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Duodenum-Jejunum Junction Gastrointestinal Stromal Tumour: A Diagnostic Dilemma on Cytology

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ABSTRACT

Gastrointestinal stromal tumors (GIST) are mesenchymal tumors arising from the interstitial cells of Cajal (pace maker cells) of the gastrointestinal tract (GIT). Stomach is the most common site (60-65%) of these tumors. 5-10% of GIT tumors is located in large intestine and rectum. Pre-operative diagnosis helps in the management of this tumor as it responds well to c-kit inhibitors. The cytological diagnosis of GIST is characteristic, however, associated with many pitfalls leading to erroneous diagnosis. Morphological resemblance is seen with other spindle cell and epithelioid cell tumors. The differentiation between high grade and low grade GISTs is described but not reliable. Cytology combined with cell block and immunocytochemistry helps in making an appropriate diagnosis. Here we present duodenal-jejunal GIST diagnosed as GIST on cytology and confirmed by histopathology and immunohistochemistry. We report this case to describe the cytological features of GIST and its pitfalls in cytology.

Keywords: Jejunum, gastrointestinal stromal tumor, interstitial cells of Cajal, immunohistochemistry.

INTRODUCTION

Gastrointestinal stromal tumors constitute 0.1-3% of gastrointestinal tumors. [1] GISTs are mesenchymal tumors having a submucosal location. Stomach is the most common site for GIST followed by the small intestine (20-25%), colon and rectum (5%), and esophagus (<5%). [2] GISTs express c-kit proto-oncogene, are immune-reactive for CD-117 and they respond to c-kit inhibitors. [3] Pre-operative diagnosis on cytology is helpful in the management of the GIST. Cytological examination helps in the pre-operative diagnosis. However, there are various pitfalls in diagnosis of GIST on cytology. Close resembles with smooth muscle tumors, nerve sheath tumors, granulation tissue, epithelial tumors, and inability to predict the long term behavior of tumors are some of the pitfalls. Cytology combined with cell block preparation and immunocytochemistry are helpful in making a confident diagnosis pre-operatively.

CASE REPORT

A 35-year-old male patient presented with pain in right lumbar region and altered bowel habits for 2 months. General physical examination was within normal limits. On abdominal examination, an ill-defined firm mass was detected in left hypochondrium. Ultrasonography revealed asymmetric circumferential thickening of small bowel forming a large hypoechoic mass measuring 11x10x7cm noted in left lumbar and umbilical region. Ultrasound guided fine-needle aspiration (FNA) was performed from the mass. Cytological smears revealed

pleomorphic cells arranged in loosely cohesive irregularly outlined cell clusters held together by thin caliber vessels and many lying singly [Figure 1]. Individual cells revealed high nucleo-cytoplasmic ratio, eccentrically placed nuclei, irregular nuclear membrane, hyperchromatic to granular chromatin, and 1-2 conspicuous nucleoli. The cytoplasm was abundant, had irregular cell borders, fine vacuolated, and showed cytoplasmic protrusions [Figure 2]. On the basis of cyto-morphological features, a diagnosis of malignant mesenchymal tumor – left hypochondrium mass was given.

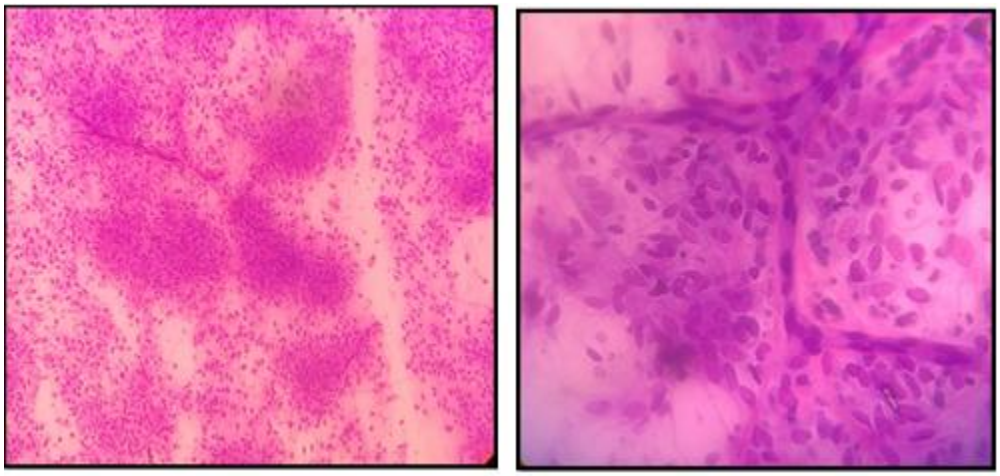

Figure 1: Highly cellular smears arranged in loosely cohesive irregularly outlined cell clusters, Giemsa 40x

Figure 2: Pleomorphic cells with thin caliber vessels. Cells have abundant finely vacuolated cytoplasm with irregular cell borders and cytoplasmic protrusions, Giemsa 400x

Retroperitoneal mass excision with portion of duodenum and jejunum was performed. Grossly, a growth was seen arising from the wall of duodenum –jejunum junction pushing and proliferating the mucosal surface of colon. Mass measured 17x12x10 cm [Figure 3]. On cut it was variegated mass with lobular appearance.

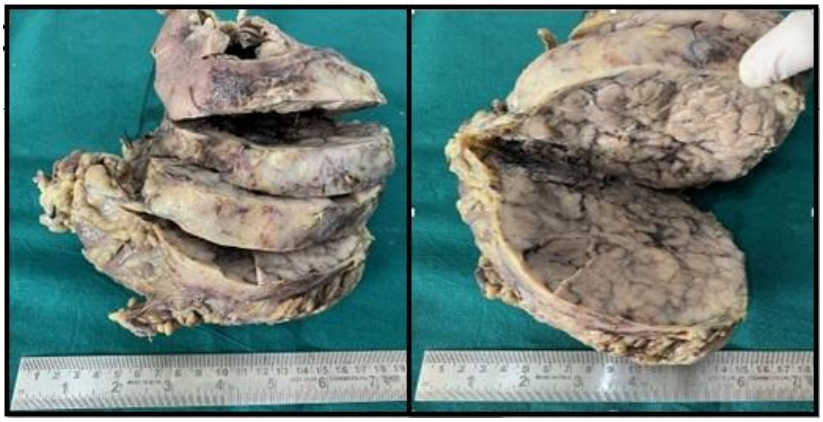

Figure 3: Mass of 17x12x10 cm arising from the serosal surface of duodenum-jejunum junction, the cut surface is lobulated and variegated.

Microscopic examination revealed spindle shaped tumor cells forming short fascicles in the submucosa [Figure 4]. The growth extended up to the serosa. Areas of necrosis were seen. The mitotic count was 1-2mitotic figures/50 high power field (hpf). Immunohistochemistry was performed for CD-117, DOG1, SMA, H-Caldesmon(focal). The tumor cells were reactive for CD-117 (cytoplasmic and nuclear)and DOG1, SMA . Based on the histopathologic features and immunohistochemistry, a diagnosis of malignant GIST- was given.

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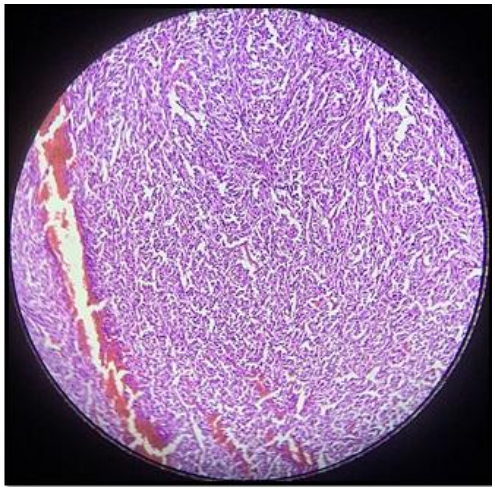

Figure 4: Tumor composed of spindle shaped tumor cells forming short fascicles, H&E 100x

DISCUSSION

Gastrointestinal stromal tumors represent a distinct clinicopathologic entity that is characterized by genetic mutations in the c-kit proto-oncogene.[7] Gastrointestinal stromal tumors accounts for only 0.1-3% of all GI neoplasms, but, simultaneously, they are the most frequent mesenchymal lesions of the gastrointestinal tract. GISTs are frankly malignant in 10-30% of cases and cause mortality in 2% of cases.[2] Clinically, most patients present with an abdominal mass, pain and melena. Criteria to predict their tumor behavior include size, necrosis, and mitotic rate as suggested by Miettinen and Lasota.[4] A mitotic count above 5/50 hpf and size above 2 cm is associated with an increased rate of progressive disease and increased risk of metastasis. GIST has no preference for gender; its peak incidence is between 40 and 70 year, with a broad age distribution.[5] These tumors do not usually involve the mucosa, but commonly originate in the wall of the GIT tract. [6] They represent a morphologically diverse group of neoplasms that display features of smooth muscle and neural differentiation. On cytology there is dilemma in its diagnosis due to the morphology. Leiomyoma and leiomyosarcoma (LMS) are very close differential of GIST on cytology. Leiomyoma which is a very common tumor of the GIT is characterized by variable cellularity and are composed of bland spindle cells with abundant cytoplasm often having fibrillary appearance [8] . No atypia, mitosis or epithelioid cells are identified. Wieczorek *et al.*[9] compared the cytology of GIST with that of LMS. The LMS showed three-dimensional, tightly cohesive, sharply marginated syncytia of spindle cells, often with nuclear crush artifact. The cytoplasm/stroma had a distinct wiry, retractile appearance. LMSs more commonly exhibit pleomorphism. Epithelioid cytomorphology, mitoses, and necrosis occasionally were observed in both tumor types. Immunocytochemistry was helpful to differentiate between these two entities. About 100% GIST showed immunostaining with CD-117 and all LMS were immunoreactive for smooth muscle actin. Other differentials on cytological smears are benign and malignant nerve sheath tumors. The distinction of metastatic GIST from other metastatic tumors is important as the former responds well to c-kit inhibitors. In practice c-kit inhibitors have become available for the treatment of GISTs, hence the pre-operative diagnosis of these tumors has gained importance. Furthermore, the question of benign versus malignant may be impossible to answer. Vij *et al.*[10] Studied the subtle differences between low grade, malignant and metastatic GIST on cytology. Malignant and metastatic GIST was more cellular than the benign GIST. Epithelioid morphology was more commonly seen in malignant and metastatic GIST. The nucleoli were indistinct in low grade GIST and prominent or multiple in high grade GIST irrespective of the cell type. Malignant GIST showed the presence of nuclear inclusions. It was concluded from the study that mitosis was the key morphologic feature that suggested high grade malignant GIST. However, it was difficult to find mitosis in cytological smears since the tumor cells occurred in closely packed cohesive thick tissue fragments. In their study Li *et al.*[11] they found that mitoses found in resected specimens were seldom found on

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cytological smears. Very little pleomorphism was found in cytological smears of malignant GIST. Dirty or necrotic background was also not a reliable criterion for differentiating low grade and high grade GIST. GISTs are believed to arise from interstitial cells of Cajal. These cells express CD-117 (c-kit) antigens. CD-117 is sensitive (79-86%) and relatively specific for GIST.

IHC

The tumour cells expressed CD-117 (cytoplasmic and nuclear) and DOG1, SMA, H-caldesmon(focal)

Thus, based on the histopathologic features and immunohistochemistry, a final diagnosis was given as malignant GIST, spindle cell type.

Histological grade: low grade. Lymphovascular invasion: absent. Subtype: spindle cell

CONCLUSION

Gastrointestinal stromal tumor show a broad morphologic variety, but nuclear pleomorphism by cytology alone, rarely correlates with malignant potential. USguided-FNAC can be being utilized to render diagnosis in palpable and deep seated masses, a cytopathologist needs to keep in mind the possibility of GIST and collect appropriate material when encountering cellular tumors with tight clusters of slender spindle cells. In the appropriate clinical and radiological setting, a confident diagnosis of GISTs can be documented by FNA cytology, cell block and immunocytochemical studies.

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