenchymal, epithelial and neural differentiation, immunohistochemistry allows them to be differentiated from other small round cell tumors (2, 7). In addition, DSRCT contains a specific chromosomal abnormality: the translocation (t11;21)(p13;q12) with subsequent fusion of the genes of Ewing's sarcoma and Wilms' tumor (4).

The annual incidence of DSRCT in the United States is 0.74 cases per million population, with prevalence in individuals aged 20 to 24 years (8). The clinical presentation is nonspecific, with abdominal pain, bloating, changes in bowel habitus, and sensation of palpable mass in the abdomen most commonly found (4, 9). The clinical manifestations are usually related to the anatomical site involved, so ureteral obstruction and secondary hydronephrosis, intestinal obstruction, obstructive biliary syndrome (10) and even increased voiding frequency due to bladder compression (11) are found in the literature.

The characteristic imaging finding is that of multiple soft tissue masses located most frequently in the pelvis (rectovesical and rectouterine spaces) and, secondly, in the omentum and mesentery of the peritoneal cavity, without a defined organ from which they originate (4, 9, 11). Presentation as a solitary peritoneal mass rarely corresponds to a DSRCT. In a study of nine cases by Pickhardt et al. (3),

only two of them corresponded to DSRCT that debuted as solitary abdominal masses. In CT, these masses usually show low-attenuation foci reflecting necrosis, hemorrhage or marked desmoplasia with a solid component that enhances heterogeneously with the contrast medium (12, 13). Areas of irregular peritoneal thickening and ascites have also been described as imaging findings in CT (6).

The dissemination pattern of the disease includes direct seeding through the peritoneal surfaces, as well as lymphatic and hematogenous dissemination (14). The liver parenchyma is the most frequent site of extraperitoneal involvement by DSRCT, followed by the lung, bone, spleen, pleura and soft tissue (14, 15). In the series described by Thomas et al. (16), adenomegaly was demonstrated in 50% of patients, with involvement of the following abdominal lymph node chains: para-aortic (100%), aortocaval (71%), portocaval (50%) and mesenteric (13%). Mediastinal adenomegaly was observed in 20 % of the cases.

In magnetic resonance imaging (MRI), DSRCT is observed as heterogeneous signal masses -predominantly medium or low signal intensity- or low signal with respect to the muscle in sequences with T1 information and high signal in sequences with T2 information; the enhancement, after the administration of paramagnetic contrast medium, is equally heterogeneous (12, 16, 17). The increase in signal intensity in sequences with T1 information and the presence of liquid-liquid levels in sequences with T2 information suggest intratumoral hemorrhage (17). Tumor calcifications have been documented in different cohorts (3-5) between 22 and 28% of patients.

Positron emission tomography (PET/CT) provides useful information about the metabolic activity of the tumor and helps to establish its stage, as well as detecting occult metastases not visualized in CT and MRI (18). PET/CT is also useful to evaluate the response to treatment and to define additional conducts or changes in therapy. The decrease in the metabolic activity of the tumor after initiating treatment can be observed even before the size of the masses is reduced or evident morphological changes occur (19).

Regarding treatment, an optimal management strategy for patients with DSRCT is still unknown. To date, the largest study that sought to establish management and survival patterns in patients with DSRCT was published by Gani et al. (18), who, through a retrospective analysis of the National Cancer Data Base of the United States, were able to identify 419 patients over 18 years of age with a diagnosis of DSRCT; of these, 86.5% received chemotherapy, 41.2% underwent surgical management and 13% received radiotherapy. During the study, 69.7 % of the patients died with a survival of 25.9 months. Survival at 1, 3 and 5 years was 78.6 %, 32.3 % and 18.4 %, respectively. Multivariate analysis showed that surgical management (HR = 0.68, 95 % CI [0.50-0.91]), radiotherapy (HR = 0.55, 95 % CI [0.33-0.92]) and chemotherapy (HR = 0.52, 95 % CI [0.35-0.78]) were independent factors associated with higher survival rates. In contrast, stage IV disease (HR = 2.12, 95 % CI [1.41-3.18]) and pre-existing comorbidities were associated with worse prognosis. The authors concluded that, although multimodality treatment in patients with DSRCT may lead to improved survival, further research and clinical trials are still required to establish the best management guidelines (18).

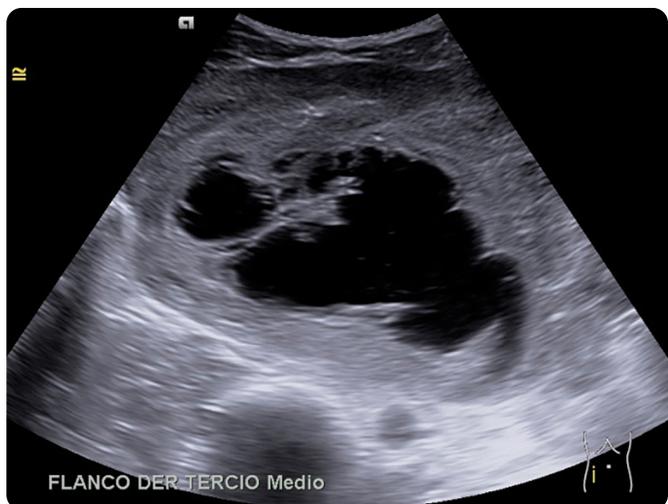


Figure 1. Abdominal ultrasound. Intra-abdominal mass of 12 × 12 cm, isoechoic with respect to the liver parenchyma, with central anechoic area corresponding to necrosis or cystic degeneration.

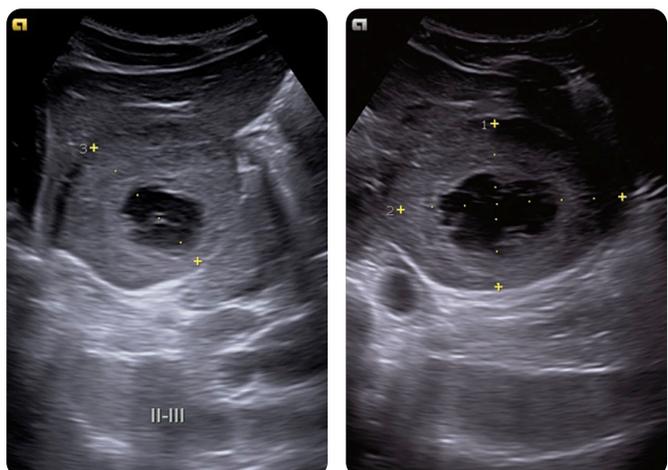


Figure 2. Focal lesion of the III hepatic segment, solid in appearance, with central anechoic area of 7.6 × 5.6 cm, probably due to necrosis. Similar characteristics to those of the abdominal mass.

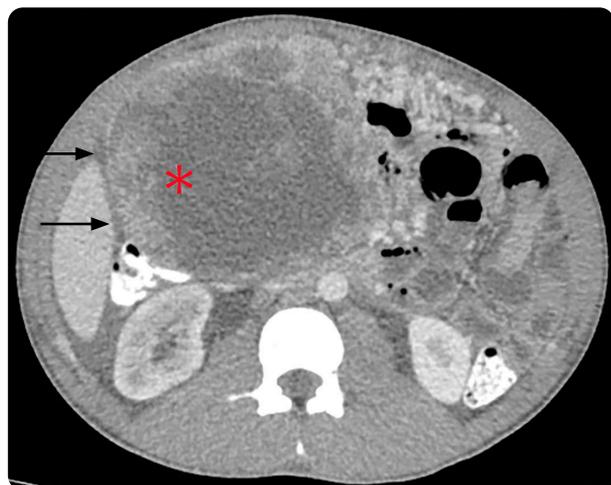


Figure 3. Abdominal CT with contrast medium in portal phase. Intra-abdominal mass, multilobulated, heterogeneous density with central areas of low attenuation due to necrosis (\*) and peripheral solid component that enhances with contrast medium (black arrows).

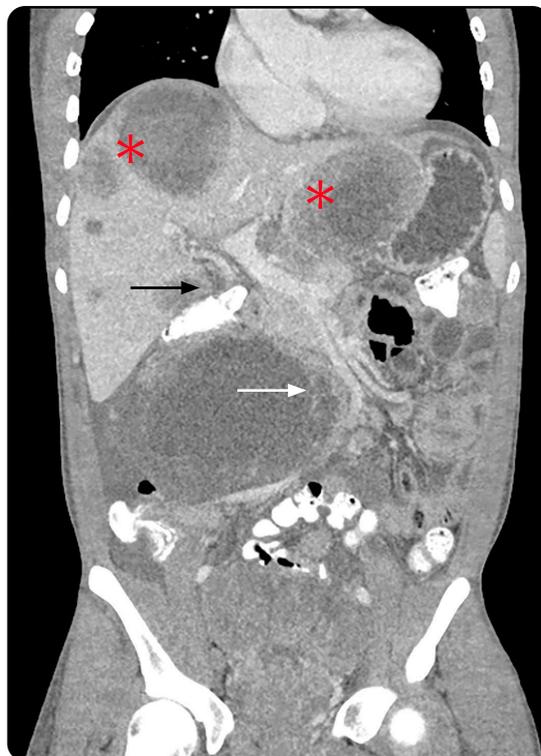


Figure 4. Coronal reconstruction: multiple focal lesions in both hepatic lobes, rounded, low signal and with slight enhancement after contrast medium administration (\*). There is no dilatation of the biliary tract and the portal is permeable (red arrow). It is also observed how the peritoneal mass displaces the mesenteric vessels without infiltrating them (white arrow).



Figure 5. Sagittal reconstruction. A second bilobulated mass is observed, located in the rectovesical space of the pelvis, predominantly solid, with central areas of low attenuation and peripheral enhancement with contrast medium. It displaces and compresses the posterior wall of the bladder (\*) and anterior wall of the rectum (black arrow) without infiltrating them.

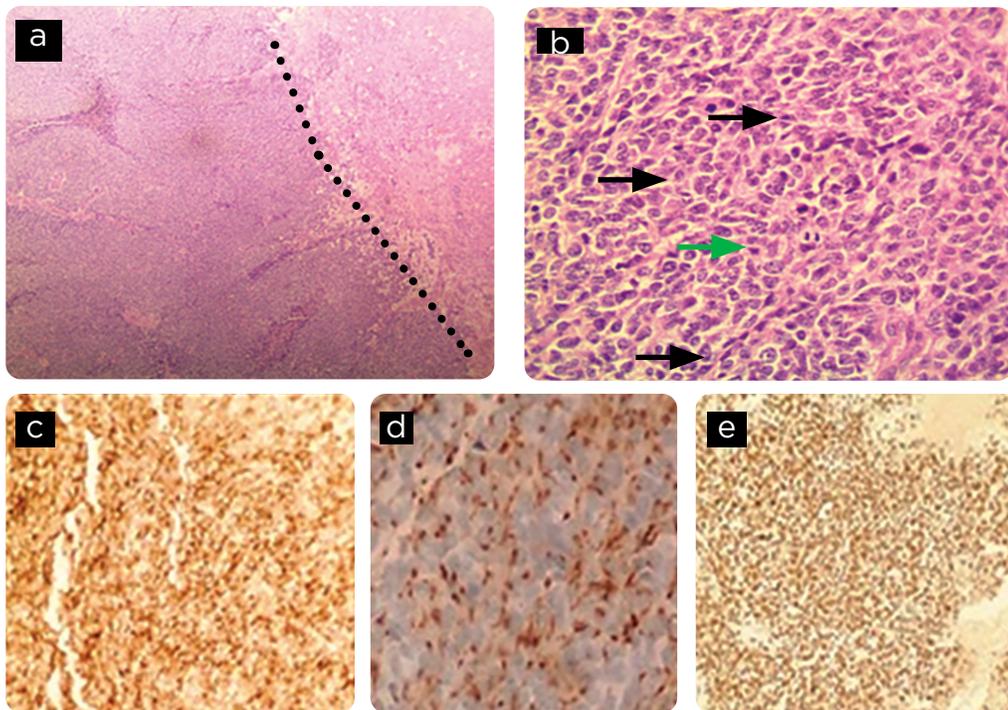


Figure 6. Histologic images. Hematoxylin and eosin (A and B). In A (microphotography at 5×) liver parenchyma with extensive infiltration by malignant tumor is observed on the left side of the photograph which is arranged in sheets (the limit of the tumor and the liver parenchyma is demarcated with a dotted line). B (microphotography at 40×), tumor cells show vesicular nuclei with clear chromatin (black arrows) and scant eosinophilic cytoplasm, with mitosis (green arrow). Immunoperoxidase studies were performed and confirmed the diagnosis: C (10× microphotography) membrane labeling for CD-56; D (10× microphotography) Golgi pattern labeling for desmin; E (10× microphotography) Ki-67 cell proliferation index of 80% in tumor cells.

## Conclusion

DSRCT is a rare sarcomatous neoplasm, most frequently affecting young white male patients. On imaging, the characteristic finding is multiple soft tissue masses primarily involving the pelvis and peritoneal cavity without a definite solid organ origin. The radiologist should be familiar with this pathology and consider it within the differential diagnosis of peritoneal masses in young patients, as well as know its behavior and potential complications in the different imaging modalities.

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