

Contents lists available at [ScienceDirect](https://www.sciencedirect.com)

International Journal of Surgery Case Reports

journal homepage: www.casereports.com

Intra-abdominal desmoplastic small round cell tumour in a 56-year-old female: Case report of a very rare presentation of an unusual tumour

Jason Russell Laurens^{a,*}, Adam John Frankel^a, Bernard Mark Smithers^b

^a Princess Alexandra Hospital, Woolloongabba, QLD, 4102, Australia

^b The University of Queensland, Upper Gastro-intestinal and Soft Tissue Unit, Princess Alexandra Hospital, Woolloongabba, QLD, 4102, Australia

ARTICLE INFO

Article history:

Received 3 November 2020

Received in revised form 13 January 2021

Accepted 14 January 2021

Available online 19 January 2021

Keywords:

Desmoplastic small round cell tumour

Intra-abdominal tumour

Surgical oncology

Case report

ABSTRACT

INTRODUCTION: This case report discusses the rare diagnosis of intra-abdominal desmoplastic small round cell tumour (DSRCT) in a 56-year-old female.

PRESENTATION OF CASE: An incidental intra-abdominal lesion was found during investigation of joint pain. Ultrasound-guided biopsy suggested desmoid tumour, after undergoing laparotomy and en-bloc excision of the tumour due to concerning radiological progression, the final histology was desmoplastic small round cell tumour. At six-week follow-up imaging, no recurrence or metastatic disease was noted. She declined chemotherapy and specialist follow-up, electing to have routine follow up with her General Practitioner only.

DISCUSSION: Intra-abdominal DSRCT is rare and mainly seen in young males. To our knowledge, this is the only reported case of DSRCT in a female over the age of 50.

CONCLUSION: There should be timely discussion between different surgical units to provide efficient care. Any disparity between radiological and histological appearance should prompt further review and investigation in order to ensure misdiagnosis is avoided and appropriate treatment is provided. Despite cytoreductive surgery, survival is dismal due to the aggressive nature of the tumour, and its low numbers limiting adequate study into post diagnosis care

© 2021 The Authors. Published by Elsevier Ltd on behalf of IJS Publishing Group Ltd. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

1. Presentation of case

A 56-year-old female had an incidental large intra-abdominal mass found on single-photon emission computed tomography (SPECT) performed for investigation of joint pain. Her medical history was osteoarthritis with chronic pain, asthma, obstructive sleep apnoea, gastro-oesophageal reflux disease, bipolar disorder and morbid obesity with a BMI of 53 kg/m². Her chronic pain management in the community included paracetamol 1 g QID, multiple opiates (250 mg daily oral morphine equivalent), gabapentin 300 mg BD, diazepam 5 mg TDS and temazepam 5 mg nocte. Her surgical history included a total abdominal hysterectomy, laparoscopic cholecystectomy and umbilical hernia repair with preperitoneal mesh. There was no relevant family history. She was referred to the local regional general surgery department who arranged an abdominal computed tomography (CT) scan to define the mass. Six weeks after the SPECT, she underwent ultrasound-guided core biopsy. Histology reported desmoid fibromatosis.

She was referred to our tertiary hospital-based sarcoma service for an opinion and management. Our multi-disciplinary team (MDT) felt that the radiological appearances were unusual for desmoid fibromatosis and a repeat CT at one month was recommended to assess progress [Fig. 1](#). In parallel and unknown to us, the local team had requested a CT/PET which was performed 10 days after the MDT meeting (3 months post initial CT). This demonstrated FDG avidity of the tumour with some free peritoneal fluid. The repeat CT we arranged occurred three weeks later and suggested tumour necrosis with possible rupture, associated with large volume ascites [Fig. 2](#). Two days after this, she attended our clinic as scheduled. Given the progressive change on imaging did not support a diagnosis of desmoid tumour, and she lived 150 km away, she was admitted. At this time, she had nausea, without vomiting, normal observations and a distended and centrally tender abdomen. A full blood count, renal and hepatic function tests and C-reactive protein were all unremarkable. She was counselled to undergo an exploratory laparotomy and likely resection based on intra-operative findings.

Under general anaesthetic, Professor Smithers performed a mid-line laparotomy, which revealed 4.5 L of straw-coloured ascites and a grapefruit-sized irregular pale mass in the mid-small bowel

* Corresponding author.

E-mail address: jasonlaurens78@gmail.com (J.R. Laurens).



Fig. 1. CT abdomen, showing intact intra-abdominal tumour.

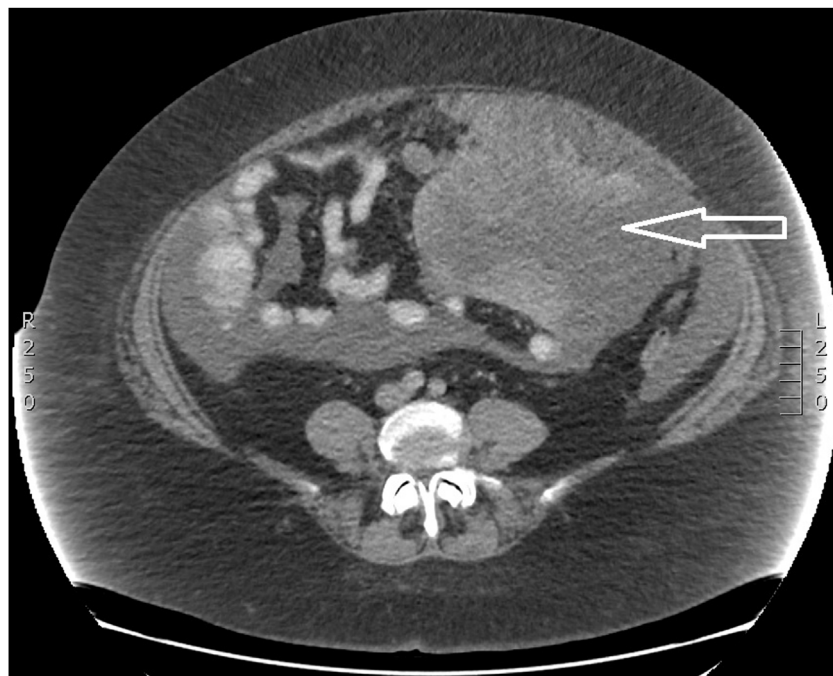


Fig. 2. CT abdomen, showing suggested tumour rupture and free fluid.

mesentery. A loop of small bowel, a short segment of transverse colon and three sections of omentum were densely adherent to the mass. The adjacent omentum contained multiple large veins approaching 1 cm in diameter but there was no evidence of varices elsewhere. The mass was excised en-bloc with bowel anastomoses Fig. 3. A nearby portion of omentum containing an enlarged firm vein was resected separately. She was admitted to the high dependency unit, to monitor her respiratory function with concerns regarding her pre-operative opiate and benzodiazepine use. She recovered quickly and was discharged home on post-operative day six with normal gut function and near-baseline mobility.

The final histology was reported as a 150 mm desmoplastic small round cell tumour (DSRCT) with adjacent small tumour deposits and foci of vascular invasion. The separate specimen of omentum showed a large vein occluded by tumour thrombus. Immunohistochemistry demonstrated positive nuclear staining for *WT1* and *SATB2*. Fluorescence In-Situ Hybridisation (FISH) demonstrated rearrangement of the *EWS-R1* gene region. Cytokeratin staining was negative, with a final diagnosis of DSRCT (Figs. 4 and 5).

The operative findings and pathology were discussed at our MDT and it was recommended to repeat the CT/PET at 6 weeks. This showed no evidence of recurrence or metastatic disease. Following consultations with a medical oncologist, she chose not

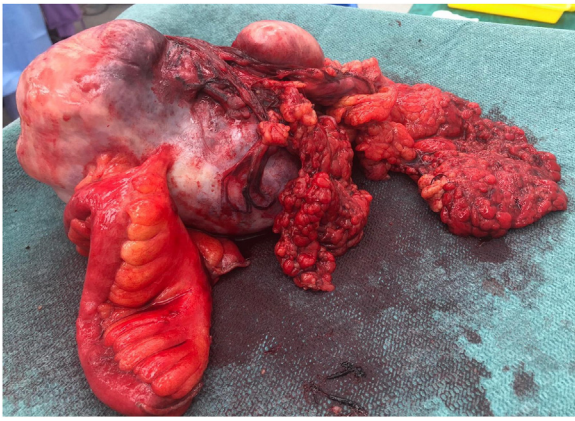


Fig. 3. Photo of en-block excision of tumour.

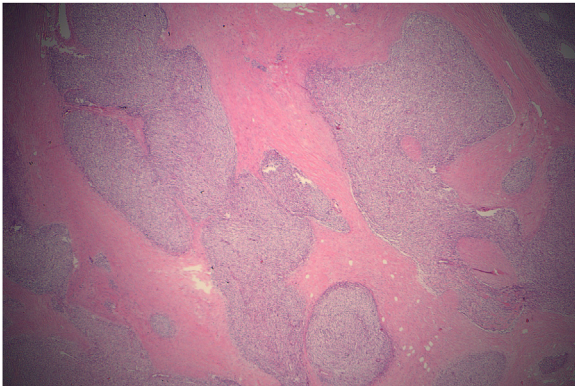


Fig. 4. H&E histology slide.

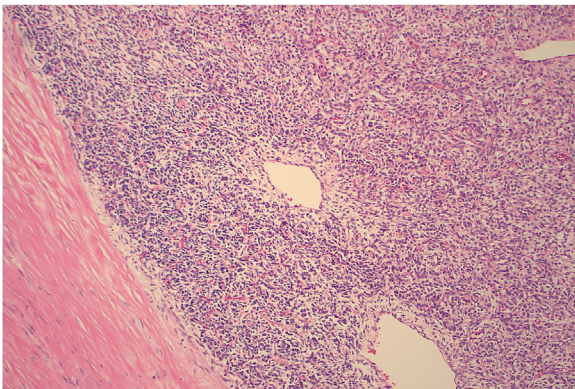


Fig. 5. H&E histology slide.

to undergo systemic therapy and declined clinical follow-up and repeat imaging. She did accept ongoing follow-up only with her general practitioner.

2. Discussion

Intra-abdominal desmoplastic small round cell tumour (IAD-SRCT) is a rare and often aggressive tumour with generally poor prognosis [1,11]. First described by Gerald and Rosai in 1989 [2], it has slowly built a presence in the literature although there are still few recorded cases. The usual presentation of IADSRCT is with abdominal pain [3–5]. It is rarely seen in females (4:1 M:F) and is usually diagnosed before the age of 30 [5]. To our knowledge, this is the only report of a case in a female over the age of 50.

Radiological studies are not specific, and diagnosis rests on histology with cytogenetic studies confirming the presence of the unique chromosomal translocation $t(11:22)(p13;q12)$. This genomic change results in the functional fusion of the *EWS* and *WT1* genes [6,7], similar to Ewing sarcoma. The rarity of the pathology means there are currently no evidence-based therapeutic guidelines on post-resection management, although several small series have shown partial response to adjuvant chemotherapy (P6 Protocol) [4,5,8]. In select cases, cytoreductive surgery (CRS) and intraperitoneal hyperthermic chemoperfusion (HIPEC) has been provided; one case had recurrence at 8.85 months [11] while a small phase 2 trial showed overall survival of 44.3 months [10]. It should be noted the patients who underwent CRS + HIPEC were young, otherwise healthy males, while our patient is a late middle-age female and morbidly obese with numerous comorbidities. It seems that regardless of treatment strategy, the long-term prognosis is very poor.

3. Conclusion

While this case report describes an unusual presentation of a rare tumour, the broader lessons are that there should be clear and timely communication between different surgical units to provide efficient care, and that disparity between radiological and histological appearance should prompt further review and investigation. This will help to ensure that misdiagnosis is avoided, appropriate treatment is provided in a timely fashion, aiming to minimise adverse outcomes. This case has been reported in line with the SCARE 2018 criteria [9].

Declaration of Competing Interest

The authors report no declarations of interest.

Funding

No sources of funding required for this case report.

Ethical approval

No ethics approval was sought as this is a retrospective de-identified case report.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Registration of research studies

Not applicable.

Guarantor

Jason Laurens.

Provenance and peer review

Not commissioned, externally peer-reviewed.

CRedit authorship contribution statement

Jason Russell Laurens: Conceptualization, Investigation, Writing - original draft, Writing - review & editing. **Adam John Frankel:** Conceptualization, Supervision, Writing - review & editing. **Bernard Mark Smithers:** Supervision, Writing - review & editing.

References

- [1] C.E. Stuart-Buttle, C.J. Smart, S. Pritchard, D. Martin, I.M. Welch, Desmoplastic small round cell tumor: a review of literature and treatment options, *Surg. Oncol.* 17 (2008) 107–112.
- [2] W.L. Gerald, J. Rosai, Desmoplastic small cell tumor with divergent differentiation, *Pediatr. Pathol.* 9 (1989) 177–183.
- [3] A. Gil, A. Gozem Portilla, E.A. Brun, P.H. Sugarbaker, Clinical perspective on desmoplastic small round-cell tumor, *Oncology* 67 (2004) 231–242.
- [4] P. Lippe, R. Berardi, C. Cappelletti, C. Massacesi, R. Mattioli, L. Latini, R. Cellerino, Desmoplastic small round cell tumour: a description of two cases and review of the literature, *Oncology* 64 (2003) 14–17.
- [5] A. Dufresne, P. Cassier, L. Couraud, P. Marec-Berard, P. Mesu, L. Alberti, J. Blay, Desmoplastic small round cell tumour: current management and recent findings, *Sarcoma* (2012), 714986.
- [6] W.L. Gerald, M. Ladanyi, E. de Alava, M. Cuatrecasas, B.H. Kushner, M.P. LaQuaglia, J. Rosai, Clinical, pathologic, and molecular spectrum of tumors associated with t(11;22)(p13;q12): desmoplastic small round-cell tumor and its variants, *J. Clin. Oncol.* 16 (September (9)) (1998) 3028–3036.
- [7] W.L. Gerald, J. Rosai, M. Ladanyi, Characterization of genomic breakpoint and chimeric transcripts in the EWS-WT1 gene fusion of desmoplastic small round cell tumor, *Proc. Natl. Acad. Sci. U. S. A.* 92 (1995) 1028–1032.
- [8] H. Fagouri, M. Ziyadi, A. Ibrahim, A. Osman, A. Babahabib, M. Oukablu, J. Kouach, D.R. Moussaoui, M. Dehayni, A Case of intra-abdominal desmoplastic small round cell tumor in a young girl patient, *Obstet. Gynaecol. Cases Rev.* 5 (4) (2018).
- [9] R.A. Agha, M.R. Borrelli, R. Farwana, K. Koshy, A. Fowler, D. Orgill, For the SCARE Group, The SCARE 2018 statement: updating consensus surgical Case Report (SCARE) guidelines, *Int. J. Surg.* (60) (2018) 132–136.
- [10] H.S. Fan, B. l'Ons, R. McConnell, V. Kumar, S. Alzahrani, D.L. Morris, Peritonectomy and hyperthermic intraperitoneal chemotherapy as treatment for desmoplastic small round cell tumour, *Int. J. Surg. Case Rep.* 7 (2015) 85–88.
- [11] A.A. Hayes-Jordan, B.A. Coakley, H.L. Green, L.C. Xiao, K.F. Fournier, C.E. Herzog, J.A. Ludwig, M.F. McAleer, P.M. Anderson, W.W. Huh, Desmoplastic small round cell tumor treated with cytoreductive surgery and hyperthermic intraperitoneal chemotherapy: Results of a Phase 2 trial, *Ann. Surg. Oncol.* 25 (2018) 872–877.

Open Access

This article is published Open Access at [sciencedirect.com](https://www.sciencedirect.com). It is distributed under the [IJSCR Supplemental terms and conditions](#), which permits unrestricted non commercial use, distribution, and reproduction in any medium, provided the original authors and source are credited.