Rare and unusual metastases of a Gastrointestinal Stromal Tumour

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Abstract

Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal tumours of the gastrointestinal tract, histological and immunohistochemistry findings help to differentiate such tumours from other gastrointestinal malignancies. Metastasis is common to the stomach and small bowel and often presents with gastrointestinal bleeding. This is a case of an 82 year old man with an inguinal mass that following exploratory examination was found on histology to be a GIST metastases, imaging also showed pulmonary metastases. Following colonoscopy the primary caecal mass was found. Such metastatic presentations are extremely rare for this type of tumour. This case report highlights these unusual findings.

Keywords: Gastrointestinal stromal tumours, Inguinal mass, Metastasis, Pulmonary.

Introduction

Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal tumours of the gastrointestinal tract.[1] GISTs can be seen as either spindled, epithelioid cells or as a combination of both of these cell types.[2] They differ from other non-epithelial GI tumours, including leiomyomatous and neurogenic tumours, on the basis of specific histological and immunophenotypic features that permit diagnosis.

They can present anywhere in the GI-tract from the lower oesophagus to the anus. A great majority of GISTs occur in the stomach (60-70%) or small intestine (25-35%).[3] Colon, caecal, rectum, appendix and oesophagus are rarer sites.[3] Gastric and small intestinal GISTs often present with vague symptoms leading to their endoscopic or radiologic detection, but sometimes they cause upper gastrointestinal bleeding.[4] Colorectal GISTs may manifest with lower GI bleeding, colonic perforation, pain, obstruction, or combination thereof.[5] A minority of GISTs, usually malignant tumours, may be externally palpable.

This report describes the case of a caecal GIST which has metastasised to both the inguinal and pulmonary regions.presenting as an isolated inguinal swelling.

Case Report

An 82-year-old gentleman presented with a two-month history of a painless swelling in his right groin. Clinically, it was immobile, non-tender and appeared to be an incarcerated inguinal hernia. It appeared to be fixed at its base and had no cough impulse, as well as lacking any scrotal descent. There was no associated nausea or vomiting, evidence of strangulation or obstruction. The patient reported that the mass seemed to be steadily increasing in size over several months. He denied any constitutional symptoms of weight loss, anorexia and lethargy.

The patient was hypertensive and suffered from hypercholesterolaemia. He did not smoke or drink alcohol and there was no other past medical history of note.

He was subsequently booked for an exploratory examination under anaesthesia (EUA) ± hernial repair/excision. Whilst in theatre the inguinal node was excised and deemed likely to be a malignant mass. A subsequent CT scan showed widespread pulmonary metastases but no obvious soft tissue disease in the abdomen. Colonoscopy revealed a caecal mass which, on biopsy, was not felt to be neoplastic. Immunohistochemistry of the inguinal node yielded positive results for CK20 but negative results for the CK7 marker. It was felt likely that this was likely to be a colorectal primary but macroscopically appearing more like a GIST, pathology confirmed positive CD117 antigens, diagnostic for GIST. C-KIT exon mutation analysis was not carried out, however macroscopically and histologically appeared to be a GIST.

The patient was then commenced on chemotherapeutic management for metastatic GI tumours.

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Discussion

This case report illustrates an unusual presentation of metastatic GIST. Subsequent histopathological assessment identified the tumour as a high grade GIST as per the Bucher grading system.[6] To our knowledge, this is one of the first cases of a GIST with inguinal lymph node and pulmonary metastasis reported in the UK.

Identification of the GIST from caecal origin is in itself rare; however metastases to the inguinal and pulmonary region too are unusual. GISTs usually become symptomatic when they reach large enough diameters with symptoms arising due to bleeding or obstruction. CT and MRI are valid for evaluating both primary tumours and metastatic lesions.

The two most common routes of GIST metastases are intraperitoneal dissemination and haematogenous spread to the liver. GIST metastasis to other visceral organs, lungs, pleura, bones, or brain is rare.[7] Lymph node metastasis is unusual too (1-2%), thus, routine lymphadenectomy is not recommended.[8] Extra abdominal lymphogenic spread is exceptionally rare and appears to also indicate a poorer prognosis.[9]

Surgery, is still the only curative treatment for resectable primary GISTs without evidence of metastasis.[10]

Conclusion

Although caecal GISTs with inguinal node metastases are rare, all groin masses must undergo careful evaluation and not presumed to be hernias. If any doubts arise, a staging CT, endoscopy and subsequent EUA with appropriate treatment by a colorectal surgeon should be carried out.

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