The radiology of desmoplastic small round cell tumour

Poster No.: C-1648
Congress: ECR 2010
Type: Educational Exhibit
Topic: GI Tract
Authors: G. Rajeswaran, S. Ganeshalingam, R. L. Jones, K. Thway, E. Moskovic; London/UK
Keywords: desmoplastic small round cell tumour, DSRCT, desmoplastic
DOI: 10.1594/ecr2010/C-1648

Any information contained in this pdf file is automatically generated from digital material submitted to EPOS by third parties in the form of scientific presentations. References to any names, marks, products, or services of third parties or hypertext links to third-party sites or information are provided solely as a convenience to you and do not in any way constitute or imply ECR's endorsement, sponsorship or recommendation of the third party, information, product or service. ECR is not responsible for the content of these pages and does not make any representations regarding the content or accuracy of material in this file.

As per copyright regulations, any unauthorised use of the material or parts thereof as well as commercial reproduction or multiple distribution by any traditional or electronically based reproduction/publication method is strictly prohibited.

You agree to defend, indemnify, and hold ECR harmless from and against any and all claims, damages, costs, and expenses, including attorneys' fees, arising from or related to your use of these pages.

Please note: Links to movies, ppt slideshows and any other multimedia files are not available in the pdf version of presentations.

www.myESR.org
Learning objectives

The purpose of this poster is to describe the imaging features of desmoplastic small round cell tumour (DSRCT) in the context of the clinical and demographic data.

Background

Desmoplastic small round cell tumour (DSRCT) is a rare but highly aggressive soft tissue sarcoma that arises most commonly in the abdomino-pelvic cavity of males in adolescence or young adulthood. [1] First reported in 1989 by Gerald et al and Ordonez et al, this tumours is so called due to its typical histological finding of nests of small blue round cells within a dense desmoplastic stroma. [1, 2]

There are only a few hundred reported cases in the literature, of which the overwhelming majority focus on the clinico-pathological features or treatment strategies, with limited radiological information.

Differentiation of DSRCT from other soft tissue sarcomas is important as it is a high grade neoplasm often presenting with advanced disease and with a mean survival time of less than 3 years. [3, 4]

Imaging plays an important role in both the diagnosis and management of DSRCT. Given the paucity of available imaging features of this condition, we present the largest radiological review of DSRCT to date (in 16 patients), in the context of the demographic and clinical data.

Imaging findings OR Procedure details

DSRCT is an extremely rare and aggressive soft tissue sarcoma, with only a few hundred cases reported in the literature generally presenting with multifocal abdomino-pelvic masses. Our practice based in a large tertiary referral centre for soft tissue sarcoma reflects this, with only 23 documented cases in an 18 year period.

Although this tumour is reported to occur in adolescence and early childhood, in our series of patients, the mean age at presentation was 28.3 years with the oldest patient being 44 years of age. This is slightly higher than the usual age range of 18-25 years,
although DSRCT has been reported in patients older than this. [5] The male to female ratio of DSRCT is reported to be 4:1. [6, 7] In our patient cohort, the male to female ratio was 3:1, confirming a male preponderance in this histological subtype.

The aetiology of DSRCT is unknown but it is thought to be a malignancy of the mesothelium and as such, occurs most commonly in the peritoneum and omentum. [8] However, it has been noted to occur in other sites of mesothelial origin such as the pleura and tunica vaginalis of the testis as well as solid organs such as the pancreas, liver and ovaries. [5, 9-12] In our series, 15 cases (94%) involved the peritoneum or omentum with the remaining case occurring in a right paravertebral location from the right crus of the diaphragm (Figure 1).

**Figure 1: Sagittal T1 weighted MRI of the lower thoracic spine in a 20 year old male with DSRCT.** There is a heterogeneous paravertebral mass lying anterior to the T11 and T12 vertebral bodies (white arrowheads). Whilst there is no direct invasion of the vertebral bodies, there is low signal intensity within the T11 and T12 vertebral bodies in keeping with bony metastatic disease (white arrows).
Fig.: Figure 1: Sagittal T1 weighted MRI of the lower thoracic spine in a 20 year old male with DSRCT. There is a heterogeneous paravertebral mass lying anterior to the T11 and T12 vertebral bodies (white arrowheads). Whilst there is no direct invasion of the vertebral bodies, there is low signal intensity within the T11 and T12 vertebral bodies in keeping with bony metastatic disease (white arrows).

References: G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

DSRCT is one of a group of soft tissue tumours that share similar histological characteristics of nests or strands of small round cells, and which includes Ewing's sarcoma/primitive neuroectodermal tumour (PNET) and Wilm's tumour. Histologically, it can be differentiated from other tumours in this group, as the small round cells are
seen embedded in a prominent desmoplastic stroma (Figure 2a). [13] Mitotic figures are frequent (Figure 2b) and central necrosis is common.

**Figure 2a: Desmoplastic small round cell tumour.** Histology shows sheets of tumour composed of small round cells (white arrow) which are surrounded by prominent sclerotic fibrous stroma (black arrow). (Haematoxylin and eosin, x100)

Fig.: Figure 2a: Desmoplastic small round cell tumour. Histology shows sheets of tumour composed of small round cells (white arrow) which are surrounded by prominent sclerotic fibrous stroma (black arrow). (Haematoxylin and eosin, x100)

**References:** G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

**Figure 2b: Desmoplastic round cell tumour.** Higher magnification shows markedly cellular tumour composed of monotonous cells with round or ovoid vesicular nuclei and scanty cytoplasm. Note the prominent mitotic figures (thin arrows) and apoptotic body (thick arrow). (Haematoxylin and eosin, x400)
**Fig.:** Figure 6b: Desmoplastic round cell tumour. Higher magnification shows markedly cellular tumour composed of monotonous cells with round or ovoid vesicular nuclei and scanty cytoplasm. Note the prominent mitotic figures (thin arrows) and apoptotic body (thick arrow). (Haematoxylin and eosin, x400)

**References:** G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

The tumour has a polyphenotypic antigen expression profile, expressing proteins associated with neural, epithelial and muscular differentiation, and as such can also be diagnosed using immunohistochemistry with a broad panel of antibodies. [14] However, definitive diagnosis requires molecular cytogenetic or molecular genetic analysis using fluorescence in situ hybridisation (FISH) and reverse transcriptase polymerase chain reaction (RT-PCR) methods to detect the presence of the characteristic t(11;22) (p13;q12) translocation. These methods demonstrate the presence of an EWS (Ewing's sarcoma)-WT1 (Wilm's tumour) gene fusion, or its RNA transcript, both of which are specific to DSRCT. [15, 16]

Presentation with DSRCT is usually late. Given that most commonly there is no organ of origin, presenting clinical signs and symptoms are often vague. In our study, the most common presenting features were of an abdominal mass (75%) and abdominal
pain (50%). This is likely to be due to the large tumour burden at presentation and in our cohort, of the 14 patients with a dominant soft tissue mass on imaging, all were \(\geq 5\) cm in size and 71% were \(\geq 10\) cm. In one case, the tumour bulk resulted in biliary obstruction and there are reports of obstruction due to mass effect on other adjacent structures such as the bowel and renal tract. [17] Other reported presenting clinical features include constipation, diarrhoea, haematuria, dysphagia and haematemesis. [4] In the absence of clinical symptoms, DSRCT is usually found incidentally on imaging studies or at laparotomy performed for other reasons. In our cohort, DSRCT was found incidentally at caesarean section and during laparotomy for appendicitis.

Imaging findings in our series included diffuse peritoneal/omental involvement (88%) with multiple soft tissue masses seen in 80% of these cases (Figure 3a).

**Figure 3a: Axial CECT of the pelvis in a 22 year old female with DSRCT.** There are multiple peritoneal soft tissue nodules in the pelvis (white arrows).
Fig.: Figure 3a: Axial CECT of the pelvis in a 22 year old female with DSRCT. There are multiple peritoneal soft tissue nodules in the pelvis (white arrows).

References: G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

A dominant soft tissue mass, either solitary (and not involving a primary organ) or at least twice the size of other peritoneal/omental lesions was seen in 88% of patients (Figure 4 & 5).

**Figure 4: Axial CECT of the abdomen in a 24 year old male with DSRCT.** There is a large, heterogeneous peritoneal mass in the abdominal cavity (white arrowheads). It is predominantly cystic but has solid enhancing tissue within it (black arrows). Peritoneal thickening and a peritoneal soft tissue nodule is seen separate to the mass (white arrow).
**Fig.** Figure 4: Axial CECT of the abdomen in a 24 year old male with DSRCT. There is a large, heterogeneous peritoneal mass in the abdominal cavity (white arrowheads). It is predominantly cystic but has solid enhancing tissue within it (black arrows). Peritoneal thickening and a peritoneal soft tissue nodule is seen separate to the mass (white arrow).

**References:** G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

**Figure 5:** Axial CECT of the abdomen in a 25 year old male with DSRCT. There is a large heterogeneous, mixed solid and cystic mass in the left upper quadrant (white arrowheads). The soft tissue component enhances. There is calcification within it (black arrow).
**Fig.:** Figure 5: Axial CECT of the abdomen in a 25 year old male with DSRCT. There is a large heterogeneous, mixed solid and cystic mass in the left upper quadrant (white arrowheads). The soft tissue component enhances. There is calcification within it (black arrow).

**References:** G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

This has been noted in previous smaller studies. [18-20] In a review of the CT findings of 11 patients with DSRCT by Bellah et al, in their cohort, 82% of the dominant lesions were in the rectovesical or rectouterine space and they postulated that this might be due to the dynamic flow of peritoneal fluid and the dependency of these locations. [18] However, our findings do not support this theory, with only 36% (5/14 patients) of the dominant lesions occurring in the pelvis, the remainder occurring in the abdomen (57%) and the right paravertebral region arising from the diaphragmatic crus (7%).
Regarding the imaging characteristics of the soft tissue lesions, enhancement following intravenous contrast was seen in all cases on both CT and MRI. This was heterogeneous in 69% of cases on CT and in 100% of the 3 cases in which lesions were characterised further using MRI. Central low attenuation was seen within lesions on CT in 50% of cases and in 100% of the 3 cases evaluated on MRI, most likely to be due to necrosis, haemorrhage or a fibromyxoid component to the tumour (Figures 4 and 5). [18, 20] Lesional calcification was noted in 25% of the cases on CT (Figure 5). These findings are consistent with previous smaller studies. [18-22]

Lymph node enlargement was a common finding, demonstrated in 50% of patients at presentation. Of those patients with lymph node involvement, 88% had retroperitoneal lymph node enlargement with the next most common sites being in the mediastinal (25%) and mesenteric (14%) regions. This is consistent with prior smaller studies noting lymphatic spread at presentation. [18-22] Ascites (31%) and pleural effusions (25%) at presentation were common findings in our study and this is again consistent with smaller studies.

Quaglia et al demonstrated distant metastases in 50% of their cases and whilst the frequency in our patient cohort was lower (25%), our findings still suggest that metastases are common at presentation. As such, imaging studies should be evaluated thoroughly for metastases, particularly in the liver and bones (Figure 3b). [4]

**Figure 3b: Axial CECT of the abdomen in a 22 year old female patient with DSRCT.** There are multiple hypoattenuating, heterogeneous liver metastases (black arrows). There is diffuse peritoneal thickening scalloping the liver edges (white arrowheads). There is small volume ascites (white arrow).
Fig.: Figure 3b: Axial CECT of the abdomen in a 22 year old female patient with DSRCT. There are multiple hypoattenuating, heterogeneous liver metastases (black arrows). There is diffuse peritoneal thickening scalloping the liver edges (white arrowheads). There is small volume ascites (white arrow).

References: G. Rajeswaran; Radiology, Chelsea & Westminster Hospital, London, UNITED KINGDOM

The differential diagnosis for DSRCT includes tumours of peritoneal/coelomic origin such as peritoneal carcinomatosis, leiomyomatosis peritonealis disseminata, pseudomyxoma peritonei, peritoneal mesothelioma, metastatic pleural disease and primary peritoneal carcinoma. Other conditions not of peritoneal origin that can be mistaken for DSRCT include rhabdomyosarcoma, metastatic ovarian carcinoma and mesenteric carcinoid. [27, 28]

Treatment strategies for DSRCT include debulking surgery, radiotherapy and chemotherapy. These approaches are based on small case series and single patient reports, and thus it is difficult to define the optimal therapy for this condition. Because
of the diffuse nature of this disease at presentation total resection is often not possible, and there are numerous studies reporting the use of various chemotherapy regimens, including alkylating agents, anthracyclines, platinum compounds, vincristine and etoposide. [3, 23, 24] Other studies have suggested a role for whole abdomino-pelvic radiotherapy as a component of multi modality management. [25] Despite these efforts the prognosis remains poor with a mean survival time of less than 3 years. [3, 4]. The findings of our study confirm this poor prognosis despite treatment, with a median overall survival from diagnosis of 22.8 months.

An aggressive multi modality approach to treatment is generally recommended involving combination chemotherapy followed if possible by surgery and/ or radiotherapy. [26] Lal and colleagues, in a series of 66 patients, demonstrated 3-year overall survival to be significantly better in those treated with chemotherapy followed by surgery and radiotherapy (55%) compared to when all 3 modalities were not used (27%). [26] All of the 3 patients treated with follow up who are still alive received multi modality therapy and our results support the use, if possible, of multi modality therapy to obtain long-term survival in the treatment of this particular soft tissue sarcoma histological subtype. However, further work is required to identify novel therapies to treat this challenging condition and ideally, patients with DSRCT should be referred to specialist soft tissue tumour centres to be included in clinical trials for this purpose.

Conclusion

The radiologist has an important role in the management of patients with DSRCT, providing diagnostic imaging, percutaneous image guided biopsy, follow up imaging after treatment and assessing response to therapy.

It is important for the radiologist to consider the diagnosis of DSRCT and differentiate it from other similar malignancies with different management options. This tumour should be suspected in adolescent/young males in whom imaging studies demonstrate: diffuse peritoneal involvement with multiple soft tissue lesions; a dominant soft tissue lesion without an organ of origin; heterogeneous tumoural enhancement, cystic change and calcification.

Personal Information

References


