Case report

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The added value of CEUS and ultrasound-guided biopsy in diagnosing an aggressive desmoplastic small round cell tumour of peritoneum in a young male. A case report

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Abstract

Desmoplastic small round cell tumour (DSRCT) is a rare and highly aggressive mesenchymal neoplasm with poor prognosis that develops in male adolescents and young adults. We report the case of a 32-year-old male admitted with abdominal distension and ascites. An ultrasonography (US) scan showed multiple peritoneal masses with large ascites. The dominant mass had a hypervascular homogenous aspect at contrast-enhanced ultrasound with wash-out in the venous phase. Thoraco-abdominal CT, performed for staging the disease, confirmed the US aspect. The US-guided percutaneous biopsy revealed DSRCT of the peritoneum. Chemotherapy was then started with minimal clinical improvement, increase in tumoral burden and death after three months. US and US-guided biopsy played an essential role in diagnosing this case. The aggressive course of the disease and seeding at paracentesis sites are the particularities of the presented case.

Keywords: desmoplastic small round cell tumour; contrast-enhanced ultrasound; percutaneous biopsy

Introduction

Desmoplastic small round cell tumour (DSRCT) is a rare and highly aggressive mesenchymal neoplasm with poor prognosis, mainly seen in adolescents and young adults [1]. This neoplasm generally affects the peritoneum as multifocal/metastatic disease without an apparent organ of origin. The tumours spread along the peritoneum and mesothelial lined surfaces [1,2]. The diagnosis is suggested by imaging methods and is established by biopsy, either surgical or percutaneous image-guided. There is limited knowledge of this disease's pathologic and clinical nature with only case reports and a small series of patients, most of them radiological series [3,4].

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Case report

A 32-year-old Caucasian male presented to the Emergency Department with symptoms of abdominal distension, discomfort and constipation. Physical examination revealed tense ascites and mild abdominal tenderness.

On admission, slight elevation of erythrocytes sedimentation rate (ESR) (18-25 mm/hr) and leukocytosis (13,700/mm^3) was present. Tumour markers were normal. Grayscale ultrasonography (US) revealed an intraperitoneal mass in the upper left quadrant (72/44 mm in diameter), other multiple mesenteric and omental masses and large ascites. The masses were 1-3 mm large, hypoechogenic, some with a polylobulated appearance. The liver was normal with no signs of portal hypertension. A moderate bilateral pleural effusion with diaphragmatic pleural lesions (up to 2 cm) was present on thoracic US (fig 1).

Diagnostic and therapeutic paracentesis was performed. The macroscopic appearance of ascites was serocitrin; the biochemical analysis was normal, while

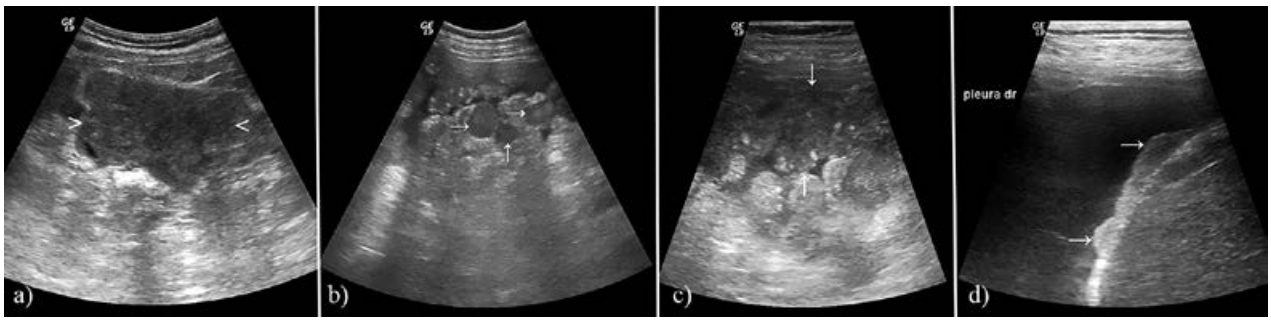


Fig 1. US image of a large intraabdominal DSRCT: a) an intraperitoneal mass in upper left quadrant; b) multiple intraperitoneal masses in the mesenteric structures (<) with ascites; c) thickened great omentum, with nodular structure; d) pleural metastasis (→) with effusion.

cytology described frequent atypical mesothelial cells. To rule out peritoneal carcinomatosis, upper and lower endoscopy was performed, both being negative.

The contrast-enhanced ultrasound (CEUS) examination (SonoVue, Bracco, Italy) revealed a homogenously enhancing of the left upper quadrant tumour with washout in the venous phase (fig 2).

CT scan showed multiple intraperitoneal masses (the biggest one in the upper left quadrant) with "omental cake" appearance, mesenteric and retroperitoneal lymphadenopathy, a large quantity of ascites, malignant infiltration of the diaphragmatic pleura (36 mm thickness in the right and 15 mm in the left), anterior and posterior mediastinal lymphadenopathy and moderate bilateral pleural effusions (fig 3).

For the final diagnosis, tissue sampling was performed using percutaneous ultrasound-guided biopsy (18G Bard needle coupled on a Biopsy Gun) from the left upper quadrant intraperitoneal mass.

Microscopically the specimens were composed of sharply demarcated nests of varying size with small round or oval cells embedded in a desmoplastic stroma. Some tumour cells had small hyperchromatic nuclei with inconspicuous nucleoli or formed cords surrounded by desmoplastic stroma. Immunohistochemistry was negative for CK5.6 and CD99 and positive for desmin, NSE and AE1/AE3. Both the morphological aspect and immunohistochemistry were compatible with DSRCT (fig 4).

The patient was referred to the Oncological Institute for chemotherapy, but the evolution was unfavourable and the patient died three months after initial presentation with multiorgan failure. During the first weeks of chemotherapy, the number of paracenteses gradually increased from 3 to 5-7 per week. Four weeks after beginning chemotherapy, US showed an increased number of peritoneal tumours and a seeding tumour at the paracentesis site (fig 5).

Discussion

DSRCT belongs to the category of "small round blue cell tumours", which also include neuroblastoma, Wilms' tumour and rhabdomyosarcoma. These malignancies have similar histological appearance in common, which consists of poorly/undifferentiated, small or medium-sized round or spindle-shaped cells, with round nuclei within a dense desmoplastic stroma [1-4]. Immunohistochemistry shows a characteristic polyphenotipic differ-

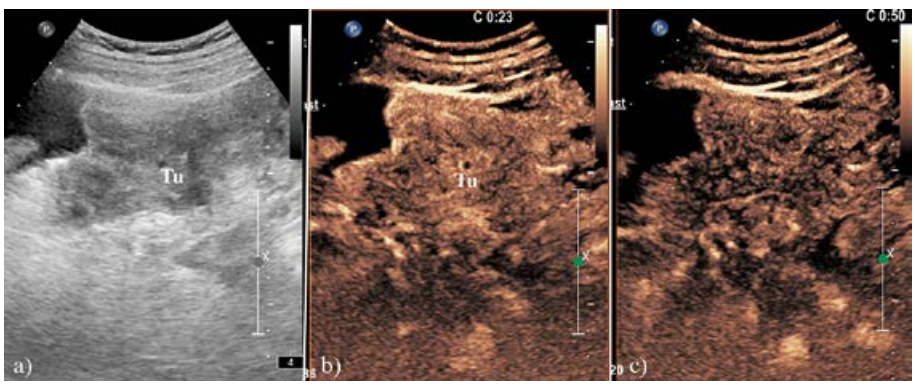


Fig 2. CEUS of the dominant tumour: a) conventional image; b) arterial phase - homogenous enhancement of the lesion without necrotic areas; c) venous phase with contrast agent wash-out.

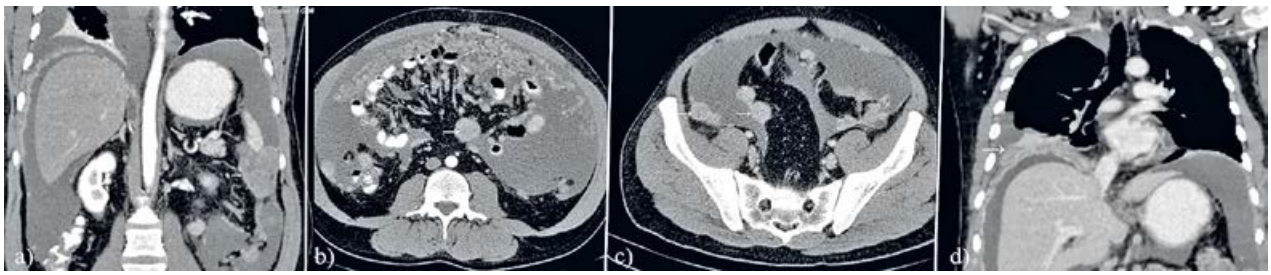


Fig 3. Abdominal and thoracic CT scans: a) the dominant intraperitoneal mass (→) in upper left quadrant, coronal view, arterial phase: homogenous enhancement; b) mesenteric nodules (>), thickened great omentum with nodules (↑↓), retroperitoneally lymphadenopathy (→); c) pelvic nodules; d) infiltration of both diaphragmatic pleurae with nodules.

entiation with co-expression of epithelial, mesenchymal and occasionally neuronal markers [3-5].

The most common signs and symptoms of intra-abdominal DSRCT include abdominal pain, distension, constipation, ascites and a palpable abdominal mass [1,2,4]. DSRCT should be considered a potential cause of ascites in adolescents and young adults [6].

The most common CT findings are multiple peritoneal, omental and serosal soft tissue masses ranging from 2 to 22 cm [4,6]. In patients with multiple peritoneal masses, a dominant mass is present, the most common sites being the retrovesical/rectouterine location (51% of cases) and peritoneal/omental area (in 48% of cases) [6]. Small peritoneal masses (<10 cm) show modest uniform enhancement on contrast-enhanced CT (CE-CT) while, larger masses demonstrate low central attenuation with modest heterogeneous enhancement on CE-CT suggestive of central haemorrhage or necrosis [3-6]. In our patient, the dominant mass, 7 cm in size located in the great omentum in the left upper quadrant, was heterogeneously enhanced without clearly necrotic areas and numerous tumoral implants were found in the mesentery, great omentum, peritoneal surface and retroperitoneal lymph nodes.

Pleural involvement has been detected as pleural masses, nodularity and effusion in 25-100% of cases [3]. There were several metastases to the diaphragmatic pleura in our patient on both sides, with important pleural effusion on the right side.

Few papers have described the US features of DSRCT [6,7]. The lesions may harbour different echogenicities depending on the amount of intratumoral necrosis and haemorrhage. Tiny calcifications have been also reported [7]. The vascularity assessed by the Doppler techniques is also heterogeneous [7]. Moreover, CEUS of the dominant peritoneal tumour allowed us to characterise the tumour. The enhancement pattern (homogeneous) was similar to that obtained with CE-CT for lesions below 10 cm [6]. To our knowledge, this is the first description of a CEUS examination of an abdominal DSRCT.

US-guided biopsy is a well-accepted method to diagnose peritoneal and omental lesions [8]. Large lesions with necrotic areas and the use of fine needles are factors associated with lower performances [8,9]. CEUS use in selecting viable sampling areas before/during percutaneous biopsy may avoid false-negative results.

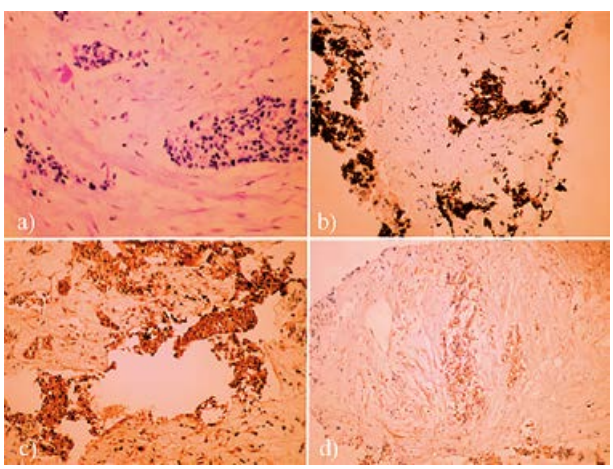


Fig 4. Microscopic appearance of the tumour: a) HE 400× desmoplastic stroma, medium-sized round cells, with indistinct borders, round/oval nucleoli increased in size, scant cytoplasm; b) Desmin 200× strong cytoplasmic staining, c) NSE 200× cytoplasmic staining; d) AE1/AE3200x positive.

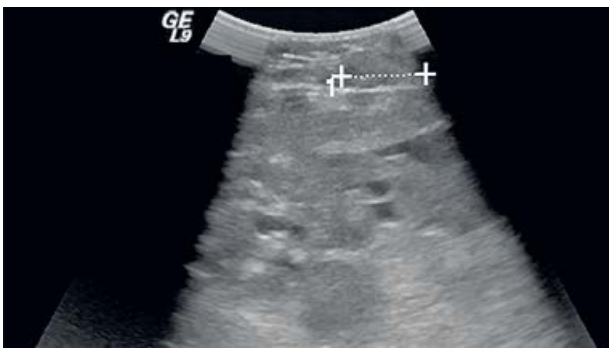


Fig 5. US image of a 2.5 cm seeding tumour in the abdominal wall at the site of numerous paracentesis P-peritoneum

As far as we know, no other case of paracentesis site seeding has been reported. In malignant ascites, paracentesis site seeding is an uncommon complication. Whether this affects the overall survival is not known.

In **conclusion,** although rare, the DSRCT should be considered a potential cause of ascites in adolescents and young adults. CT has a central role in diagnosing and staging the disease, but US (conventional, contrast-enhanced) and US-guided biopsy may play an important role in the diagnosis. Seeding at the paracentesis site and very short survival are the particularities of the presented case.

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