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# Pathogenesis

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## Low-grade fibromyxoid sarcoma of the transverse colon: A case report

I.S. Farrell <sup>a,\*</sup>, K. Brelsford <sup>b</sup>, W. Salman <sup>b</sup>, C.J. Smart <sup>a</sup><sup>a</sup> Department of Colorectal Surgery, Royal Blackburn Hospital, East Lancashire NHS Trust, United Kingdom<sup>b</sup> Department of Histopathology, Royal Blackburn Hospital, East Lancashire NHS Trust, United Kingdom

### ARTICLE INFO

#### Article history:

Received 20 May 2014

Accepted 10 July 2014

Available online 21 July 2014

#### Keywords:

Low-grade fibromyxoid sarcoma

Colon

FISH

### ABSTRACT

A 52 year old lady presented with non-specific abdominal symptoms and menorrhagia. Ultrasound scan revealed an incidental finding of a large soft tissue mass. This was further characterised with a CT scan which revealed a 15 cm × 10 cm × 7 cm multilobulated mass. This was removed via a subtotal colectomy. Initial histology was reported as a most likely diagnosis of gastro-intestinal stromal tumour (GIST) but further characterisation revealed this to be a low-grade fibromyxoid sarcoma.

This is only the second reported case of low-grade fibromyxoid sarcoma of the colon and would potentially have been missed had not the sample been sent for second opinion at a regional specialist centre.

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### 1. Case history

A 52 year old lady was referred to gynaecology outpatients with a 2 year history of menorrhagia and intermenstrual bleeding. She also described occasional mild abdominal distension, a sensation of bloating and feeling nauseated. There was no recent change in her weight, appetite or bowel habit. She reported occasional 'hot flushes' which she had put down to the beginnings of menopause.

She had a past history of hypertension and anxiety and a family history of her father suffering from gastric carcinoma. In view of her menorrhagia, the gynaecologists arranged an ultrasound scan which revealed 4 soft tissue masses in the right adnexa extending above the umbilicus measuring 10 cm, 6 cm, 3.7 cm and 3.5 cm. They were separate to the ovaries and the uterus appeared normal. The tissue masses were reported to contain small blood vessels and a large vessel connecting them. A CT scan, with contrast, revealed a 15 cm × 10 cm × 7.5 cm multilobulated soft tissue mass in the central lower abdomen and pelvis, abutting and displacing the transverse colon and stomach, which was felt to be separate to both the ovaries and uterus (Fig. 1).

The colorectal MDT decision was for a laparotomy and resection, with a likely diagnosis of gastrointestinal stromal tumour (GIST) of small bowel origin. Laparotomy revealed a large cystic tumour in the mid transverse colon emanating from the bowel wall and mesentery. An additional small deposit was found in the distal sigmoid colon and posterior uterine wall.

A total colectomy was performed to incorporate the transverse and sigmoid lesion with side to side stapled anastomosis of ileum to rectum, the posterior uterine deposit was also resected. She made a routine recovery but this was complicated by ileus which required placement of an NG tube for 5 days and a chest infection which required IV antibiotics. She was discharged 17 days after her surgery.

Macroscopically the specimen showed a 15 cm × 9 cm × 6 cm tumour which was adjacent to the serosal surface. The mucosa showed no inflammation or ulceration (Fig. 2).

Microscopy showed a tumour with spindle shaped cells with eosinophilic, pigment free cytoplasm and elongated nuclei showing a mild degree of pleomorphism. The initial impression was of a CD117 negative GIST. An expert external opinion to include DOG1 staining was requested from the regional histopathology centre. The referral centre reported a well circumscribed tumour in the mesenteric/pericolic fatty tissue with invasion into the muscularis propria. The tumour consisted of spindled to stellate cells in an alternating fibrous and myxoid matrix. They were negative for DOG1 antibodies, but were strongly positive for MUC4 and EMA. Fluorescence in-situ hybridisation (FISH)

\* Corresponding author.

E-mail address: [ian.farrell@doctors.org.uk](mailto:ian.farrell@doctors.org.uk) (I.S. Farrell).

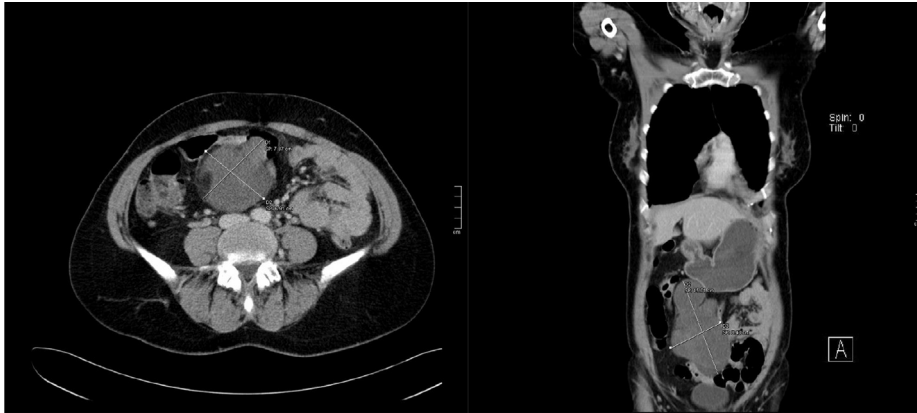


Fig. 1. Transverse and coronal CT images showing the soft tissue mass.

studies showed a pattern of atypical FUS gene rearrangement. The microscopy and immunohistochemistry were in keeping with a low grade fibromyxoid sarcoma (LGFS) rather than GIST as originally postulated.

The lesion in the uterus was a small benign leiomyoma and the sigmoid lesion was benign tissue and not microscopically significant (Figs. 3 and 4).

Supra-regional sarcoma MDT discussion did recommend adjuvant therapy and standard radiological/surgical follow up.

## 2. Discussion

The immunohistochemistry stains performed on this sample were the key to the correct diagnosis. CD117 and DOG1 have been shown to be the most sensitive stains for the diagnosis of gastrointestinal stromal tumours [1] both of which were negative in this case. MUC4 is positive in several different tumours including pancreatic and oesophageal adenocarcinomas but is extremely sensitive and specific for fibromyxoid sarcoma where it is suspected [2]. FISH studies are used to detect the presence or absence of specific DNA sequences on chromosomes. FUS (fused in sarcoma) gene rearrangement is highly specific for fibromyxoid sarcoma [3].

Low grade fibromyxoid sarcoma is in itself a rare tumour which most frequently presents in the trunk and deep tissues of the lower extremities. It is a slow growing tumour, growing between 1 and 18 cm per year [4]. There is a high rate of recurrence (64%) and a high rate of metastasis (45%), often very late (median 5 years) [5]. Death from the disease is common, occurring in 42% of cases [5]. LGFS appears to be extremely uncommon in the GI tract. There have been 6 cases reported in the small bowel mesentery in the past but there appears to be only one previous case of colonic fibromyxoid sarcoma reported [6]. Had the DOG1 stain been available at this hospital, it is possible that this

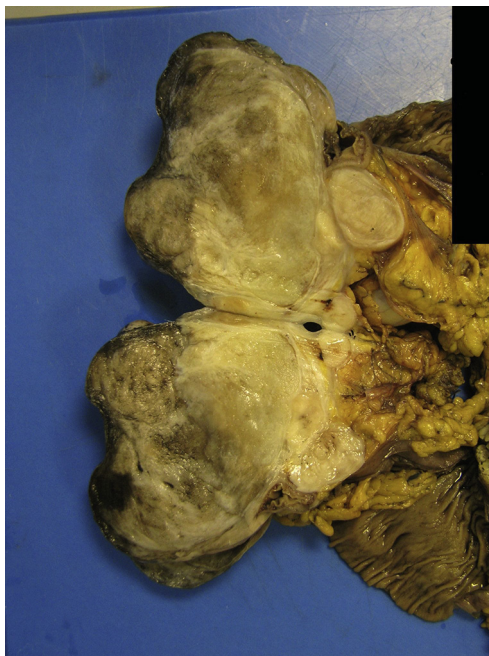
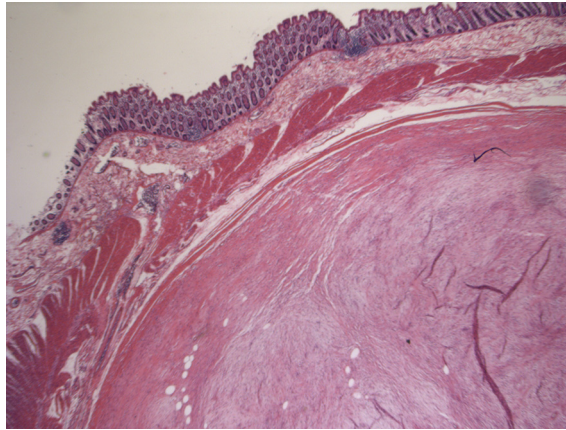
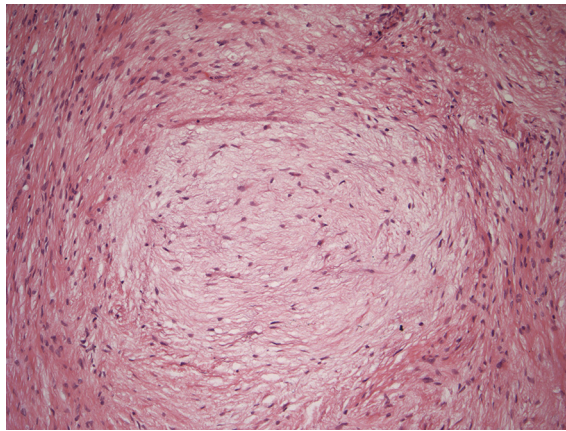


Fig. 2. Macroscopic image of the mass within the mesentery with adjacent colon.



**Fig. 3.** 16× power image showing spindled nature of the tumour cells.



**Fig. 4.** 100× power image showing surrounding myxoid change.

tumour may have been misdiagnosed as a GIST because it would not have gone on to have the MUC4 and FISH studies which were strongly positive for a diagnosis of LGFS.

### Acknowledgements

Dr P Shenjere – Department of Histopathology, The Christie NHS Trust, Manchester.

### References

- [1] Novelli, et al. DOG1 and CD117 are the antibodies of choice in the diagnosis of gastrointestinal stromal tumours. *Histopathology* 2010 Aug;57(2):259–70.
- [2] Doyle, et al. MUC4 is a highly sensitive and specific marker for low-grade fibromyxoid sarcoma. *Am J Pathol* 2011 May;35(5):733–41.
- [3] Maretty-Nielsen, et al. Low-grade fibromyxoid sarcoma: incidence, treatment strategy of metastases, and clinical significance of the FUS gene. *Sarcoma* 2013;2013. Article ID 256280.
- [4] Gardner J. Soft tissue fibroblastic/myofibroblastic tumors; low-grade fibromyxoid sarcoma. [www.pathologyoutlines.com/topic/softtissue/fibromyxoid.html](http://www.pathologyoutlines.com/topic/softtissue/fibromyxoid.html) [accessed 20.03.14].
- [5] Evans HL. Low-grade fibromyxoid sarcoma: a clinicopathologic study of 33 cases with long term follow-up. *Am J Pathol* 2011 Oct;35(10):1450–62.
- [6] Park, et al. Low-grade fibromyxoid sarcoma of the colon. *Dig Liver Dis* 2007 Mar;39(3):274–7.