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Pictorial Review

Gastrointestinal stromal tumours (GISTs) with a thousand faces: atypical manifestations and causes of misdiagnosis on imaging

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Gastrointestinal stromal tumours (GISTs) can lead to emergency situations, such as gastrointestinal bleeding, intestinal obstruction, and tumoural rupture with haemoperitoneum or peritonitis. In addition, if a GIST grows exophytically to a large size, it is often misdiagnosed as a tumour arising from adjacent organs. Sometimes, the atypical appearance of GISTs on imaging causes diagnostic confusion. In this article, we illustrate a variety of GISTs with atypical presentations and also discuss the important diagnostic clues for differentiating GISTs from other lesions.

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Introduction

Gastrointestinal stromal tumours (GISTs), derived from interstitial cells of Cajal, are the most common mesenchymal tumours found in the gastrointestinal tract. The majority of GISTs express a tyrosine kinase growth factor receptor (c-KIT), which is a key factor for diagnosing GISTs.[1] The most common site of GISTs is the stomach (70%), followed by the small bowel (30%), and anorectum (7%); however, GISTs occur very rarely in the colon and oesophagus.[1,2]

GISTs usually appear as well-defined, enhanced masses with intact overlying mucosa. They can grow in one of the following patterns: endoluminal, exophytic, or dumbbell-shaped. Occasionally, complications associated with GISTs, including gastrointestinal bleeding, intestinal obstruction, rupture with haemoperitoneum, or peritonitis, occur and cause acute abdominal symptoms.[2] In these situations, accurate diagnosis of the underlying cause on imaging is essential for treatment.

In particular, large GISTs may be difficult to differentiate from other gastrointestinal tumours as well as tumours arising from other organs, such as the liver, pancreas, ovary, or uterus, because it is often difficult to identify the organ of origin of large masses. Infrequently, correct preoperative diagnosis of GISTs is not possible due to unusual imaging manifestations.

In this review, we illustrate various atypical manifestations of GISTs including complicated GISTs, GISTs mimicking other tumours, and GISTs with rare presentations. Among the various imaging techniques, magnetic resonance imaging (MRI) and positron-emission tomography (PET) combined with computed tomography (CT) have been reported to be useful for predicting the malignant potential of

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GISTs.[3,4] Therefore, we will focus on the CT imaging findings of GISTs. We also discuss diagnostic clues for correct diagnosis.

Complications of GISTs

Complications such as gastrointestinal bleeding, intestinal obstruction, and tumoural rupture with haemoperitoneum or peritonitis can occur as a result of a GIST.[2,5–7] The majority of cases with complications of GISTs are emergency situations that require accurate preoperative diagnosis and urgent surgery. In a study of GISTs associated with emergency complications (n=92), the most frequent presentation was gastrointestinal bleeding (49%), followed by intestinal obstruction (28%), haemoperitoneum (15%), and rupture with peritonitis (8%).[2]

Gastrointestinal bleeding occurs as a result of tumour ulceration at the mucosal level or intraluminal tumour rupture.[2,5] Clinical presentation varies from anaemia to melaena or haematemesis/haematochezia according to the amount of active bleeding and tumour location in the gastrointestinal tract. Demonstration of intraluminal extravasation of contrast material by CT allows correct diagnosis of a bleeding GIST. Occasionally, it is important to identify small bleeding GISTs admixed with extravasated contrast material (Fig 1); however, it is not always easy because a bleeding tumour with reduced perfusion may mimic an intraluminal haematoma.

Most intestinal obstructions caused by GISTs occur in the small bowel with three patterns.[2,6] First, GISTs as a leading mass induce intussusception with a bowel-within-bowel configuration on CT (Fig 2). Second, GISTs with exophytic and endophytic growth cause direct occlusion of the bowels. In the third pattern, a volvulus-like torsion of the small bowel on the mesenteric root by GISTs, especially exophytic tumours, develops. In a study of GIST-related intestinal obstruction (n=22), the small bowel was obstructed by two patterns: intussusception (n=12) and direct occlusion (n=10).[2]

Although the majority of GIST ruptures occur spontaneously within the gastrointestinal lumen; infrequently, intraperitoneal rupture of GISTs can result in massive haemoperitoneum or peritonitis.[2,7] Features of ruptured GISTs include high-risk pathology, large size (mean, 10 cm), exophytic growth, internal necrosis or cystic degeneration, and interval rapid growth.[7] Intratumoural necrosis, intratumoural bleeding, or bowel ischaemia induced by tumour embolisation is regarded as the mechanism for GIST rupture or bowel perforation.[2] At CT, a ruptured GIST should be considered when necrotic or haemorrhagic portions are seen within the tumour with the presence of ascites or haemoperitoneum, which is rare even in peritoneal metastasis[7] (Fig 3). Panperitonitis and pneumoperitoneum can be a consequence if an extensive necrotising GIST with communication to the bowel lumen ruptures (Fig 4).

Misdiagnosis of GISTs

At CT, misdiagnosis of GISTs as tumours arising from other organs can often occur, particularly in cases

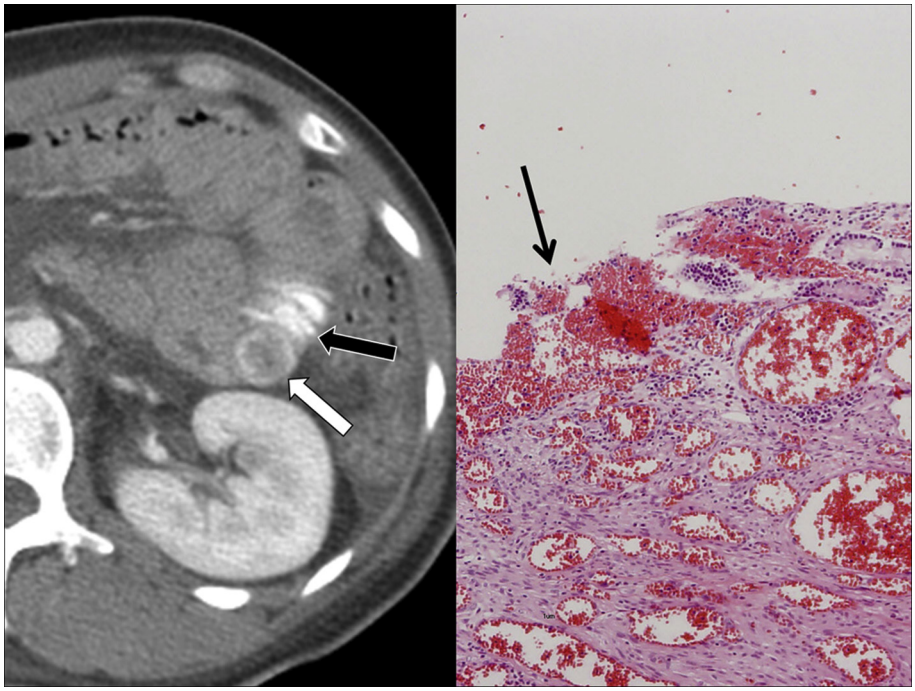

Figure 1 A 39-year-old woman with a GIST in the jejunum. The patient visited the emergency room with melaena, hematemesis, and haematochezia. Enhanced CT (left) reveals extravasation of contrast media (black arrow) and a small soft-tissue density lesion (white arrow) with a peripheral hyperdense rim within the jejunum. This hyperdense rim was presumed to be contrast media around the mass. A very low-risk GIST with mucosal necrosis and bleeding (arrow, right) was confirmed after surgical resection.

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