

CASE REPORT

Diagnosis of gastrointestinal stromal tumor in a 25-year-old female presenting as huge abdominal mass and mandibular metastasis on fine needle aspiration cytology

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are rare forms of mesenchymal tumors of the digestive tract. We present a case of 25-year-old female who presented with abdominal pain, vomiting and mandibular swelling. Buccal mucosa at the site of mandibular swelling was normal on histopathological examination. CT scan revealed a huge intra abdominal mass. Fine needle aspiration cytology (FNAC) was performed from the abdominal mass and mandibular swelling. It revealed similar morphology from both the sites, confirming metastasis of GIST to mandible. The highlight of this case was presentation of malignant GIST in a young female along with rare mandibular metastasis. Diagnosis by FNAC is rarely reported.

Key Words: Gastrointestinal stromal tumor, Fine needle aspiration cytology, Mandibular metastasis

1. INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal tumors of the digestive tract. Most common site of GIST is stomach followed by small bowel and other sites (colon, rectum, esophagus). They are believed to arise from interstitial cells of Cajal. These can present as abdominal pain, bleeding or mechanical obstruction. Liver is the most common site of metastasis.^[1] Second most common site is the peritoneum whereas bone metastasis is rare.^[2] The incidence of bone metastases among other metastatic locations has been estimated to be less than 5%.^[3] Standard treatment for GIST is surgery, depending upon the size of tumor.

2. CASE PRESENTATION

2.1 History

A 25-year-old female presented to G.B. Pant hospital, Andaman and Nicobar islands, with complaints of diffuse abdominal pain for 3 to 4 months, episodes of vomiting on and off and mandibular swelling. Patient had noticed the mandibular swelling for 3 weeks. The patient was very cachexic, had decreased appetite and loss of weight. Oral hygiene was very poor. On local examination, there was an irregular tissue on right buccal mucosa which was excised for histopathological examination. It turned out to be unremarkable on biopsy report. Thus, ruling out the possibility of a primary tumor of oral cavity. The patient was shifted to

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department of surgery for management of pain abdomen and vomiting.

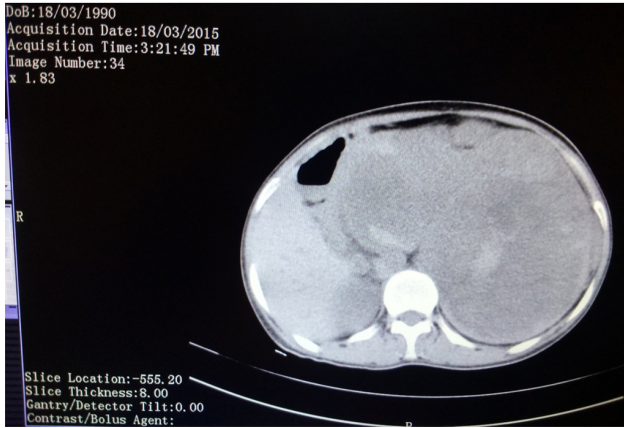


Figure 1. CT scan abdomen
CT scan showed huge intra abdominal mass

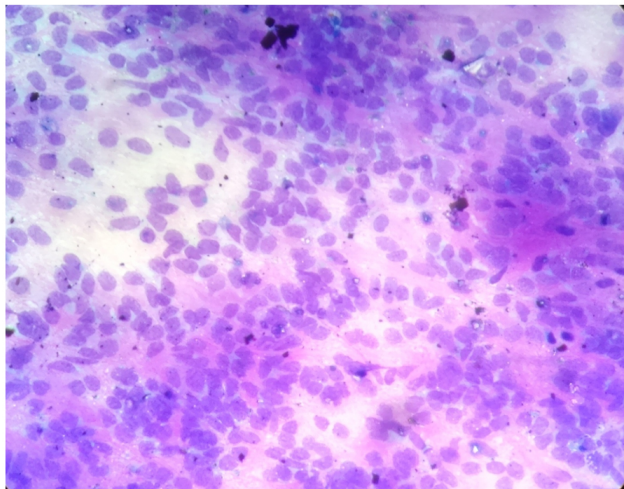


Figure 2. Giemsa stain
The tumor (abdominal mass) was composed of spindle shaped cells along with myxoid material in the background (giemsa, ×400)

2.2 Investigations

Local examination revealed a lump in abdomen occupying whole of the epigastrium, left lumbar region and extending to the right side, with mild tenderness. Right mandibular swelling was also slightly tender. Hematological reports revealed iron deficiency anemia, and other serological and biochemical tests were within normal limits. CT scan abdomen revealed a lobulated mass measuring 20 cm × 15 cm × 14 cm in left renal fossa displacing spleen anteriorly (see Figure 1). A provisional diagnosis of wilms tumor/renal cell carcinoma was made on CT scan. CT scan also revealed lytic lesions in cervical vertebrae, scapula and left humeral neck. Fine needle aspiration cytology (FNAC) was requested

by the clinician. FNAC was done blindly from abdominal mass (as the mass was palpable subcutaneously) and from mandibular swelling. The right mandibular swelling was 5 cm × 4 cm, firm in consistency. FNAC from abdomen as well as mandible revealed mucoid material which was blood tinged and particulate. Air dried and wet fixed smears were prepared and subsequently stained with Giemsa and haematoxylin-eosin stain respectively. Smears from abdominal mass as well as right mandibular swelling were cellular and showed clusters and singly scattered cells in a myxoid background (see Figure 2). The cells had spindle to ovoid nuclei and scant amount of cytoplasm (see Figure 3). Numerous mitotic figures were noted (see Figure 4). Cytological features were those of malignant gastrointestinal stromal tumor.

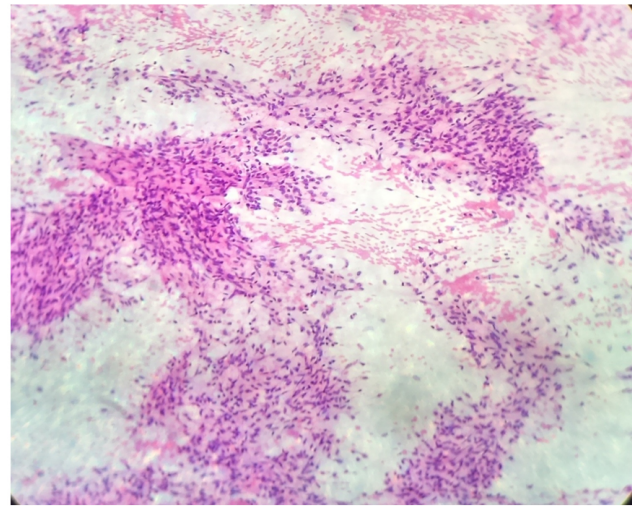


Figure 3. H & E stain
The tumor cells (from mandibular swelling) were arranged in clusters and scattered singly, with scant amount of cytoplasm (H & E, ×100)

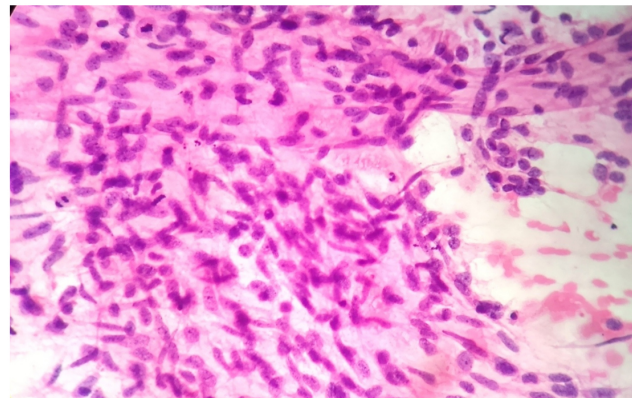


Figure 4. H & E, mitotic figures
Tumor cells (from mandibular swelling) showed atypical mitotic figures (H & E, ×400)

2.3 Clinical course

General condition of the patient was very poor. Patient was cachexic with significant weight loss in the past 6 months. Patient was married and nullipara. The patient was declared unfit for surgery in pre anaesthetic check up. Hence, she was not operated upon and was kept on conservative treatment. After three days, the patient died.

3. DISCUSSION

GISTs account for 1% of all gastrointestinal tract malignancies.^[4] The predominant site of GISTs is the stomach (60% - 70%), followed by small intestine (25% - 35%) and less common than 5% in rectum, esophagus, omentum, and mesentery.^[5] The interstitial cell of Cajal is a mesodermally derived cell located in the wall of gastrointestinal tract and thought to function as a pacemaker cell of GI tract. This cell expresses CD 117.^[6] The age of onset is 4th to 6th decade, with no sex predilection. Most GISTs present with gastrointestinal symptoms like upper abdominal pain, gastrointestinal bleeding and symptoms due to mass effect depending upon the size of the tumor. Though benign GIST outnumber malignant GIST by the ratio of 10 : 1, the malignant potential of benign tumors ranges from 3% - 38%.^[7] GISTs are usually

between 0.5 cm and 8 cm with a range from few mm to larger than 30 cm.^[8] In our case, the size of primary tumor was 20 cm, in its largest dimension. Metastasis of GIST to mandible is very rare. In literature, Gil-Arnaiz *et al.*^[9] reported a tumor in skull that was a distant metastasis from primary GIST of the pelvic region. Another report indicated the mandibular metastasis of a GIST.^[10] Our case clearly revealed mandibular metastasis. Reinhard E. Friedrich *et al.* published recent data on GIST showing metastasis to oral cavity.^[11] The tumor was immobilized in the soft tissues of the cheek, protruding into the oral cavity.

4. CONCLUSION

The aim of this case was to focus on the rare young age presentation of a large intra abdominal mass of GIST. Presentation at the age of twenty five is very rare for malignant GIST. The case highlights diagnostic difficulties and unusual metastasis. Distant metastasis to mandible are rare findings in GIST and indicate end stage of the disease. In addition, it highlights the significance of FNAC in the diagnosis of GIST.

CONFLICTS OF INTEREST DISCLOSURE

The authors declare no conflicts of interest.

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