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CASE REPORT

Gastrointestinal stromal tumour presenting as a palpable, undifferentiated abdominal mass

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Background: We present the case of a previously healthy 60-year-old female with a long-standing history of lower abdominal pain radiating to her back, abdominal distension, and weight loss. She had no abnormal gynaecological history, with four normal vaginal deliveries without complications. A normal vaginal and rectal examination was reported. Transvaginal ultrasound showed a 30-week fetal-size mass with a solid wall, minimal vascularity, a homogeneous cystic centre, septations, and clear contents. Baseline bloods reported microcytic anaemia, thrombocytosis, and no acute rise in tumour markers.

Case report: After correcting her haemoglobin levels with four units of packed red cells, a laparotomy was performed by the gynaecologists with suspicion of an ovarian tumour. General surgery was called based on the cystic nature of the mass, and with concerns about seeding. Peritoneal and omental nodes were excised and sent for histology. A gastrointestinal stromal tumour (GIST) was confirmed with strong positivity of both DOG1 and CD34 receptors. A year course of imatinib was started, with serial computed tomography (CT) scans demonstrating mass shrinkage. A resection was performed. A follow-up CT scan two years after the resection showed no peritoneal disease or local recurrence.

Discussion: A palpable, undifferentiated abdominal mass yields several possible differential diagnoses. Although rare, they are the most frequent mesenchymal tumours of the gastrointestinal (GI) tract. Previously, GISTs were considered benign or malignant. Current European Society for Medical Oncology (ESMO) guidelines state that all GISTs are considered malignant and should be treated appropriately. Occurring primarily in the stomach, extraperitoneal GISTs that mimic ovarian tumours have proven extremely rare, as noted by Tonni et al.[11] Following a histological diagnosis, GISTs are treated with tyrosine kinase inhibitors as first-line therapy.

Conclusion: Although rare, this case highlights the consideration of a GIST in the differential diagnosis when confronted with a palpable, undifferentiated abdominal mass.

Keywords: gastrointestinal stromal tumour, abdominal mass, mesenchymal tumour, imatinib, undifferentiated mass

Background

A previously healthy 60-year-old female initially presented to the emergency centre with a two-month history of anorexia, nausea, vomiting, lower abdominal pain radiating to her back, weight loss, and progressive abdominal distension. She was gravida 4 parity 4, all normal vaginal deliveries with no complications, and she reached menopause at 56 years. She reported no history of abnormal uterine bleeding, never had a pap smear, and had no family history of malignancy.

Upon examination, she was noted to be cachectic with subconjunctival pallor. A large, firm abdominal mass was noted originating from the pelvis, equivalent to a 34-week gestational size. Vaginal and rectal examinations were normal. She was referred to gynaecology for further investigations. Transvaginal ultrasound revealed a normally sized, anteverted uterus with a normal endometrial cavity. The right ovary appeared to contain a large mass equivalent to a 30-week fetal size, with a solid wall, minimal vascularity, a homogeneous cystic centre, septations, and clear contents. The left ovary was normal. Abdominal CT scan showed a large, lobulated pelvic-abdominal mass measuring 135 × 231 × 207 mm, with areas of necrosis, reported as most likely originating from the left ovary (Figure 1). The mass displaced the bowel and abutted the anterior abdominal wall muscles, with ascites present.

Baseline bloodwork revealed normal renal function, microcytic anaemia with a haemoglobin of 4.7 (13.0–17.0), mean corpuscular volume of 76.6 (83.1–101.6), and an elevated platelet count of 1 095 (171–388). Tumour markers were not markedly elevated, with a carcinoembryonic antigen of 1.1 ug/L (2.5–3.0) and cancer antigen (CA)-125 of 67 kU/L (< 35.0).

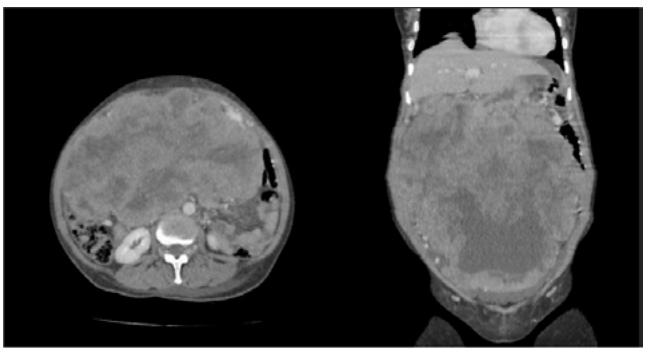

Figure 1: Abdominal computed tomography showing an extensive lobulated pelvic-abdominal mass with areas of necrosis

Case report

The patient received a transfusion of four units of packed red cells and was taken for an explorative laparotomy with a presumed diagnosis of an ovarian mass. However, intraoperatively, the ovaries were noted to be normal, and the general surgeon was called to assist. Due to the extensive and cystic nature of the mass, the origin could not be confidently ascertained intraoperatively, with concerns that it may extend from the retroperitoneum and risk potential rupture and seeding, limiting dissection (Figure 2). Peritoneal and omental nodes were excised, and the procedure was aborted. Histology of the peritoneal nodule confirmed a spindle cell-type GIST, with diffuse strong positivity for both DOG1 and CD34 receptors.

The patient was referred to oncology and initiated on imatinib. After a significant improvement in performance status and an excellent clinical and radiological response after more than a year of treatment, she was considered for possible resection. Preoperative gastroscopy did not demonstrate any lesions, and subsequent explorative laparotomy revealed a mass that eventually was found to be originating from the greater curvature of the stomach while also densely adherent to the transverse colon and its mesentery. Resection of the mass was performed with en bloc segmental transverse colon resection and anastomosis. Peritoneal inspection revealed multiple possible small tumour deposits onto the bladder peritoneum, which were biopsied but not resected.

Histology revealed a 120 × 85 × 50 mm low-grade T4 epithelioid-type gastric GIST, with a mitotic rate of ≤ 5 mitoses per 5 mm^2. Peritoneal biopsies showed a good treatment response to imatinib, with no residual tumour seen on histology. Regrettably, genetic analysis of the tumour is not available in the public sector and was not performed. The GIST was considered to have moderate risk for malignant potential by the National Comprehensive Cancer Network (NCCN).[1] This was in view of its size being more than 10 cm and a low mitotic count of ≤ 5 mitoses/50 high-power fields (HPF). However, based on the initial diagnosis of peritoneal metastases, the patient was recommenced on imatinib.

Figure 2: Intraoperative findings of an extensive solid/cystic intra-abdominal mass at initial laparotomy

At the last review, two years after the resection, the patient remains clinically well, and surveillance abdominal CT showed no features of persistent peritoneal disease or local recurrence.

Discussion

A palpable, undifferentiated abdominal mass, as in this case, can be challenged with a wide differential diagnosis, including gynaecological aetiologies, lymphomas, sarcomas, and GISTs. Signs and symptoms may be highly non-specific, and obtaining a tissue diagnosis may be risky due to concerns about tumour seeding. GISTs typically arise from a gain-of-function mutation in a single KIT gene on chromosome 4. This mutation leads to heightened tyrosine kinase activity, resulting in uncontrolled tumour growth. GIST treatment primarily involves tyrosine kinase inhibitors and, where possible, resection.

Developed in 1999, imatinib is a targeted tyrosine kinase inhibitor and remains the first-line medical treatment for GISTs due to its efficacy and relatively low toxicity. Although surgery remains the cornerstone and only curative management option, survival rates have significantly improved with the introduction of imatinib. Specifically, in non-metastasised GISTs, adjuvant treatment with imatinib for three years was associated with an overall survival and relapse-free survival advantage compared with one year of therapy. Imatinib also remains the standard treatment for advanced disease and for those with inoperable GISTs.[6]

Historically, GISTs were grouped with leiomyosarcomas, but are now known to arise from the interstitial cells of Cajal (ICC), which are the pacemaker cells of the GI tract and thus have a distinct origin. They typically express CD117 receptor positivity, but may also be CD34- or DOG1-positive.[2] Although GISTs are rare (7–15 cases per million people per year worldwide), they are the most common mesenchymal tumour of the GI tract, accounting for approximately 80% of all GI mesenchymal tumours and between 0.1% and 3% of all GI malignancies.[3,4] Literature on GISTs presenting as a palpable abdominal mass is sparse, with only 25 cases published from 2001 to 2011.[5] Anecdotally, this type of presentation may be more common in our South African setting, possibly due to socio-economic factors and delays in presentation.

GISTs are subclassified based on their predominant cell type: spindle cell (75% of cases) or epithelioid. In rare instances, a mixed histological pattern containing both spindle and epithelioid cells may be observed. GISTs were previously considered benign or malignant. However, in the more recent past, as noted in ESMO guidelines, all GISTs are considered malignant and managed accordingly.[6]

GISTs most commonly develop in the stomach (60%), particularly along the lesser curvature.[7] When present on the greater curvature, the malignant potential is reduced due to

lower concentrations of ICCs in this region.[8] The small intestine accounts for 20–30% of GISTs, the remaining 10% occurring in the colon, with rare instances in the oesophagus or appendix.[9] Tonni et al.[11] published a systematic literature review regarding GISTs and their ability to mimic primary ovarian tumours and metastases to the ovary. The result showed a rare correlation between these two categories of pelvic-abdominal masses.[11]

A rare subset of GISTs arises outside the GI tract and is classified as extra-gastrointestinal stromal tumours (EGISTs). The term was introduced by Reith et al. in 2000 to describe tumours with clinical, imaging, and histopathological features similar to GISTs but with a primary site outside the GI tract.[12] EGISTs are categorised into three subtypes: omental, pancreatic, and retroperitoneal, with the latter being the rarest, accounting for < 20% of all EGISTs.[10] Theoretically, retroperitoneal GISTs should not occur, as they originate from ICCs, which are absent in the retroperitoneum. However, they are sometimes diagnosed based on exophytic growth patterns, with the muscularis propria involved in > 50% of cases.

Conclusion

Although documented as a rare presentation, this case highlights the importance of considering a GIST in the differential diagnosis of a palpable, undifferentiated abdominal mass. This consideration is especially important in a resource-constrained setting, such as South Africa, where delays in presentation are common.

Conflict of interest

The authors declare no conflict of interest.

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Ethical approval

The authors declare that this submission is in accordance with the principles laid down by the Responsible Research Publication Position Statements as developed at the 2nd World Conference on Research Integrity in Singapore, 2010. Verbal consent was obtained from the patient, and they are aware of this article's submission for publication.

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