ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 09 | September - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

MYXOID EPITHELIOID EXTRAGASTROINTESTINAL STROMAL TUMOUR PRESENTING AS UMBILICAL HERNIA – A CASE REPORT

Oncology

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ABSTRACT

Extragastrointestinal stromal tumours (EGIST) are mesenchymal tumours with histological and immunohistochemical features similar to Gastointestinal stromal tumours (GIST). The most common site of EGIST is mesentry and omentum. These tumours can present as vague abdominal discomfort to surgical emergencies and can have variable morphological features and prognosis. The diagnosis is confirmed by immunohistochemistry for CD117 and DOG 1. A minority of cases which are negative for them may show a PDGFRA mutation. We herein present a case report of a 79 year old lady who underwent hernioplasty for umbilical hernia, the content of which showed EGIST with myxoid and epithelioid morphology. This case is presented in view of its rare clinical presentation and unusual morphology.

KEYWORDS

Cd117, Extragastrointestinal stromal tumours (EGIST), umbilical hernia

INTRODUCTION

Gastrointestinal stromal tumours (GIST) represent the majority of mesenchymal neoplasms of the gastrointestinal tract. Extra gastrointestinal stromal tumours (EGISTs), which are neoplasms with histological and immunohistological features overlapping with those of GISTs, are found in the abdomen outside of the gastrointestinal tract with no connection to the gastric or intestinal wall[1]. EGIST is a type of soft tissue tumour with variable presentation and prognosis. GISTS whether intestinal or EGIST, have a broad spectrum of morphological variants with distinct prognosis. We present here a case of a 79 year old lady who underwent umbilical hernia reduction with the excised content showing features morphologically consistent with EGIST.

Case Report

A 79 year old female presented with complaints of swelling in the umbilical region associated with pain and gradual increase in size for one year. Local examination showed an irreducible 3x2 cm swelling in the umbilicus. USG revealed narrow umbilical hernia with inflammatory changes. She underwent hernioplasty with excision of umbilical mass which intraoperatively was suggestive of a metastatic deposit. With a provisional diagnosis of myxoid spindle cell neoplasm, the specimen, slides and blocks were sent to our centre for further analysis. Gross examination of the mass revealed a 5x 4x 4cm nodular tissue with haemorrhagic and slimy cut surface. Histopathological evaluation showed a spindle and epithelioid cell neoplasm showing clear cell morphology, myxoid areas, thin vascular channels, areas of hyalinisation and haemorrhage.

The initial panel of IHC done showed vimentin, CD34 and CD117 positivity while all other markers including CK7, EMA, SMA,HMB 45, Melan A, S100, WT1 and CD 10 were negative. We proceeded with DOG 1 which also turned out to be positive. Ki 67 was 6 – 8%. Correlating histopatholoigcal and IHC features, a diagnosis of extraintestinal GIST, intermediate risk was made.

DISCUSSION

EGIST was first described in 1999, by Miettinen et al[2]. The occurrence of EGIST is extremely rare and little is known about its actual origin[3]. The main distinction between GIST and EGIST is the site of origin of the primary tumor. While GIST occurs throughout the GI tract, from the esophagus to the anus, EGIST is a tumor without any connection with the intestinal wall and are reported in the retroperitoneum, mesentery, and omentum, paravaginal tissue, periprostatic tissue and in the pleura[3].

Grossly they range in size from 1mm to more than 20cm with tan and fleshy cut surface which often show haemorrhage and cystic change. GISTS have a broad spectrum of morphological variants with distinct prognosis. Cases with epithelioid or myxoid histology have higher chances of being malignant with increased 5 year recurrence risk. These cases also frequently show PDGFRA mutation in contrast to our case which was ckit positive[4].

The malignant potential of EGIST is determined by evaluating the same parameters used in GIST which include tumor size, mitotic rate, and the presence of tumor necrosis. However, whether this approach to the evaluation of GIST can reasonably be applied to EGIST is still unclear, as there is some evidence that patients with EGIST have a lower age of onset, a larger tumor size and poorer prognosis compared with patients with GIST

In diagnosing GIST composed of epithelioid tumor cells, it is necessary to rule out adenocarcinomas, epithelioid leiomyomas, epithelioid type PEComas, glomus tumour, carcinoid and melanomas[5].

Myxoid morphology in GIST is rare and is therefore essential to rule out other soft tissue myxoid lesions like myxoma, benign peripheral nerve sheath tumor, including neurofibroma and schwannoma, myxoid liposarcoma, myxoid leiomyosarcoma, myxoid chondrosarcoma, ossifying fibromyxoid tumor, and myxofibrosarcoma[5].

Majority of GIST are known to express CD117(ckit) with <5% associated with PDGFRA mutation. The chloride channel protein ANO1/DOG 1 is equally sensitive and specific for GIST as CD 117.

Other markers expressed in GIST include CD 34, with a minority showing positivity for SMA, S100, CK 18, h caldesmon and desmin[6].

The findings of previously published studies on EGIST suggest that these tumors are histologically and immunohistochemically similar to gastrointestinal stromal tumors (GIST), including the expression of *c-KIT and PDGFRA gene* mutations. Omental EGIST often show similar histological features as gastric GIST which further supports the theory of common cell of origin for GIST and EGIST. Omental EGIST, mesenteric EGIST, and gastric GIST have been shown to have a better prognosis compared with intestinal GIST as per previously reported studies.

Review of the current published literature on EGIST has shown that the most common anatomic location of the tumor was in the intra-abdominal cavity, including the mesentery or omentum in 78.6% followed by retroperitoneum in 10.7%[7].While GIST presenting as inguinal hernia is rarely reported[8], EGIST with epithelioid and myxoid morphology, presenting as content of umbilical hernia, is the first reported case in literature to the best of our knowledge.

Myxoid epithelioid GIST although a rare entity, should therefore be considered in the differentials of intrabdominal mesenchymal neoplasms.

Financial Support And Sponsorship

Nil

Conflicts Of Interest

There are no conflicts of interest.

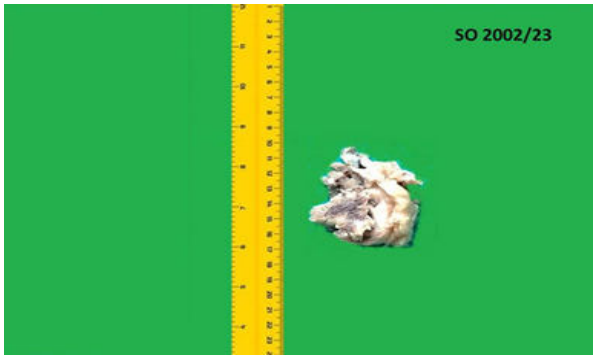

Fig 1: Gross specimen showing nodular mass measuring 5x4x4cm with haemorrhagic and slimy cut surface.

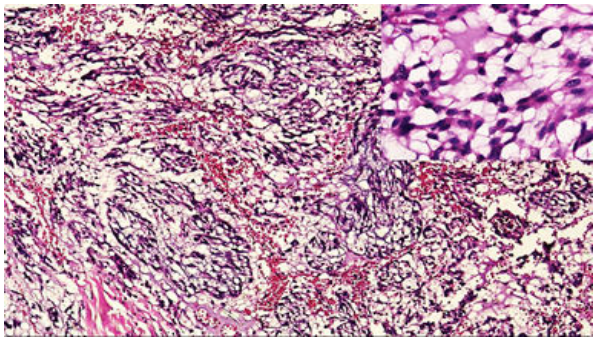

Fig 2: Photomicrograph showing spindle and epithelioid cell neoplasm with myxoid areas, areas of haemorrhage and hyalinisation (100x). Inset shows epithelioid cells with clear cell morphology (400x).

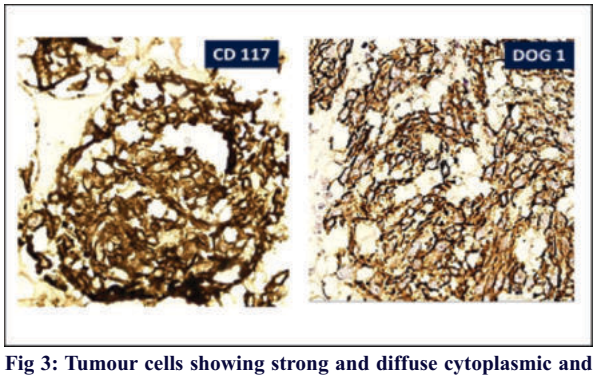

Fig 3: Tumour cells showing strong and diffuse cytoplasmic and membranous positivity for CD117 and DOG 1 (400x).

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