ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 08 | August - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

JEJUNAL LEIOMYOMA VIS-À-VIS JEJUNAL GIST – RARE MESENCHYMAL MIMICS & ROLE OF IMMUNOHISTOCHEMISTRY (IHC) IN PROPER DIAGNOSIS : A CASE REPORT

General Surgery

Dr. Gaurab Das Mbbs, Ms General Surgery , Assistant Professor , Department Of Surgery , Iq City Medical College And Hospital, Durgapur, West Bengal, India.

Dr. Moumita Das Mbbs , Academic Junior Resident , Department Of Surgery , Bankura Sammilani Medical College, West Bengal, India

ABSTRACT

Mesenchymal tumours of GI tract represent a heterogenous group of neoplasms encompassing benign , intermediate, and malignant entities. In consideration of their rarity as well as of intrinsic complexity, diagnostic accuracy represents a major challenge. Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors in the gastrointestinal (GI) tract. GISTs account for approximately 80% of the clinically relevant GI mesenchymal tumors. They are basically positive for KIT by immunohistochemistry. Almost all GISTs are negative for desmin, which is a positive marker for mature smooth muscle cells. Smooth muscle tumors such as leiomyomas and leiomyosarcomas, which usually show the spindle cell morphology, consist of approximately 10% of the clinically relevant GI mesenchymal tumors and are almost positive for desmin and negative for KIT. Thus, most GI mesenchymal tumors are differentially diagnosed by immunohistochemistry (IHC) of KIT, desmin.

KEYWORDS

Mesenchymal tumours ; GIST; Leiomyoma ; Gastrointestinal stromal tumour.

INTRODUCTION :

Gastrointestinal (GI) mesenchymal tumors are rare entities . Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal tumours in the stomach as well as in the small bowel[4]. Approximately 60% of them arise in the stomach and approximately 30% in the small intestine. Duodenal and rectal GISTs account for about 5% each among whole GIST cases, and esophageal GISTs are rarer and colonic GISTs are extremely unusual. GISTs are considered to be derived from interstitial cells of Cajal (ICCs). GISTs are typically composed of spindle cells. Most GISTs diffusely and strongly express KIT. That's why tyrosine kinase inhibitors such as imatinib has a major role in its treatment[2].

Leiomyoma is a rare benign mesenchymal tumour showing smooth muscle differentiation and represents approximately one third all mesenchymal neoplasms of the GI tract. They predominantly occur in the oesophagus, colon, and rectum and rarely arise in the stomach and small intestine. They be confused with GISTs, and IHC in this differential diagnosis can be quite helpful. Leiomyomas are invariably strongly positive for desmin, but are negative for CD117 & the reverse is true for GIST.

Here , we have discussed about a patient who came to our OPD with provisional diagnosis of jejunal GIST but afterwards IHC was suggestive of a different pathology altogether. Informed consent of the patient was taken.

The aim of the reporting of this case is to describe the two most common mesenchymal tumours occurring in the GI tract, focusing on the application of immunohistochemistry to improve diagnostic accuracy and consequently promote the most appropriate therapeutic approach.

Case Report :

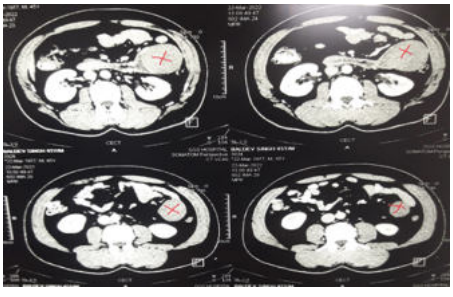
A 45 yr old gentleman presented in our OPD with history of feeling of a lump in left upper part of abdomen with mild pain for last 6 months. He had no history of nausea , vomiting , abdominal distension , alteration of bowel habit , jaundice , loss of weight, loss of appetite , fever , haematemesis , hematochezia. On general examination, his vital signs were stable. On local examination, a non-tender abdominal lump was palpable in deep palpation on left hypochondrium. No other organomegaly was present.

He was investigated further with CECT whole abdomen which revealed a 12 x 8 cm homogeneous mass which is exophytic in nature arising from proximal jejunum suggestive of GIST. There was no gross attachment with surrounding organs and no features of any distant metastasis was present. He was planned for surgery after thorough optimisation.

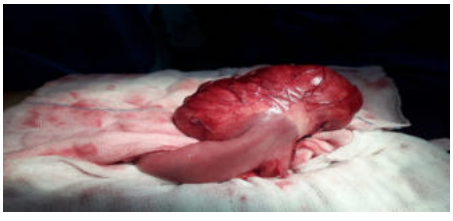
During surgery , mass with above mentioned features was revealed which was very close to ligament of Treitz . Segmental resection and anastomosis was done after taking 1.5 cm margin all around the growth. Post-operative course was uneventful.

The histopathological examination report revealed well circumscribed spindle cell variant GIST with moderate risk of disease progression with mitosis less than 5/ 50 with absent necrosis or hemorrhage.

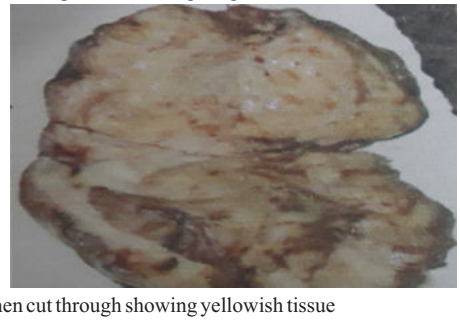
IHC was done next which revealed the the growth was CD117 , DOG1, CD34 , S100 negative which is very much unlike of GIST. But it was positive for SMA and Desmin consistent with Leiomyoma.



CECT showing the growth(marked with a cross)



Intraoperative picture showing the growth



Specimen cut through showing yellowish tissue

DISCUSSION :

Mesenchymal tumours represent an extremely heterogeneous group of lesions. GISTs, leiomyomas are main GI mesenchymal tumors. Since we have expensive but very effective drugs such as imatinib for GIST

treatment, incorrect diagnosis of GIST in non-GIST cases give disadvantage to patients. For correct diagnosis of GISTs, IHC for differential diagnoses are necessary.

In our patient, although histopathological examination was suggestive of GIST, IHC indicated otherwise due to the presence of desmin in these cells. The spindle shaped GISTs share characteristic histological findings with the leiomyomas, which is why it is important to differentiate between the two using immunohistochemical techniques for proper prognostication and treatment regimens[5].

Where a large number of cases of misdiagnosis of leiomyomas as GISTs have been reported earlier in the literature, these tumors arising from the jejunum have been next to none. *Ye et al* in their study reviewed 46 esophageal tumors and suggested that there was presence of both spindle-shaped mast cells as well as hyperplasia of interstitial cells of Cajal in leiomyomas, which could be falsely diagnosed as GISTs[6]. *Hallin et al* reported a lesion of the gastroesophageal junction which showed the presence of spindle shaped cells with interspersed cells of Cajal with a strong expression of CD117 and DOG1 in a large number of cells. They indicated the importance of differentiating a leiomyoma from a GIST, as misdiagnosis could result in incorrect treatment of the lesion diagnosed[7] . A similar case was reported by *Zainudin et al* where a 30 year old male patient had a 10 x 10 cm mass in the terminal ileum which was initially suspected as a GIST but on proper investigation, was diagnosed as a leiomyoma [8].

The case report strongly emphasizes on importance of differentiation between a GIST and a leiomyoma through the necessary investigations, as it would determine the patient's line of treatment as well as the overall prognosis of the condition. We also believe that it would add to the scarce literature that is available on the jejunal leiomyomas that present as GISTs histologically and morphologically.

REFERENCES :

[1] Gheorghe, M., Predescu, D., Iosif, C., Ardeleanu, C., Băcanu, F., & Constantinoiu, S. (2014). Clinical and therapeutic considerations of GIST. *Journal of medicine and life*, *7*(2), 139–149.

[2] Akahoshi, K., Oya, M., Koga, T., & Shiratsuchi, Y. (2018). Current clinical management of gastrointestinal stromal tumor. *World journal of gastroenterology*, *24*(26), 2806–2817.

[3] Wrońśki K, Kaczor J, Masłowski Z. Duodenalis leiomyoma – egy ritka betegség esettanulmánya [Duodenal leiomyoma - a rare case report]. Magy Seb. 2017 Sep;70(3):232-234. Hungarian. doi: 10.1556/1046.70.2017.3.3. PMID: 28876117.

[4] Kang HC, Menias CO, Gaballah AH, Shroff S, Taggart MW, Garg N, Elsayes KM. Beyond the GIST: mesenchymal tumors of the stomach. Radiographics. 2013 Oct;33(6):1673-90. doi: 10.1148/rg.336135507. PMID: 24108557; PMCID: PMC3794320.

[5] Hirota S. Differential diagnosis of gastrointestinal stromal tumor by histopathology and immunohistochemistry. Transl Gastroenterol Hepatol. 2018 May 12;3:27. doi: 10.21037/tgh.2018.04.01. PMID: 29971258; PMCID: PMC6002266.

[6] Ye JX, Liu Y, Qin Y, Ma XL, Zhong HH, Zhang Y, Shi XY. Mast cells or not? - CD117 positive cells in esophageal leiomyoma. Histol Histopathol. 2015 May;30(5):581-8. doi: 10.14670/HH-30.581. Epub 2014 Dec 9. PMID: 25487836.

[7] Hallin M, Mudan S, Thway K. Interstitial Cells of Cajal in Deep Esophageal Leiomyoma. Int J Surg Pathol. 2017 Feb;25(1):51-53. doi: 10.1177/1066896916660197. Epub 2016 Jul 28. PMID: 27422468.

[8] Zainudin S, Rajanthran SK, Azizan N, Hayati F, Ginawoi J, Suhaimi KA, Voo WYL, Kiram MF, Md Jaki MI, Abd Aziz SH, Nacathiran STN. An oncological curiosity of a male patient with a huge leiomyoma of the terminal ileum. Oxf Med Case Reports. 2020 Oct 23;2020(10):omaa086. doi: 10.1093/omcr/omaa086. PMID: 33133619; PMCID: PMC7583423.

[9] S. C. Abraham, "Distinguishing gastrointestinal stromal tumors from their mimics: an update," Advances in Anatomic Pathology, vol. 14, no. 3, pp. 178–188, 2007.

[10] Sbaraglia M, Bellan E, Dei Tos AP. The 2020 WHO Classification of Soft Tissue Tumours: news and perspectives. Pathologica 2021;113:70-84.