**Introduction**

Gastro-intestinal stromal tumours (GISTs) usually occur in the stomach (70%) or small intestine (20%) with the oesophagus being a rare site accounting for between 1 and 2% of all cases. Such GISTs are usually asymptomatic and tumour perforation is an uncommon presentation. In the literature there are reported cases of ruptured gastric and small bowel GISTs but a ruptured oesophageal GIST is very rarely heard of. In this report, we review the diagnosis, pathology, and treatment of a patient presenting with a ruptured oesophageal gastro-intestinal stromal tumour.

**Case report**

**History**

A 32-year-old man presented to the Accident and Emergency Department with a 2-week history of left upper back pain in February 2006. After admission he developed chest pain and fever. Physical examination and a urine culture were reportedly normal but blood investigations revealed a markedly raised white blood cell count of 38.6 x 10^9/L. The other blood parameters were normal.

**Initial imaging findings**

An initial chest X-ray showed a hydro-pneumothorax with an air-fluid level in the left hemithorax. The trachea was deviated to the right side. A chest drain was inserted through which purulent fluid was drained.

A provisional diagnosis of empyema was made prior to an urgent CT of the thorax. Oral contrast, 800 mL, was given 2 hours before the CT, which demonstrated a bulky 4 x 4 x 8 cm posterior mediastinal mass at the distal oesophagus, with eccentric wall thickening and irregular air cavitation. After intravenous contrast injection, heterogeneous enhancement of the mass could be seen. A huge air-containing collection, measuring 6 x 9 x 12 cm, was closely related to the mass at the posterior and lateral aspect of the pleural cavity. It exhibited direct communication with the cavity of the aforementioned mass (Fig 1). This, together with the presence of oral contrast in the lateral pleural collection, suggested a pre-existing neoplasm with rupture of the oesophagus.

A subsequent contrast study of the upper gastro-intestinal tract using Gastrograffin confirmed substantial contrast leakage from the distal oesophagus into the posterior mediastinum.

**Surgical management**

A left thoracotomy was performed 2 weeks after admission and the tumour was found to be a GIST. The mass was dissected and removed. Histopathological examination confirmed that it was a GIST.
to be unresectable. The lower thoracic oesophagus had been replaced by a tumour of 10 cm in length extending to the cardio-oesophageal junction. It was connected to the lateral pleural cavity. The patient became haemodynamically unstable during the operation and further tumour dissection was abandoned. An incisional tumour biopsy and a jejunostomy with an open approach were then performed for palliative care.

Pathological evaluation
Microscopic examination of the material removed at surgery demonstrated a tumour composed of epithelioid and spindle cells arranged in cords and sheets in myxoid stroma. The tumour cells exhibited mild pleomorphism without necrosis. Immunohistochemical staining showed that the tumour cells were diffuse and strongly positive for CD117 (c-kit), CD34, while focally positive for desmin (Fig 2). The tumour cells were negative for smooth muscle actin, cytokeratin (Cam 5.2 and AE1/AE3), and S-100 protein. All these features, especially the CD117 positivity, were very suggestive of a GIST with uncertain malignant potential.

Clinical progress
Multiple antibiotics were prescribed to treat the sepsis secondary to the empyema. The patient improved clinically and a trial of imatinib (400 mg daily for 8 weeks on a self-payment basis) was started.

The tumour showed interval regression in
the follow-up CT scan done 3 months later. With the significant shrinkage in tumour size, an elective partial oesophagectomy using a thoracoabdominal approach became possible.

Discussion

The name GIST was originally introduced as a neutral term for tumours that were neither leiomyomas nor schwannomas. This name is now used to describe a specific group of tumours encompassing most gastric and intestinal mesenchymal tumours earlier designated as leiomyomas, leimyoblastomas, and leiomyosarcomas. Schaldenbrand and Appelman first used the term ‘GIST’ in 1984.1

Gastro-intestinal stromal tumours are typically immunohistochemically positive for the KIT tyrosine kinase receptor, which is perhaps their single best defining feature. The c-kit positivity parallels that seen in the interstitial cells of Cajal, the pacemaker cells regulating autonomic activity. It is currently thought that GISTs originate from a precursor cell pool with differentiation towards the Cajal cell phenotype.

This protein, CD117, acts as a specific antigen and constitutes a portion of the KIT enzyme, which is present in most GISTs. Therefore, detection of CD117 with a specific diagnostic test helps to prove that the growth is a GIST. CD117 is also normally present in breast epithelium, germ cells, melanocytes, stem cells, and mast cells.2 Most GISTs (70-80%) occur in the stomach while 20 to 30% occur in the small intestine.3 They are rarely found in the colon/rectum (5%) and the oesophagus (1-2%).

Gastro-intestinal stromal tumours are usually asymptomatic. Symptomatic GISTs can present with a spectrum of symptoms including dysphagia,4 a palpable mass, abdominal pain, or gastro-intestinal (GI) bleeding manifest by haematemesis or melena.5,6 Tumour perforation is an uncommon but not especially rare presenting feature that occurs in up to 20% of cases. Depending on the location of the GIST, it may also rupture into the abdominal cavity resulting in a haemoperitoneum, peritonitis,6 or features mimicking appendicitis7 in the case of a small bowel GIST rupture.8 In our case, the patient presented with upper back pain and a hydro-pneumothorax, features that have not been documented in the English literature on GIST rupture.

Different imaging modalities including double barium contrast study, ultrasound, CT, magnetic resonance imaging (MRI), and positron emission tomography (PET) have been used to assess GIST but CT is used most frequently due to its high sensitivity and availability. Nonetheless, imaging features can never be pathognomonic and diagnostic confirmation requires the aforementioned immunohistochemical techniques.

Plain radiographs usually offer little assistance with the evaluation of a GIST but a double barium contrast study can demonstrate a predominantly intra-mural mass with an extra-phytic component. Other features such as overlying mucosal ulceration leading to a bull’s eye or target lesion appearance are mainly seen in malignant stromal tumours. It has been reported that a double barium contrast study shows abnormalities in 80% of cases,11 which is slightly less than the rates seen with CT (87%).

A main role for CT is localisation of the lesion. It shows the endoluminal and exophytic extent of the tumour. Contrast enhancement may be rim-like and uniform. Larger GISTs with necrosis appear as heterogeneously enhancing masses with irregular central areas of fluid, air or oral contrast and variable wall thickness.

Another important role for CT is recognition of an aggressive tumour. Features that are suggestive of malignancy include (a) heterogeneous contrast enhancement, (b) central cystic/necrotic degeneration, (c) size greater than 5 cm, (d) extension into adjacent organs, and (e) more than one mitosis per 50 high-power field. All these features carry a negative prognostic value. Computed tomography is also sensitive for detection of metastatic liver, peritoneal, lung, and bone lesions. Gastro-intestinal stromal tumour is a likely diagnosis in the presence of a large, complex, intestinal mass with liver lesions and no significant lymphadenopathy.12

Ultrasonography, apart from endoscopic ultrasound, has a minimal role in the evaluation of GISTs. It is generally believed that ultrasonography is only moderately sensitive for detecting GISTs because bowel gas and acoustic shadowing obscure portions of the bowel and mesentery. Nonetheless, when indicated, ultrasonography can be considered for guiding a needle biopsy of a known lesion in selected cases, although there is a risk of tumour seeding and peritoneal spillage. On sonograms, larger GISTs appear as complex masses with cystic and solid components, consistent with their tendency to necrose.

In difficult cases, MRI and PET may also be employed to evaluate GISTs. The former has the best tissue contrast and helps identify masses within the GI tract. In the old days, MRI was believed to be superior to CT because it can localise the mass in different planes. The evolution of multi-detector computed tomography (MDCT) has removed this advantage because reconstructed coronal13/axial images suffice for this purpose. Last but not least, PET scanning has recently been shown to be more sensitive than MRI.

While surgery remains the mainstay of treatment, imatinib, a new cancer drug, is an effective adjuvant therapy for GISTs. It is a c-kit growth factor tyrosine kinase inhibitor, STI571. Two reports of early
clinical trials presented at the 2001 Annual Meeting of the American Society of Clinical Oncology have promising results. 

**Conclusion**

The oesophagus is a rare site for a GIST and tumour rupture is an uncommon clinical presentation. A high level of suspicion can lead to effective surgical treatment and a better clinical outcome. Computed tomography remains the imaging technique most frequently used to assess GISTs because of its high sensitivity and availability. The different reconstructed multi-planar images provided by MDCT enable stromal tumours of the distal oesophagus to be better assessed and localised.

**References**