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Gastrointestinal Stromal Tumors (GIST) - A rare case report at atypical site

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ABSTRACT

GIST can be present anywhere in the GIT but it is most commonly found in the stomach and small intestine and rarely on extra visceral sites. These are one of the most common mesenchymal[1]. Approximately, 60% arise in the stomach and 30% arise in the small bowel, the other 10% arise in the oesophagus, colon, rectum, gallbladder, appendix, omentum, mesentry, retroperitoneum and pelvis[2].Symptoms can vary upon location, size and growth rate. Due to its location and hence the inability to identify by endoscopy, it is difficult to diagnose. Small lesions usually asymptomatic are incidentally found as a result of imaging, endoscopy,pathologic specimens following surgery for another cause and post mortem[3].We reported a case of GIST arising in the jejunum in 61 year female presented with pain in abdomen since 20 days.

KEY WORDS: Gastro intestinal Stromal Tumors (GIST), Mesenchymal tumor, intestinal tumors.

I. INTRODUCTION

Small bowel gastrointestinal stromal tumor (GIST) is an uncommon tumor, with an estimated frequency of 10–20/L million populations, occurring usually in the 6th decade of life. GISTs, mainly ones larger than 4 cm have a wide range of presentations from abdominal discomfort and bloating to abdominal emergencies due to massive gastrointestinal (GI) hemorrhage, with pressure necrosis and ulceration of the overlying mucosa is the main mechanism leading to GI hemorrhage. Life-threatening hemorrhage is a rare initial presentation.

Among the wide differential diagnosis for GI bleeding, jejunal GIST is one of the rarest etiologies. Due to its location and hence the inability to identify by endoscopy it is difficult to diagnose. From here, computed tomography (CT) angiography is considered essential for identification of the site of GI bleed, which aids in diagnosis of GIST. The mainstay treatment for this emergent presentation is resection, which if done in a timely manner results in a good clinical outcome.[4]

II. CASE REPORT

A 61-year-old female came to the surgery OPD of Bhagat Phool Singh Government Medical College & Hospital, Khanpur Kalan, Sonepat with pain in epigastrium since 20 days. The pain was dull aching and associated with vomiting (not blood stained). Surgical excision of the lesion was performed. The excised specimen was sent for histopathological examination. Specimen comprised of small intestine 19x5 cm, with a well-defined solid tumor, firm in consistency measuring 9.5x7x5 cm. On cut, tumor was grey white with areas of hemorrhage. The tumor appeared to involve whole wall of intestine however mucosa remained uninvolved.

Microscopically, tumor area comprised of cells arranged in bundles and intersecting fascicles. These cells had oval to elongated nuclei with inconspicuous nucleoli and moderate amount of

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pale to eosinophilic cytoplasm, 1 mitosis/50 HPF observed. Tumor involved muscle layer however overlying mucosa was free from tumor infiltration.. Areas of hemorrhagic necrosis were seen at periphery. Overlying serosa show acute on chronic inflammatory infiltrate.Tumor involved radial margin. Tumors cells were immunoreactivity for CD117 and vimentin. Focal aberrant expression of desmin also observed. The cells did not show the expression for CK, CD-31, SMA, S-100, HMB45, CD-68 and Ki67. The final histopathology diagnosis of GIST was randered. The patient was advised for the regular follow up.

III. DISCUSSION

GISTs are thought to grow from specialised cells found in gastrointestinal tract called interstitial cells of cajal[5]. It is of 2 types: sporadic and familial GIST. Most GISTs are sporadic (97%). Sporadic form occurs in individuals who do not have positive family history and usually present with only one tumor while in familial type, patients has family history of GIST with multiple tumor. They also present with other signs and symptoms of noncancerous growth such as hyperplasia of other cells in GIT. They may also present with a maculo-papular cutaneous lesion i.e urticaria pigmentosa. Some cases of familial GIST are associated with KIT and PDGFRA gene mutation and have autosomal inheritance. Activating mutations in KIT and PDGFRA (encoding KIT and platelet derived growth factor receptor tyrosine kinases, respectively) are considered the main oncogenic drivers of GIST[6]. GIST is expressed by the expression of the receptor tyrosine kinase growth factor.CD117, which differentiates it from other mesenchymal tumors such as leiomyomas, leiomyosarcomas, leiomyoblastomas and eurogenic tumors that do not express this protein[7]

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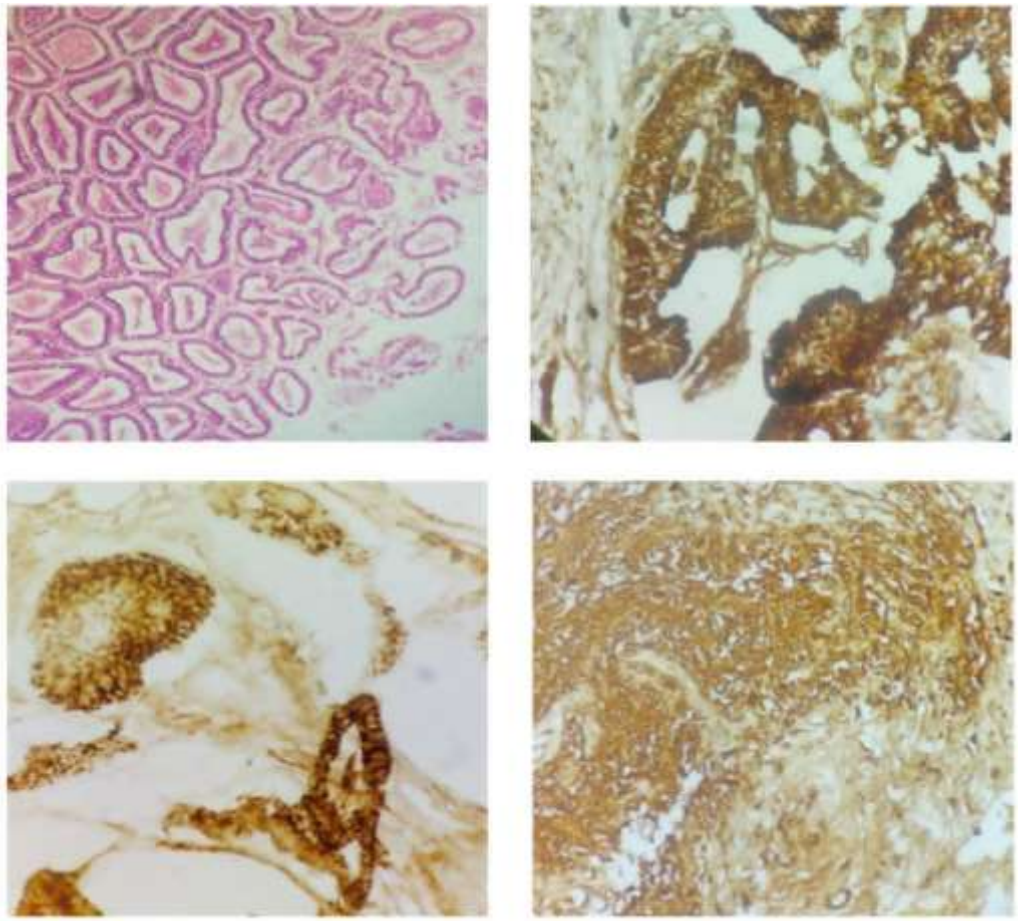

Fig: At 40X Microphotograph showing tumor cells and various IHC vimentin, EMA, S-100 positivity

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In summary, detailed understanding about GIST as a notable GI tract tumor is important in the treatment of this rare disease entity that is known to have a high recurrence rate.surveillance continues to be the key for early detection among patients with prior history[3].Originally described in the 1980s, GISTs were thought to represent smooth muscle tumors, however improvements in immunohistochemistry and the recognition of gain of function mutations over the last 20 years, have led to the recognition of GISTs as a separate entity[5].

IV. CONCLUSION

GIST is a rare mesenchymal tumor which is difficult to diagnose and treat. We report this case because of its rarity. GIST should be included in the differential diagnosis of any intra-abdominal non epithelial malignancy.

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