## Case Report

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# Unusual occurrence of two sister tumours in the same patientgastrointestinal stromal tumour and gastrointestinal autonomic nerve tumour

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#### **ABSTRACT**

Gastrointestinal stromal tumours (GIST) account for less than 1% of gastrointestinal tumours, whereas Gastrointestinal autonomic nerve tumour (GANT) is a rare variant of gastrointestinal stromal tumour (GIST) and occurs at an estimated frequency of 1% of all malignant gastrointestinal tumours. We report the case of a 38-year old male which represents the rarity in the incidence of a Gastrointestinal Stromal tumour as well as the occurrence of a gastrointestinal autonomic nerve tumour together in the same patient which presented as a jejunal lump along with a terminal ileocecal intussusception. Multiple GISTs are associated with familial or hereditary syndromes are described only in few case reports whereas multiple sporadic GISTs have been rarely reported. This review aims to report the multiple sporadic occurrence of two types of GISTs as well as discuss highlight the features and prognosis of GANTs as surgeons and gastroenterologists are unaware of the existence of such an entity.

**Keywords:** Gastrointestinal stromal tumour, Gastrointestinal autonomic nerve tumour, Ileo caecal intussusception, Multiple occurrence of GIST

#### INTRODUCTION

Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal tract. GISTs are usually found in the stomach followed by jejunum and then ileum. They derived from transformed neoplastic precursors of Cajal's interstitial cell.<sup>1</sup>

While, GANT is a rare variant of gastrointestinal stromal tumour (GIST) and occurs at an estimated frequency of 1% of all malignant gastrointestinal tumours. They are histogenetically related to the gastrointestinal autonomic nervous plexuses and sometimes considered as a rare subtype of the gastrointestinal stromal tumours (GIST). They can however be distinguished based on their unique ultrastructural properties.<sup>1</sup>

Generally, GISTs are solitary tumours and their occurrence as multiple primary tumours is an exceptional event, this type of occurrence is found in familial GISTs or distinct paediatrics syndromes such as neurofibromatosis type 1 (NF1) or Carney's syndrome.<sup>2</sup>

But here we report a rare case with an incidence of multiple types of gastrointestinal stromal tumours in a 38-year-old male who presented with a lump in the abdomen and incidentally on further radiological evaluation a terminal ileo-colic intussusception was noted with the terminal ileum herniating in to the caecum apart from a large mass arising from the level of the jejunum. The patient was treated surgically by resection of both masses found one at the caecum and jejunum and a quarter colectomy was done.

#### **CASE REPORT**

A 38-year-old male had noticed a lump in the lower portion of the abdomen in the last two weeks and presented with complaints of passing black stools intermittently for 8-10 days along with vague complaints of malaise and fatigue. The patient gave no history of pain, fever, nausea or vomiting, loss of weight or appetite. There were no complaints of constipation, loose stools or any urinary complaints associated. He gave a history of having undergone open appendectomy 22 years back. The patient had no co morbidities.

On examination, the patient appeared to be pale. There was a large, solid, smooth and mobile palpable lump noted in the hypogastric region of the abdomen which was around 8\*6cm and non-tender.

Laboratory tests revealed a low Haemoglobin of 4mg/dl and Carcino Embryonic Antigen level (CEA) of 1.2ng/ml.

Rest of the reports were within normal limits. Computed Tomography (CT) of Abdomen and Pelvis reported the presence of a large heterogeneously enhancing mass arising from the small bowel loops in central abdomen. The level of lesion appeared to be in jejunal loops measuring 8\*5\*6cm with No obvious signs of bowel obstruction. Incidentally terminal ileo-colic intussusception noted with terminal ileum herniating in to the caecum. S/O malignant jejunal gist.

On positron emission tomography (PET scan), increased FDG uptake was noted in the heterogeneously enhancing small bowel mass in mid abdomen measuring 8\*5\*6cm (site of primary malignancy). No perilesional nodes noted. Ileo-colic intussusception was noted.



Figure 1: Pet scan image showing the transverse section of the small bowel mass (Jejunal).

Patient was explored with a Midline Incision. On opening the abdomen, a large mass of 8\*6cm was identified in the small bowel. Multiple polyps were studded all over small bowel. The mass was present around 5\*6cm from

duodenojejunal junction. Local surgical resection of the jejunal was performed and sent for histopathology.



Figure 2: Pet scan image showing the sagittal section of the mass.

Another mass was identified at the caecum, excised and sent for histopathology. Distal ileum and proximal colon were resected, and intestinal tract was restored by means of end-to-end anastomosis between the ileum and the ascending colon (Quarter Colectomy) was done. Pelvic drain inserted, and the patient tolerated the procedure well.

Histopathologic report of the jejunal mass revealed features suggestive of Gastrointestinal stromal tumour (GIST) of Uncertain Malignant Potential (due to large tumour size of 10cm and low mitotic activity). While the histopathology report of the ileo-caecal intussusception revealed, benign gastrointestinal stromal tumour (GIST) with neural differentiation (Gastrointestinal Autonomic Nerve Tumour- GANT) of the caecum.



Figure 3: Intra Operative image showing the location of the jejunal mass. Multiple polyps are noted over the small bowel.

#### **DISCUSSION**

Gastrointestinal stromal tumours (GISTs) account for less than 1% of gastrointestinal tumours, but they are the most common mesenchymal neoplasms of the gastrointestinal tract. GISTs are usually found in the stomach followed by jejunum and then ileum. More often malignant in the small bowel than in the stomach. Tumours smaller than 2cms are usually benign, whereas more than 10cms are often malignant. No laboratory test can specifically confirm to rule out GIST.GIST is also not associated with elevation of any serum tumour markers. Imaging studies such as ultrasonography, computed tomography (CT) and positron emission tomography (PET SCAN) can help identity and localise the mass. Although Complete Surgical Resection is the main treatment protocol for GISTs chemotherapy maybe indicated in certain cases. In cases of small GISTs, complete resection is the main stay of treatment. If the GIST is locally invasive, then En Bloc Resection of the involved organ should be done. If the GIST is not resectable or metastatic or both then chemotherapy is indicated. This is also indicated as adjuvant treatment following complete resection. Once histologic findings have been confirmed (study of GISTs by immunohistochemistry reveals expression of CD117 strongly among other antigens), Tyrosine kinase inhibitor (TKI) IMATINIB MESYLATE plays a key role. At a standard dose of 400mg daily then escalate to 800mg daily if tolerated over 1 month. Evaluation of tumour response is done by evaluating the tumour density on CT scan.3

Gastrointestinal autonomic nerve tumour, GANT is a sub type of gastrointestinal stromal tumour (GIST) and has an incidence of only 1% of all malignant gastrointestinal tumours. They are histogenetically related to the gastrointestinal autonomic nervous plexuses however, they can be distinguished based on their unique ultrastructural features. On ultrastructural examination, the cells of Cajal (intestinal pacemaker) show both smooth muscle and neural differentiation, accounting for different variants of GIST viz. smooth muscle GIST (myogenic features), gastrointestinal autonomic nerve tumour (GANT with neural features), mixed GIST (both muscle and nerve), or GIST-not otherwise specified (GIST-NOS lacking differentiation). Multiple GISTs are extremely rare, may be familial. The diagnosis of GANT is usually made on electron microscopy, and immunohistochemical analyses. The tumours were found slightly more often in males. GANTs remain mainly asymptomatic until the tumour reaches a sufficient size to produce abdominal pain or mucosal ulceration with gastrointestinal bleeding. In most of cases, the primary site of the tumour is the stomach, duodenum, jejunum and ileum.

Radiological techniques do not permit a distinction between GANTs and other gastrointestinal stromal tumours. Radical surgical resection appears to be the most promising and solely curative treatment regimen for gastrointestinal autonomic nerve tumours. Conventional chemotherapy and radiotherapy is ineffective in the treatment of GANT. However, for metastatic CD117-positive tumours, tyrosine kinase inhibitors might be an appropriate palliative treatment. Imatinib mesilate, or Glivec (a tyrosine kinase inhibitor), was confirmed to be effective against metastatic or unresectable CD117-positive GANT.<sup>4</sup>

Gastrointestinal stromal tumours (GIST) are commonly regarded as solitary tumours, hence the incidence of multiple lesions is considered unique and is restricted to paediatric GISTs and rare hereditary conditions only. Multiplicity in sporadic GIST patients without family history is uncommon and it was described only in few reports.<sup>2</sup> Earlier, multiple GISTs were categorised as metastatic disease because they were believed to disseminate from one primary GIST. This theory was altered by the reports of authors which described multifocal GISTs in the same patient with well-defined clinical and pathological characteristics are different mutations of KIT and PDGFR which are present in each individual tumour of each patient.<sup>2</sup>

Our case not only reports the unique occurrence of multiple sporadic GISTs but also, it's rare presentation as an ileo caecal intussusception. Histopathological reporting further suggests that one of the GIST presents with neural Differentiation as well thus subsequently resulting in the rare incidence of gastrointestinal autonomic nerve tumour as well.

#### **CONCLUSION**

GIST patients with multifocal presentation are affected by multiple primary tumours. Thus, in the presence of multifocal GIST manifestations, the individual tumours of each different tumour site must be differentiated accurately for a proper staging and planning the right course of further management.<sup>5</sup>

GANTs are rare tumours that arise from the autonomic nerve plexuses. The diagnosis is often made by electron microscopy and immunohistochemical analysis. They present aggressively which correlates with the tumour size, presence of high mitotic figures, degree of necrosis and DNA ploidy. They may present with poor prognosis even after surgical resection due to ineffectiveness of conventional chemo as well as radiotherapy.

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