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Gastrointestinal Stromal Tumour in a Young Adult: A Case Report

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Abstract

Gastrointestinal Stromal Tumour (GIST) is the most common mesenchymal tumour. Peak ages are typically 60-65 years hence very few young adults present with GIST. Here, we present a case of a 26-year-old male with an acute chronic history of melena subsequently diagnose with GIST. No pathology was initially identified upon multiple gastroscopies and a colonoscopy. The patient then underwent Magnetic Resonance Imaging (MRI) Enterography which revealed a 5.6cm soft tissue mass in the right pelvis without any established evidence of bowel invasion. A Positron Emission Tomography (PET) scan did not reveal metastases. Laparoscopy was done and a small bowel resection was performed upon identifying the mass in the bowel. Histopathology analysis showed GIST with a TNM staging of IIIA. The patient was discharged 9 days later with Imatinib to be considered as adjuvant therapy. This case highlights the rarity of GIST seen in young adults presenting with undiagnosed abdominal symptoms as well as peri-operative investigations and surgical management.

Introduction

Gastrointestinal Stromal Tumour (GIST) is the most commonly seen mesenchymal tumour. It is typically observed in patients aged 60-65 years without a specific inclination to any sex. The incidence rate is reported between 11 to 20 per 1 million people with above 50% of these tumors occurring in the stomach [1]. It is challenging to diagnose patients as such tumors can be clinically silent. Here we report a case of a patient diagnosed with GIST after extensive investigations for acute on chronic melaena.

Case description

A 26-year-old male with a medical history of Attention Deficit Hyperactive Disorder (ADHD) presents to the Emergency Department with acute on chronic melaena. The first onset of melaena noticed by the patient was 2 years ago with the most recent episode occuring 2 days prior to presentation. This was associated with intermittent abdominal pain with no other symptoms reported. On the most recent presentation, Haemoglobin (Hb) was 119 g/L, Haematocrit 0.346L/L, Mean Cell Volume (MCV) 86 fl and White Cell Count (WCC) $9.3x10^9$/L. One colonoscopy and three gastroscopies were performed since the patient's first onset of melaena with all of them unable to demonstrate any significant findings. Magnetic Resonance Imaging (MRI) Enterography was done on the most recent presentation, and a 56 x 48 x 34mm well-defined soft tissue mass was noted in the right pelvis which was isointense to muscle on T1 sequences, hyperintense to muscle on T2-FS sequences and demonstrated mild heterogeneous enhancement (Figure 1). It was displacing the adjacent sigmoid colon and small bowel loops without direct involvement although that was not entirely certain. A Positron Emission Tomography (PET) scan was then performed which reported no metastases (Figure 2). Patient was transferred to a tertiary hospital for operative management. He underwent a diagnostic laparoscopy with subsequent small bowel resection with primary anatomosis as the mass was identified to be within the small bowel. Histopathology analy-

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ses demonstrated clear margins of 65mm of small bowel GIST with 2 mitoses per 5mm^2 categorising it's TNM staging to IIIA as per American Joint Committee on Cancer [2]. The patient was discharged 9 days post-operatively however re-presented as he experienced vomiting and significant abdominal pain. Computed Tomography (CT) imaging of the abdomen revealed multiple distended small bowel loops involving the mid and proximal small bowel up to 3.5 cm with decompressed distal small bowel loops and normal-appearing large bowel in keeping with a mechanical small bowel obstruction. Anastomotic dehiscence couldn't be excluded due to lack of oral contrast. He was managed conservatively without any nasogastric tube and recovered well during his admission.

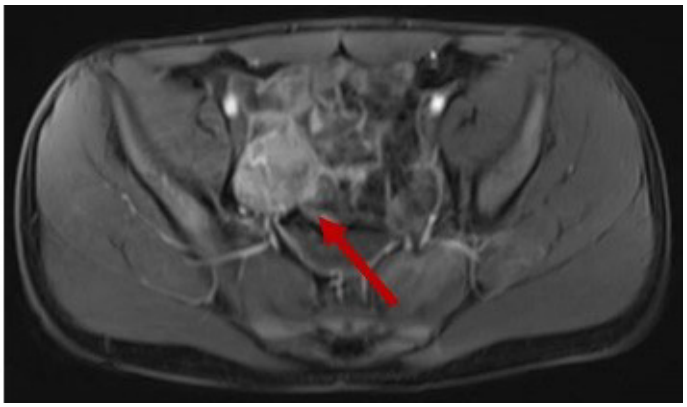

Figure 1: MRI demonstrating a well-defined soft tissue mass on T1 sequence (red arrow).

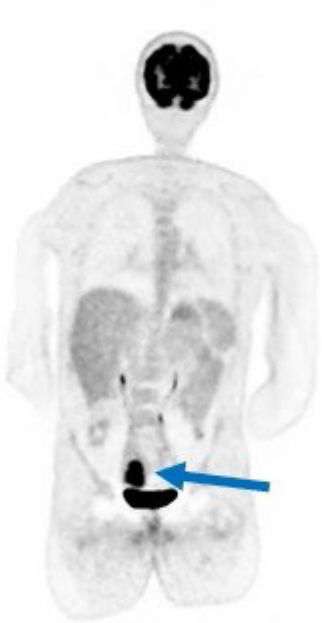

Figure 2: PET scan demonstrating lesion in the right pelvis (blue arrow) with no evidence of metastasis.

Discussion

Once known as Gastrointestinal (GI) smooth muscle tumour, studies utilizing electron microscope observed such tumours were significantly different than a classical smooth muscle tumour. It lacks smooth muscle-specific ultra-structures, antigens as well as Schwan cell features [3-5]. Hence, the term 'gastrointestinal stromal tumor' was then used to reflect the range of features seen in these tumours. It is now established that GIST arises from the pacemaker cells of the intestines i.e. Interstitial cells of Cajal. Mutations of tyrosine kinase receptor KIT and platelet-derived growth factor receptor α (PDGFRA) are identified as having oncogenic properties that leads to GIST [1]. As previously mentioned, at least 50% occur in the stomach; whilst 30% in the jejunum or ileum, 5% in the duodenum and rectum each and less than 1% in the oesophagus [6]. In our case, the tumour was laparoscopically resected from the distal small bowel. Laparoscopy surgery has an increased advantage especially for tumours with ≤5cm as they show exceptional survival rates (92-96%), irrespective of age [7,8]. This also concurrently illustrates the excellent prognosis for GIST that are ≤5cm [9]. Fortunately for our patient, no metastasis was detected giving a good survival prognosis. It was however unanticipated for him to re-present with a small bowel obstruction. Compared to open bowel resections, laparoscopic surgery causes lesser 'Early Small Bowel Obstruction' (ESBO) due to reduced adhesion formation. However, ESBO persists [10]. Non-operative approach with gastric decompression e.g. nasogastric tube insertion is preferred in this context to avoid bowel manipulation which may delay healing [11]. Given the progressive risk of disease for our patient is 24% as per Miettinen *et al*, administrating Imatinib as an adjuvant therapy for 3 years is currently considered in our multidisciplinary team meetings [12,13]. Used for metastatic or non-resectable GIST, Imatinib is now introduced to prevent tumour recurrence despite complete tumour resection [14-16]. Considering our patient's age and its rarity to present with GIST, it highly important that timely follow-ups, symptom monitoring and treatment compliance is embedded into his care. This is to ensure recurrence of disease is identified and managed promptly. In conclusion, GIST in young adults is uncommon but nonetheless have very good prognosis if the condition is recognised and treated early.

Conflict of interest

None declared.

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