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A RARE CASE OF ANTERIOR ABDOMINAL WALL EXTRA-GASTROINTESTINAL STROMAL TUMOUR (EGIST)

Oncology

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ABSTRACT

Introduction: GIST that arises primarily outside the GI tract is rare, and is termed as Extra Gastro Intestinal Stromal Tumor (EGIST). CD117 positivity is invariably found and is a hallmark of GIST. EGIST in anterior abdominal wall are extremely rare and very few cases had been reported in past. Majority of EGIST reported in past were actually GIST with metastasis to an Extra GI site. But present case shows origin of tumor over abdominal wall.

Case report: We report a 65 year old male with EGIST in anterior abdominal wall. A well defined heterogenous soft tissue mass measuring 1.0 x 2.0 x 2.2cm was found over left anterolateral abdominal wall. Fat plane with skin, left rectus muscle, and underlying bowel loop was maintained but were lost with External oblique muscle. Excisional biopsy of the lesion shown a circumscribed mass having spindle cell pattern with focal fascicular pattern and with 30-40 mitosis per 50 High power field (HPF). Immunohistochemistry shows strong cytoplasmic positivity for CD117, and also for smooth muscle antigen (SMA). Classifying this case in high risk category, Imatinib mesylate 400mg once daily was prescribed as adjuvant treatment.

KEYWORDS

GIST; CD117, Imatinib Mesylate; Abdominal wall mass

BACKGROUND

Gastrointestinal Stromal Tumour (GIST) is the most common tumour arising from mesenchymal tissue of gastrointestinal tract (GI). GISTs most commonly arise from the stomach, followed by jejunum and ileum, colon, rectum and oesophagus. However, GIST that arises primarily outside the gastro Intestinal tract is rare, and was termed as Extra Gastro Intestinal Stromal Tumor (EGIST) by Reith et al. (1)

EGIST arising in abdominal wall are rare and few cases has been reported in past. (2-4). Herein, we report a denovo case of EGIST in the anterior abdominal wall in a 65 yr old patient.

CASE REPORT:

A 65 year old male developed a small firm nodular mass of 3x3 cm^2 over left hypochondrium since 3 month duration. The mass was non tender, fixed and non-pulsatile on examination. Contrast enhanced computed tomography (CECT) whole abdomen and pelvis interpreted a well-defined, heterogenous soft tissue mass of approximate size 1.0x2.0x2.2cm^3in left anterolateral abdominal wall. Fat plane with skin, left rectus muscle, and underlying bowel loop was maintained. However, there was suspicious loss of fat plane with External oblique muscle [Fig-1]. Also, no evidence of metastasis in other intrabdominal organ, viscera or pelvis was noted. Routine blood examination was unremarkable. Chest X ray showed no lung parenchymal lesion. Differential diagnosis of lipoma was made. Patient underwent excisional biopsy of the lesion. Tab Imatinib mesylate 400mg once daily (OD) was prescribed as adjuvant treatment and patient is on regular follow up and is doing well.

Histopathological examination: Excisional biopsy specimen on gross examination showed, a fairly circumscribed, grey white mass measuring 2.2 x 2.4 x 3.2 cm^3, with focal area of congestion. All margins were free. Microscopy revealed spindle cell tumor with focal fascicular pattern [Fig-2]. The cells had elongated nuclei with cytoplasmic vacuoles. Also, 30-40 mitosis per 50 High power field (HPF) was seen [Fig-3].

Immunohistochemistry (IHC)

The tumor shows strong cytoplasmic positivity for CD117, and also positive stain for smooth muscle antigen (SMA) [Fig-4]. Therefore, pathological diagnosis of EGIST was made.

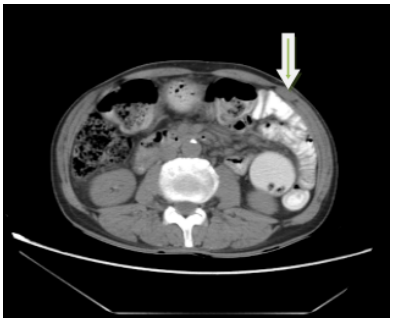

Fig-1: A 1.0 x 2.0 x 2.2 cm3 mass (arrow) in left anterolateral abdominal wall with loss of fat plane with external oblique muscle.

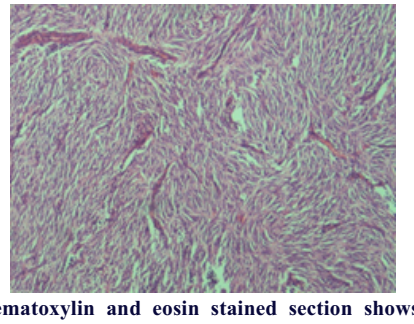

Fig-2: Hematoxylin and eosin stained section shows spindle shaped cells with fascicular pattern

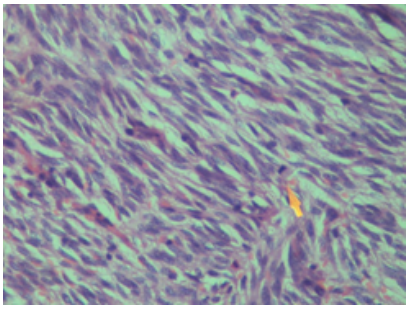

Fig-3: Individual cells (arrow) and high mitosis >30/50HPF

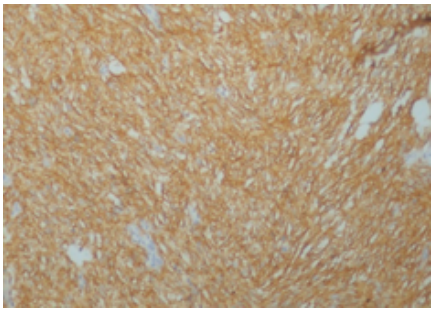

Fig-4: Immunohistochemistry evaluation Positive for CD117 (40X)

DISCUSSION:

GISTs are mesenchymal tumours usually arising from the gut wall. The presumed cellular origin of GIST is from Interstitial Cell of Cajal (ICC) which is the pacemaker cells, intercalated between nerve and muscle fibres in GI tract. (5) It forms a network that is widely distributed within mucosal, submucosal and intramuscular layers from esophagus to internal anal sphincter of gastrointestinal system. (6)

EGIST are the GISTs which occur outside the GI system and has similar histological morphology and immunophenotype to their GI counterpart. However, cell of origin of EGIST is unknown. (7) Pathophysiology of occurrences of EGISTs is the gain of function mutation in KIT-protooncogene, located on chromosome 4q11-12, which encodes for a transmembrane KIT- protein. (8) Cases which does not have KIT mutation are found to have another class II receptor tyrosine kinase gene, platelet derived growth factor receptor alpha (PDGFR), a receptor tyrosine kinase protein as reported by Yamamoto H. et al. (9)

GIST expresses c-KIT protein tyrosine kinase (CD117) and shows an increase in function mutation of C-Kit gene (8). EGIST similar to GIST are invariably positive for CD117 antibody (KIT protein) but also, 50-62% shows positivity for CD34, 26-31% are positive for smooth muscle antigen (SMA), 4-5% are positive for S-100, and few <5% shows Desmin positivity (1,9). The present case also showed positivity for CD117 and SMA.

Major histological variants of EGISTs are spindle cell morphology, epithelial cell morphology and few cases have mixed pattern. Spindle cell morphology and epithelial cell morphology are found in equal incidences in EGISTs. (9) Also, studies had confirmed that histological variant *per se* has no prognostic significance. (1,9) The present case also demonstrated spindle cell morphology with focal fascicular pattern. Reported cases of EGIST in abdominal wall by Kumar AS et al. were spindle cell (4) or epitheloid cell histology as reported by L Alkhatiba et al. (2)

Agaimy et al. carried a detail study on 200 neoplasm consisting GIST and EGISTs and concluded that EGISTs has incidence rate lower than <1.5% , and most of the apparent EGIST would have arisen from GI tract and may have lost contact with their origin due to extensive extramural growth. However, in present case fat plane between bowel loop and tumor is well maintained suggesting de novo origin of tumor in abdominal wall. (7)

Behaviour of stromal tumor depends upon the location, size, and is more aggressive as proceeds from proximal to distal location in GI tract. Data classifying prognostic factors for EGISTs are limited because of its rare occurrences. However, Miettinen M, Lasota (10) suggested prognosis of EGIST akin to that of GIST of distal gastrointestinal tract.

Reith et al. analysed biological behaviour of 48 cases of EGISTs, to find an association of histological features with outcome. After a median follow up 24 months, 39% patients developed metastasis or died, which shows aggressive behaviour of these tumors. High cellularity, high mitotic count (>2/50 HPF) or presence of necrosis were shown to be high risk factor in additive fashion for adverse outcome. Neither size nor the histological patterns had shown any prognostic significance. The reasons were, EGISTs are generally larger (>10cm) at presentation and outcome of small (<5cm) tumors are difficult to interpret because of limited data. (1)

The National Institute of Health (NIH) developed a new classification for risk of malignant behaviour of GIST in 2001. GIST is classified as low to high risk depending upon either tumour size and/or mitotic count. Tumor with any size with mitotic count >10/50 HPF are classified as high risk tumor. (11)

Standard guideline for management of EGIST has not yet evolved. So, they are managed as similar to GIST. In majority of localised cases Surgery as complete tumor removal with clear resection margin followed by adjuvant chemotherapy depending upon risk category is generally followed. (12) Imatinib mesylate is the drug that has shown a significant response in GIST harbouring KIT mutation. (13) However, optimal duration of therapy is still debatable.

The present case had mitotic count of 30-40/50 HPF, thus places it into the category of high risk tumour. Based on earlier study, Tab. Imatinib mesylate 400mg OD for 1 year was prescribed as adjuvant chemotherapy. We decided to keep the treatment duration for 1 year because present data is still not robust regarding the optimal duration of therapy and so would review for further extension depending upon patient tolerance. Patient is on regular follow up and is disease free on clinical and radiological examination.

CONCLUSIONS:

Existing data is insufficient to study the pathogenesis and management of EGIST. EGIST though rare, should be considered in the differential diagnosis of mesenchymal tumour which arises in abdominal wall. Disease risk stratification as per the consensus guideline for GISTs can be followed for EGISTs, with surgery and a long term follow up in low risk cases and a target therapy in high risk cases.

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Ethical clearance: Written and informed consent was obtained from patient for publication of case and images.

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