Gastroblastoma with MALAT1-GLI1 fusion gene: a case report

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Case presentation

A 49-year-old woman presented to our hospital with a gastric submucosal mass in March 2024. She was asymptomatic with no abdominal signs, and laboratory tests revealed no significant abnormalities. Abdominal contrast-enhanced computed tomography (CE-CT) identified a 3.2×2.2 cm^2 rounded mass on the greater curvature of the gastric antrum. The mass protruded into and out of the cavity, displaying a clear boundary, slightly uneven density, evident inhomogeneous enhancement, and marked vascularity, indicative of abundant blood supply. Endoscopic ultrasound (EUS) examination revealed a smooth-surfaced spherical bulge originating from the muscularis propria layer. It appeared hypoechoic with areas of hyperechoic signals, protruding into and out of the lumen, measuring 24.5 × 20.3 mm^2 in cross-section (Fig 1).

The tentative diagnosis was gastric stromal tumour. The patient underwent laparoscopy endoscopy cooperative surgery (LECS). During the operation, the tumour was observed to have an irregular shape, with no invasion or adhesion to surrounding organs and no enlarged lymph nodes in the abdominal cavity. The surgical specimen revealed a tan-white solid tumour measuring 3.2×2.5×2 cm^3, with a complete capsule. The margins

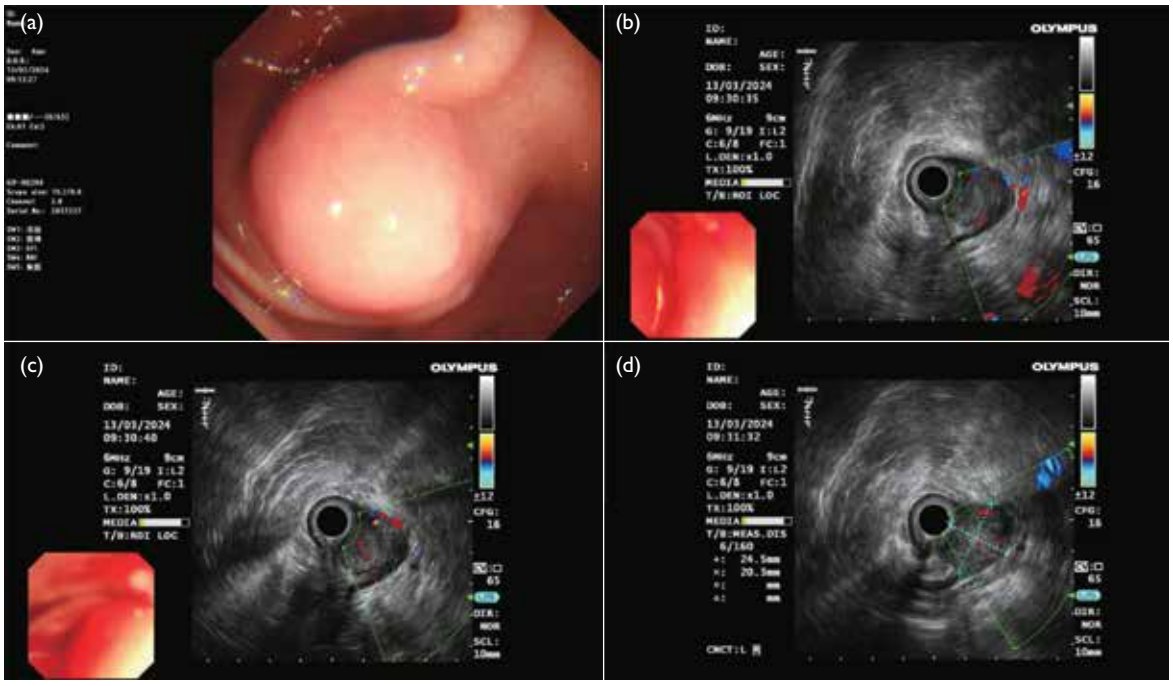

FIG 1. (a) Endoscopic image. (b-d) Endoscopic ultrasound images

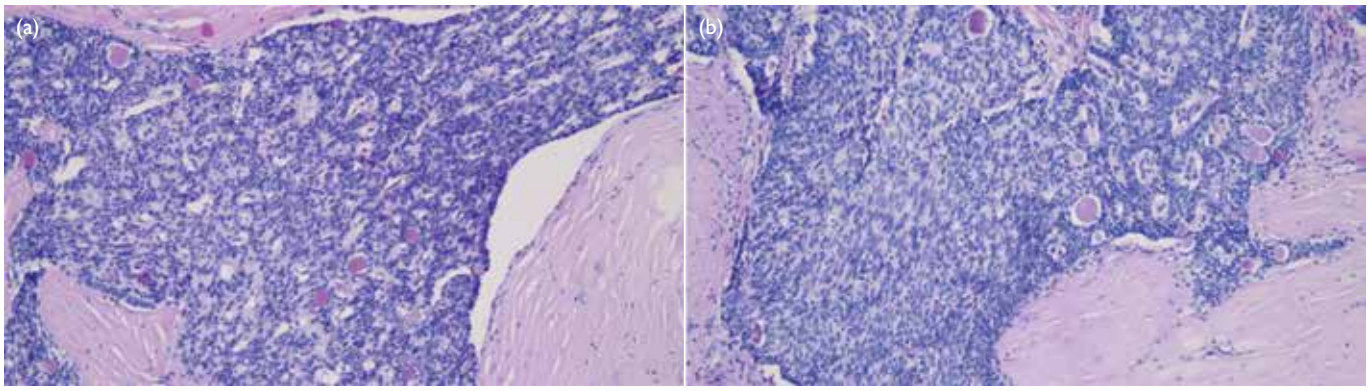

FIG 2. Histological findings (haematoxylin-eosin staining, ×200). (a) Oval cells arranged in nest-bulk clusters. (b) A small number of spindle cells

of the specimen were clean. Postoperatively, routine symptomatic supportive treatments such as fasting, gastrointestinal decompression, anti-infection therapy, acid suppression, and nutritional support were provided. The patient recovered uneventfully.

Histological examination revealed the tumour to be multifocally centred in the muscularis propria, arranged in nests and cribriform patterns, with intraluminal dense eosinophilic material. The cells were oval, with a few fusiform in shape (Fig 2). Immunohistochemistry revealed tumour cells to be positive for CD56, cytokeratin, CD34, CD10, and vimentin. The Ki-67 proliferation index was 3%. We performed whole-transcriptome messenger RNA sequencing of tumour tissue, which revealed a fusion between *MALAT1* (exon 1) and *GLI1* (exon 3). Based on the above evidence, the patient was definitively diagnosed with gastroblastoma.

Discussion

Gastroblastoma is a rare biphasic gastric tumour characterised by its distinctive biphasic epithelial-mesenchymal morphology. Diagnosis from biopsy specimens is typically challenging and often requires additional immunohistochemistry.[1] In 2009, Miettinen et al[1] reported the first case of gastroblastoma. At the time of writing, only 27 cases had been reported.[2]

According to the review by Luo et al,[2] the 27 patients ranged from 5 to 74 years, with a mean age of 35 years. There were 14 male and 13 female patients. Tumour size ranged from 1.3 to 15 cm, with an average of 5.7 cm. Most lesions occurred in the gastric antrum. Clinical manifestations were non-specific, and the tumours primarily involved the muscularis propria. Computed tomography (CT) and EUS were the most commonly used diagnostic methods, typically revealing a mixed solid-cystic mass with heterogeneous hypoechoic areas, occasionally accompanied by ulcers. Two patients underwent preoperative EUS-guided fine-needle aspiration. In our case, the preliminary diagnosis was gastrointestinal stromal tumour based on EUS and CE-CT findings. Endoscopic ultrasound–guided fine-needle aspiration was not performed to avoid damaging the tumour and increasing the risk of metastasis. No clear associated with systemic conditions was observed.

Gastroblastoma rarely expresses markers typically positive in gastrointestinal stromal tumours, solitary fibrous tumours, gastric schwannomas, or mesotheliomas. It also generally lacks expression of markers characteristic of gastric neuroendocrine tumours or leiomyomas.[3] In the 27 reported cases,[2] most tumour cells were positive for vimentin, CD56, CD10, and PCK. The *MALAT1-GLI1* fusion gene was detected in nine patients and is considered a valuable diagnostic marker for this tumour.[2]

Only three cases presented with organ or lymph node metastasis.[2] One patient had peritoneal, liver, and pelvic metastases, as well as bladder adhesion and lymph node metastasis at diagnosis. Following partial gastrectomy, no recurrence or metastasis was observed at 3-month follow-up. Another patient had two lymph node metastases at the splenic hilum prior to surgery. After partial gastrectomy and splenectomy, local recurrence was noted at 6 months, and surgical debulking was performed. Another patient had liver metastasis preoperatively, but no postoperative follow-up information was available. Among the other surgically treated patients, only one experienced local recurrence. These findings suggest that while gastroblastoma may exhibit indolent behaviour, preoperative metastasis or invasion may be associated with a poor prognosis.

Surgery remains the mainstay of treatment. Of the 27 cases,[2] 23 underwent surgical resection and three received endoscopic treatment. The mean tumour size in the surgical group was 5.80 cm,

compared to 1.91 cm in the endoscopic treatment group, suggesting that tumour size influenced the choice of treatment modality.[2] Among the endoscopically treated patients, one underwent endoscopic full-thickness resection, and two underwent endoscopic submucosal dissection.[2] The choice of endoscopic resection generally depends on lesion depth, size, and location. When endoscopic resection is not feasible, laparoscopy endoscopy cooperative surgery may be considered.[4] In our case, laparoscopy endoscopy cooperative surgery was used, offering the benefits of minimally invasive surgery while reducing postoperative complications. In one previous case, a patient with a 15-cm gastroblastoma underwent postoperative radiotherapy and remained disease-free after 14 years of follow-up.[2] The remaining patients received routine postoperative follow-up.[2] Given the potential for recurrence, particularly in cases with preoperative metastasis or invasion, adjuvant radiotherapy or chemotherapy may be considered on an individual basis. The average follow-up duration across reported cases was 31 months.[2] Given its generally indolent nature, annual follow-up is recommended for most patients with gastroblastoma. For those with preoperative metastasis or invasion, more frequent follow-up every 3 to 6 months is advisable. As gastroblastoma remains rare, additional case reports and studies are needed to enhance our understanding of its biological behaviour, diagnosis, and optimal management.

Author contributions

Concept or design: Q Chen.
Acquisition of data: Y Sun, H Shi, X Wu, Y Wang, E Linghu.
Analysis or interpretation of data: Y Sun, H Shi, J Wang, Y Yuan, J Pang, L Wei, S Song.
Drafting of the manuscript: Y Sun.
Critical revision of the manuscript for important intellectual content: Q Chen.

All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of interest

All authors have disclosed no conflicts of interest.

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Ethics approval

This study was approved by the Ethics Committee of Chinese People's Liberation Army General Hospital, China (Ref No.: S2023-188-01). The patient was treated in accordance with the Declaration of Helsinki and provided written informed consent for all treatments, procedures and the publication of this case report.

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