Intestinal GIST masquerading as an ovarian mass: Diagnosed on FNAC

ABSTRACT

The preoperative diagnosis of metastatic intestinal gastrointestinal stromal tumors (GIST) on cytology can be quite difficult at times. The present case characterizes the cytomorphological and immunocytochemical features of GIST, emphasizing the utility of fine-needle aspiration cytology (FNAC) in the evaluation of spindle cell tumors of gastrointestinal tract. An accurate and early diagnosis of GIST affects the treatment, primarily allowing the use of tyrosine kinase inhibitors in unresectable or metastatic cases. Presence of highly cellular fragments of spindle-to-oval cells with variable degree of pleomorphism, atypia, and necrosis supplemented by immunocytochemistry can render a cytological diagnosis of GIST in dilemmatic clinical situations. Our case highlights the diagnostic role of FNAC in the evaluation of a pelvic mass, which was clinicoradiologically misdiagnosed as ovarian carcinoma.

Key words: CD117; GIST; FNAC; ovarian mass

Introduction

Gastrointestinal stromal tumor (GIST) is the most frequent mesenchymal tumor of the gastrointestinal tract (GIT), with a malignant potential which typically presents in older individuals.[1] Though cytological features of GISTS have been documented in literature, cytological evaluation remains limited in its ability to distinguish between benign and malignant tumors.[2-4] We report fine needle aspiration cytology (FNAC) of intestinal GIST mimicking a malignant ovarian mass in a postmenopausal female. The present case discusses the role of FNAC in the evaluation of spindle cell tumors of GIT.

Case Report

A 54-year-old multiparous postmenopausal female presented to the gynaecology outpatient department with gradual onset pain in the right lower abdomen for last 1 month. There was no history of anorexia, bone pains, bowel and bladder complaints, postmenopausal bleeding, or symptoms of hyperestrogenism/androgenism. General physical examination was unremarkable. Per abdomen examination revealed a mobile, nontender, firm lump of approximately 8 × 8 cm in the right iliac region extending into the pelvis.

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Per speculum examination showed chronic cervicitis with a healthy vagina. On pervaginal examination, a firm irregular nontender mass with restricted mobility was felt in the right fornix, which could not be made out separately from the uterus. Clinically, a possibility of right ovarian mass was considered.

Ultrasound abdomen showed a well-defined, heterogeneous mass measuring $13 \times 9 \times 7.9$ cm with internal vascularity and echogenic foci in right pelvic and right iliac fossa, suggestive of right ovarian mass. CECT abdomen revealed a heterogeneously enhancing mass lesion in the region of right adnexa [Figure 1a]. Lesion was displacing and abutting the adjacent small bowel loops and sigmoid colon [Figure 1b]. Multiple well-defined nodular lesions were seen in omentum, mesentery, and bilateral round ligaments. Because the right ovary was not visualized separately, possibility of carcinoma ovary was suggested. Serum CA-125, CA 19.9, $\beta$-hCG, and CEA levels were found to be within normal limits.

Ultrasound-guided FNAC was done from the right adnexal mass using lumbar 23 guage puncture needle. Aspirate smears were cellular and showed predominantly cellular cohesive fragments of oval-to-spindle shaped tumor cells with high vascularity [Figure 1c]. Some loose clusters and singly scattered cells of similar morphology were also seen. The tumor cells had scant-to-moderate amount of cytoplasm, bipolar cytoplasmic processes and spindle shaped nuclei. The cells were pleomorphic with hyperchromatic nuclei, granular chromatin and small conspicuous single nucleolus [Figure 1d]. Parallel, side-by-side arrangement of nuclei was noted. At places cells had epithelioid morphology and were arranged in vague glandular structures [Figure 2a]. Focal areas of necrosis were seen, however, no mitosis was found [Figure 2b].

Cell block showed predominantly interlacing spindle cells with few round-to-oval cells. On immunocytochemistry, the tumor cells were negative for cytokeratin, inhibin and calretinin, desmin, and S-100, and showed strong positivity for vimentin, smooth muscle actin (SMA), and CD117. Based on high cellularity, presence of loose fragments with single cells, focal areas of necrosis, and immunocytochemistry, diagnosis of GIST – inconclusive for malignancy was offered.

The patient underwent an exploratory laparotomy that revealed a large peritoneal mass which was attached with the serosa of small bowel, 5 cm distal to the duodenojejunal junction. Bilateral adnexa and uterus were normal. Excision of the tumor with resection anastomosis of an ileal segment (7.5 cm) was done.

On the basis of morphology and immunohistochemistry, brisk mitoses >5/50 hpf, necrosis, tumor size of 18 cm, and presence of multiple implants all over the mesentery and peritoneum, a diagnosis of malignant intestinal GIST was offered on histopathology [Figure 2c and d].

**Discussion**

GIST constitutes approximately 1% of gastrointestinal malignancies. A rapid and accurate preoperative cytological diagnosis is important in metastatic or unresectable cases because these patients are ideal candidates for targeted treatment.

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**Figure 1:** (a) CECT abdomen shows a heterogeneously enhancing mass lesion in right adnexa. (b) The lesion is displacing and abutting the adjacent small bowel loops and sigmoid colon. Papanicolaou stained aspirate smears show: (c) Predominantly cellular fragments of tumor cells with high vascularity (Pap stain x200). (d) Oval-to-spindle shaped cells with scant amount of cytoplasm, spindle-shaped hyperchromatic nuclei, and granular chromatin (Pap stain x400)

**Figure 2:** Papanicolaou stained aspirate smears. (a) Round-to-oval cells arranged in vague glandular (structures (Pap stain x200). (b) Foci of granular necrosis (Pap stain x200). (c) Histopathology section shows tumor arising from the wall of the small intestine (H and E stain x 100). (d) Tumor cells show a strong membranous staining (400) (IHC : CD117 x200)
therapy. There are a few case series illustrating the cytomorphological and immunocytochemical features of GIST with histopathological correlation. It is imperative for the cytologist to be aware of the spectrum of cytological features and unusual clinical presentations of GIST that can aid in diagnosis.

Based on the presence of predominantly cohesive and highly cellular clusters of spindle-to-oval cells with focal epithelioid areas in an adnexal mass in a postmenopausal female, the possibilities considered in our case were mesenchymal tumor, malignant mixed mullerian tumor (MMMT), and metaplastic carcinoma. Possibility of ovarian sex cord stromal tumor was unlikely due to the lack of characteristic nuclear features and absence of hormonal symptoms. Negative immunocytochemistry for cytokерatin excluded the possibility of MMMT/metaplastic carcinoma. Uniform strong CD117 (c-kit) and SMA positivity along with negative desmin excluded the diagnosis of leiomyosarcoma and clinched the diagnosis of GIST.

A few studies have addressed the utility and limitations of FNAC in the diagnosis of spindle cell tumors of GIT. Important differential diagnoses of GIST include leiomyomas and nerve sheath tumors. Cellular density of tissue fragments is relatively higher in GIST than the latter. Leiomyomas are strongly positive for desmin and SMA, whereas schwannomas show positivity for S100; however, both are negative for CD117 and CD34. Metastatic melanoma is another differential which is positive for CD117, S-100, and HMB-45. CD117 positivity excludes the possibility of inflammatory myofibroblastic tumor, which is vimentin and SMA positive. A diagnosis of GIST can only be suspected on cytology and needs confirmation by immunocytochemistry. However, Li et al. have reported that GIST could be diagnosed with confidence on cytology alone.

Malignancy is difficult to predict on cytology alone unless there is marked nuclear pleomorphism, singly scattered cells, mitoses and necrosis, but might not hold always true. Large tumor size, foci of granular necrosis, and loose clusters with scattered cells favoured malignancy in our case, although mitosis could not be identified on smears. This could probably be due to closely packed cohesive thick cellular fragments. The most relevant and widely applied morphological parameters to predict malignant behavior are tumor size >5 cm and >5 mitoses per 50 HPFs.

Conclusion

To conclude, cytopathologists need to keep in mind the possibility of extraintestinal or metastatic GIST on encountering cellular fragments of spindle cells in such a dilemmatic clinical situation and to resort to immunocytochemistry for a definitive diagnosis.

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Conflicts of interest
There are no conflicts of interest.

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