World Journal of Surgical Oncology

CASE REPORT

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Gastroblastoma — a case report and literature review

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Abstract

Objective To report a new case of gastroblastoma and conduct an exhaustive review of the clinical, morphological, immunohistochemical, molecular features, diagnosis, treatment, and prognosis, to enhance understanding of this condition.

Methods We retrospectively analyzed the case of a 50-year-old woman diagnosed with gastroblastoma and conducted a review and summary of relevant literature.

Results To date, 27 cases have been reported, including the present case. The mean patient age at the time of presentation was 35.0 years (range, 5–74 years), and the disease showed no sex predilection. The most common location was the gastric antrum, and the average lesions size was 5.7 cm (range, 1.3–15 cm). Most patients underwent gastrectomy($n=23$), while several underwent ESD($n=2$) or EFTR($n=1$). Fusion genes were identified, including MALAT1–GLI1($n=8$), EWSR1-CTBP1($n=1$), PTCH1:GLI2($n=1$), and ACTB-GLI1($n=1$)Four patients had metastasis and one of them dead of disease. Immunohistochemical (IHC) analysis revealed that pancytokeratin was always positive in epithelioid components, while vimentin and CD10 were always positive in mesenchymal components. CD56 were often positive in both two components.

Conclusion A comprehensive evaluation of clinical and pathological features is crucial for accurate diagnosis. Partial gastrectomy and EFTR could be an appropriate treatment. The risk factors that affect the prognosis need more cases to be clearly defined. We present this exhaustive literature review to increase awareness of gastroblastoma, better characterize the disease, and provide a reference point for gastroblastoma research in the future.

Keywords Gastroblastoma, Stomach tumor, Biphenotypic tumors

Introduction

Gastroblastoma is a rare tumor of the stomach characterized by biphasic morphology with variable proportions of epithelioid and mesenchymal components [1]. In the 2019 World Health Organisation (WHO) classification of tumours of the digestive system, gastroblastoma was included in the 'malignant epithelial tumours' section, with ICD-O code of 1, suggesting that it has low malignant potential [2]. Herein, we present a new case of a 50-year-old woman, and performed an exhaustive review search to summarize the clinical, morphologic, immunohistochemical, molecular features, diagnosis, treatment

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and prognosis in order to deepen the understanding of the gastroblastoma.

Case descrption

A 50-year-old woman exhibited no symptoms such as nausea, vomiting, abdominal pain, hematemesis, melena, fatigue, fever, jaundice, diarrhea, or significant weight loss. The patient underwent a gastroscopy during a health checkup, which revealed a 3 cm sized mass at the greater curvature of the stomach. Physical examination showed no positive signs in the abdomen. Abdominal computed tomography showed a lobulated mass with clear boundary that was 2.5×2 cm in the largest dimension and appeared to protrude from the greater curvature of the stomach (Fig. 1). Imaging suggested a gastric stromal tumor. The initial surgery, laparoscopic partial gastrectomy, was conducted based on this assumption, and no preoperatively biopsy was performed. During the operation the tumor was excised and the site sutured. No invasion of surrounding organs and no enlarged lymph nodes in the abdominal cavity were observed. Given the rarity of gastroblastoma, the pathologist responsible for the diagnosis lacked direct experience in diagnosing. Initially, the presence of atypical glandular epithelial components led to a diagnosis of highly differentiated adenocarcinoma. Laparoscopic radical gastrectomy was performed 15 days later. In this procedure, an additional 2 cm of the gastric wall, adjacent to the previous resection and suture line, was removed and the gastric wall was re-sutured. Additionally, 26 lymph nodes from groups 1, 2, 3, 4, 5, 7, 8, 11, 19, and 20 were cleared during the surgery and pathological reports showed no lymph node metastasis. The patient had a smooth recovery postoperatively, with no postoperative complications.

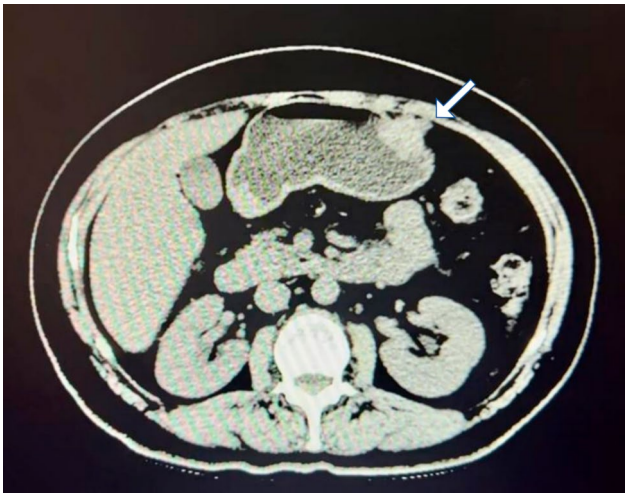

Fig. 1 The preoperative abdominal computed tomographic Images shows a lobulated mass(white arrow) with clear boundary, protruding from the greater curvature of the stomach

On gross inspection, the partial gastrectomy specimen consisted of a submucoal mass, measuring 4.1×2.5×2.0 cm. The cut surfaces varied from yellow-gray to pink-tan.

Histologic examination disclosed a tumor with clear boundaries centered in the muscularis propria, spanning from mucosa into the serosa. The tumor showed biphasic morphology, with well-demarcated epithelial and mesenchymal cells (Fig. 2A) .The predominant component was the epithelial cells, which formed glands or luminal structures containing inspissated eosinophilic material (Fig. 2B). Necrosis could be found in the glands or luminal structures of the tumor. The mesenchymal cells with eosinophilic cytoplasm and ovoid nuclei were distributed around epithelial cells (Fig. 2C). There was no evidence of vascular emboli or perineural tumor invasion. Mitotic figures were rare. The proliferation index (Ki67) in the epithelial and mesenchymal cells was 40% and 3%, respectively.

By immunohistochemistry, the epithelial cells expressed Pancytokeratin (AE1/AE3) (Fig. 2D), CEA, MUC5ac, and Syn. The spindle cells expressed TLE1 (Fig. 2E), Desmin (focally), Syn (focally), CD10 (focally), CD68, and CD163. S100, DOG-1, HER-2, PDGFRA, and SMA were uniformly negative in both tumor cell components.

According to the DNA-based and RNA-based next-generation sequencing (NGS), our case showed low tumor mutation burden (TMB-L) and microsatellite stability (MSS). Neither c-KIT mutation nor fusion gene were detected. Fluorescence in situhybridization (FISH) using an SS18 (SYT) gene probe showed no gene rearrangement (Fig. 2F).

Fourteen months after surgery, no radiotherapy or chemotherapy was performed, and the patient presented with no evidence of tumor recurrence or metastatic disease.

Methods

In February 2024, PubMed was queried using the term “gastroblastoma” and the resultant articles were reviewed. Cases with clear diagnoses and complete information on both histopathology and immunohistochemistry were included in this study.The search was not limited by language and included articles from America, Europe, Africa, and Asia.

Result

A total of 37 articles were found through a PubMed search. These articles and their references were reviewed, and 25 of them reported 26 cases overall. The detailed information of the cases was shown in Table 1. The articles were all published between 2009 and April 2024,

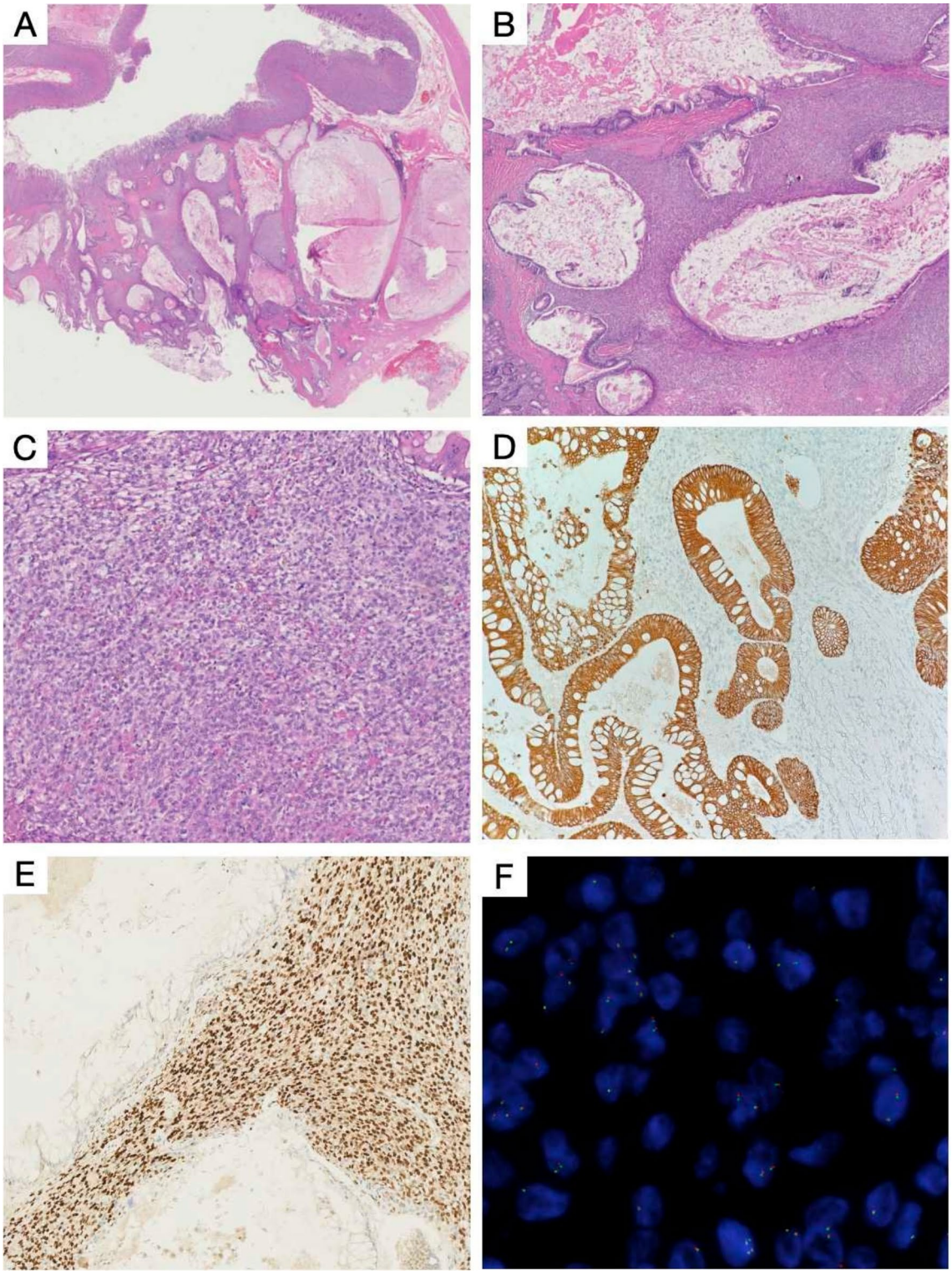

Fig. 2 (**A**) The tumor centered in the muscular layer, spanned from mucosa into the serosa and was well circumscribed. The tumor showed a biphasic morphology, with well-demarcated epithelial and mesenchymal cells. (**B**) The epithelial cells formed glands or luminal structures containing inspissated eosinophilic material. Necrosis could be found in the glands or luminal structures of the tumor. (**C**) The mesenchymal cells with eosinophilic cytoplasm and ovoid nuclei. (**D**) The epithelial component strongly express CK-AE1/ AE3. (**E**) The spindle cells strongly express TLE-1. (**F**) Fluorescence in situ hybridization with an SS18 (SYT) break-apart probe reveals double signals without evidence for a gene rearrangement

Table 1 List of all gastroblastoma cases reported

	Age(y)/Sex	Symptoms	Size (cm)	Location	Layer	Ki-67	Mitoses	Molecular	Treatment	Metastases	Follow-up(mo)
1 [3]	19/M	Nonspecific abdominal pain	5	Greater curvature	TM	N/A	30/50HPF	N/A	Subtotal gastrectomy	No	ANED, 41
2 [3]	27/F	Nonspecific abdominal pain	6	Greater curvature	MP, SubS	N/A	4/50HPF	N/A	Partial gastrectomy	No	ANED, 60
3 [3]	30/M	Anemia, fatigue	15	Gastric antrum	MP, SubS	N/A	1/50HPF	N/A	Antrectomy Postoperative radiation	No	ANED, 168
4 [4]	9/M	Abdominal pain, mass	9	Gastric antrum	MP	N/A	No	MALAT1–GLI1	Antrectomy	No	ANED, 93 [9]
5 [5]	28/M	Constipation	3.8	Gastric antrum	TM	10%	2 /10 HPF	MALAT1–GLI1	Chemotherapy Partial gastrectomy	Liver, LN, retroperitoneal, bladder	AWD, 3
6 [6]	19/F	Abdominal pain, radiating to the back	13	Gastric antrum	MP	N/A	<5/50HPF	N/A	Partial gastrectomy	No	ANED, 20
7 [7]	12/M	Bloody stool, abdominal mass	4.5	Gastric antrum	TM	major: 1% focally: 40%	2/10 HPF	N/A	Subtotal gastrectomy	No	ANED, 8
8 [8]	29/F	Epigastric pain, hematemesis	7	Proximal stomach	TM	N/A	21/10HPF	N/A	Partial gastrectomy Splenectomy Surgical debulking	LN, LR	LR, 6 DOD, 7
9 [9]	27/M	N/A	N/A	Gastric antrum	N/A	N/A	N/A	MALAT1–GLI1	Partial gastrectomy	No	ANED, 12
10 [9]	56/F	N/A	4	N/A	N/A	10%	N/A	MALAT1–GLI1	N/A	Liver	N/A
11 [10]	65/F	No	1.3	Greater curvature	MP	2%	Infrequent	N/A	EFTR	No	ANED, 3
12 [11]	74/M	Weight loss, dysphagia	9	Gastric antrum	NA	N/A	No	MALAT1–GLI1	Partial gastrectomy	LR	ANED, 52 LR, 51
13 [12]	43/F	Intestinal bleeding	5.3	Gastric antrum	MP	N/A	Epithelial: 2/ 20 HPF Spindle cell: 0/ 20 HPF	N/A	Partial gastrectomy	No	ANED, 100
14 [13]	53/F	Dyspepsia	2.27	Greater curvature	MP	<5%	<2/50HPF	N/A	Partial gastrectomy	No	ANED, 18
15 [14, 15]	53/F	Abdominal pain	5.0	Gastric antrum	SubM, MP	<1%	N/A	N/A	Partial gastrectomy	No	ANED, 14
16 [16]	17/M	Hematemesis, melena.	6.3	Fundus	MP	5%	0/ 20 HPF	EWSR1-CTBP1	Partial gastrectomy	No	ANED, 23
17 [17, 18]	58/M	No	2.43	Lesser curvature	SubM, MP	5%	Infrequent	PTCH1::GLI2	ESD	No	ANED, 12
18 [19]	51/F	Black stool, dizziness	2.8	Gastric antrum	Muc, SubM, MP	<5%	<5/50HPF	MALAT1–GLI1	Partial gastrectomy	No	ANED, 9
19 [20]	43/M	Reverse the acid, black stool	5.5	Gastric antrum	MP	2%	Infrequent	N/A	Partial gastrectomy	No	ANED, 24

Table 1 (continued)

	Age(y)/Sex	Symptoms	Size (cm)	Location	Layer	Ki-67	Mitoses	Molecular	Treatment	Metastases	Follow-up(mo)
20 [21]	5/F	Upper abdominal pain, melena, fever, headache, dizziness	3	Greater curvature	SubM, MP	N/A	Infrequent	N/A	Partial gastrectomy	No	ANED, 24
21 [22]	19/F	Loss of appetite, weight loss	8.1	Gastric antrum	Muc, SubM, MP	3%	<5/50HPF	No	Partial gastrectomy	No	ANED, 19
22 [23]	29/F	Upper abdominal pain	7	Gastric antrum	N/A	N/A	N/A	MALAT1–GLI1	Partial gastrectomy	No	ANED, 8
23 [24]	55/M	No	2	Lesser curvature	SubM, MP	20%	10/50HPF	ACTB-GLI1	ESD	No	ANED, 12
24 [25]	26/M	Abdominal pain, nausea, vomiting	6	Pylorus	N/A	N/A	N/A	MALAT1–GLI1	Partial gastrectomy	No	ANED, 2
25 [26]	19/M	Epigastric abdominal discomfort, decreased appetite	5.6	Gastric antrum	SubM, MP, SubS	2%	Rare	ACTB-GLI1	Antrectomy	No	ANED, 9
26 [27]	28/M	Epigastric pain	5.3	Gastric antrum	N/A	N/A	N/A	ACTB-GLI1	Antrectomy	No	ANED, 50
27	50/F	No	4.1	Greater curvature	TM	Epithelial: 40% Mesenchymal: 3%	Rare	No	Radical gastrectomy	No	ANED, 14

F, female; M, male; N/A, not available; ANED indicates alive with no evidence of disease; AWD, Alive with disease; DOD, Dead of disease; LN, Lymph node; LR, Local relapse; Muc, mucosa; SubM, submucosa; MP, muscularis propria; SubS, subserosa; S, serosa; TM, transmura

including 8 Chinese, 10 American, 2 Portuguese, 2 Italian, 1 Japanese, 1 French, and 1 Tunisian literature.

Clinicopathologic Features

The clinicopathologic findings are summarized in Tables 2 and 3. The patients included 13 women and 14 men, ranging in age from 5 to 74 years (mean: 35.0 years; median: 29 years). The most common tumor site was the gastric antrum (15; 57.7%), followed by the greater curvature (6; 23.1%), the lesser curvature (2; 7.7%), fundus (1; 3.8%), pylorus (1; 3.8%) and proximal stomach (1; 3.8%). 26 patients underwent surgery, including partial gastrectomy ($n=16$), subtotal gastrectomy ($n=2$), radical gastrectomy ($n=1$), antrectomy ($n=4$), ESD ($n=2$), and EFTR ($n=1$). One patient received postoperative radiation. One patient received preoperative chemotherapy. Only 6 patients underwent biopsy, and the lesions were diagnosed as adenocarcinoma ($n=1$), gastroblastoma ($n=1$), mesenchymal lesion ($n=1$), GIST ($n=1$), epithelioid neoplasm ($n=1$),and neuroendocrine origin ($n=1$).

The tumor size ranged from 1.3 to 15 cm (mean: 5.7 cm; median: 5.4 cm). The neoplasms were predominantly centered in the muscularis propria (15; 78.9%), with variable extension into other layers. The proportions of mesenchymal and epithelial component are variable. Ten cases had a dominant mesenchymal component(58.8%). The epithelial component in 5 cases comprised the majority of the tumor(29.4%). Another 2 cases showed relatively even proportions of these two components(11.8%). Nearly half of cases (13; 48.1%) had the special structure, lumina containing eosinophilic, inspissated secretions. Several neoplasms had haemorrhage(16; 66.7%), ulcer(9; 40.9%), necrosis(8; 32%) and peri-tumoral vascular emboli(2; 8%).

Immunophenotype

The positive and negative immunohistochemical markers reported in previous literature are summarized in eTable 1 and eTable 2 in the supplement, respectively. The epithelial component expressed pancytokeratin (24/25), CK18 (3/3), CK7 (7/9), Vimentin (3/21), CD10 (12/25), CD117 (2/24), CD99 (2/13), S100 (1/22), Syn (2/24), CD56 (15/18), LMWCK (9/10), EMA (7/12), CEA (1/3), NSE (1/3), GLI1 (4/4), bcl-2 (3/4), and MUC5ac (1/1). The mesenchymal component expressed Vimentin (21/21), CD10 (23/25), CD99 (2/13), SMA (1/21), Desmin

Table 2 Clinical features of 27 patients with gastroblastoma

Characteristic	
Sex(*n*=27)	
Men	14
Women	13
Age, mean, median (range), y (*n*=2)	35.0, 29(5-74)
Tumor site(*n*=26)	
Greater curvature	6
Gastric antrum	15
Lesser curvature	2
Fundus	1
Proximal stomach	1
Pylorus	1
Tumor size, mean, median (range), cm (*n* = 26)	5.7, 5.4(1.3-15)
Biopsy (*n*=6)	
Adenocarcinoma	1
Gastroblastoma	1
Mesenchymal lesion	1
GIST	1
Neuroendocrine origin	1
Epithelioid neoplasm	1
Treatment (*n*=26)	
Gastrectomy	21
Chemotherapy+Gastrectomy	1
Gastrectomy+Radiation	1
EFTR	1
ESD	2
Metastases/Relapse (*n*=4)	
Liver, lymph node, retroperitoneal and bladder	1
Liver	1
Lymph node	1
Local relapse	1
Follow-up (*n*=26)	
Alive	25
Dead of disease	1

Table 3 Pathologic features of 27 patients with gastroblastoma

Characteristic	
Ulcerated(*n*=22)	9
Center(*n*=19)	
Muscularis propria	15
Submucosa	3
Muscularis propria and submucosa	1
Prominent component(*n*=17)	
Epithelial component	5
Mesenchymal component	10
Even proportions	2
Luminal structures containing eosinophilic material/Special structure(*n*=27)	13
Layer(*n*=21)	
Only muscularis propria	7
Mucosa and submucosa	1
Submucosa and muscularis propria	3
Muscularis propria and subserosa	2
Mucosa, submucosa and muscularis propria	2
Submucosa, muscularis propria and subserosa	1
Transmural	5
Haemorrhagic(*n*=24)	16
Necrosis(*n*=25)	8
Peri-tumoral vascular emboli(*n*=25)	2
Fusion gene(*n*=15)	
MALAT1–GLI1	8
EWSR1-CTBP1	1
PTCH1-GLI2	1
ACTB-GLI1	3
GLI(*n*=10)	6
C-KIT gene mutation(*n*=5)	0

(1/22), Syn (2/24), CD56 (17/18), TLE-1 (1/4), NSE (1/3), GLI1 (4/4), and bcl-2 (3/4), PDL1 (1/1), HDAC2 (1/1), CD68 (1/1), and CD163 (1/1). Both epithelial and mesenchymal components are negative for ChromograninA (0/22), CD34 (0/21), Calretinin (0/10), P63 (0/7), CDX2 (0/7), TTF1 (0/4), Inhibin (0/4), CK20 (0/6), DOG1 (0/20), CK5/6 (0/4), PLAP (0/2), ALK (0/1), Melan-A (0/3), SOX10 (0/4), HMWCK (0/2), MOC31 (0/1), HMB45 (0/4), WT1 (0/3), CD31 (0/3), ERG (0/1), GFAP (0/1), STAT6 (0/4), Caldesmon (0/3), Calponin (0/3), PDGFRA (0/2), P16 (0/1), ER (0/1), PR (0/1), SDHB (0/1), HER-2 (0/1), MyoD1 (0/1), TFE3 (0/1), SSTR2 (0/1).

Molecular pathology

Fusion genes was found in 13 of 15 cases, including MALAT1–GLI1(*n*=8), EWSR1-CTBP1(*n*=1), PTCH1-GLI2(*n*=1), and ACTB-GLI1(*n*=3). None of the 5 cases had C-KIT gene mutation.

Follow-up

Follow-up information was available for 26 patients ranging from 2 to 168 months (mean: 31.0 ; median: 16 months). There were 4 patients who had metastases. Case #5 was noted to have liver, bladder, and retroperitoneal metastases during the surgery and had metastatic tumor in 1 of the 4 perigastric lymph nodes. Case #8 developed loco-regional recurrence in the retro-gastric area. The liver metastasis of Case #10 was present at the time of consultation. Case #12 had local relapse.

Discussion

Gastroblastoma, a rare biphasic neoplasm, was first reported by Miettinen et al. in 2009, and up to now a total of 27 cases have been reported. The term "gastroblastoma" was proposed in view of the resemblance to the childhood biphasic neoplasms termed blastomas, especially (pleuro) pulmonary blastomas and nephroblastomas [3]. The pathogenesis of this tumor is still unknown, while it has been proposed that gastroblastoma may relate better to the spindle epithelial tumor with thymus like differentiation and the desmoplastic nested spindle cell tumor of liver rather than to classic 'blastic' tumors

because of the favorable prognosis [4]. Perhaps due to the limited number of cases reported, no epidemiological studies have reported incidence and prevalence of this tumor to date.

Gastroblastoma mostly occurs in patients under 60 years old, with no sex preference. Patients present nonspecific symptoms or even no symptoms at all. The most common location of gastroblastoma is the gastric antrum. In terms of histopathology, the neoplasm consists of both epithelial and mesenchymal component, and predominantly centered in the muscularis propria, with variable extension into other layers. Nearly half of the cases have luminal structures consisting of epithelial cells containing eosinophilic material. Most cases had low Ki-67 and an infrequent mitotic index ranging from 0 to 10 mitoses per 50 high-power fields. The immunohistochemical staining analysis shows that the epithelial and the mesenchymal component of most cases express Pancytokeratin and CD10 respectively, and all the available cases express Vimentin in the mesenchymal components. In addition, both the two components often express CD56.

In terms of cellular and molecular genetics, four fusion gene have been found in gastroblastoma. MALAT1-GLI1 fusions were first identified by Graham and colleagues in their series of four cases in 2017 [9]. Subsequently, another 2 cases also found this fusion genes [11, 19]. Koo et al. [16] identified a novel EWSR1-CTBP1 fusion in a 17-year-old man with Wiskott-Aldrich syndrome and a history of multiple allogeneic bone marrow transplantation and radiotherapy in infancy. In 2022, Chen et al. [17] reported a case of a 58-year-old man that demonstrated a novel PTCH1-GLI2 gene fusion. ACTB-GLI1 fusion was found in three cases [24, 26, 27]. These fusion genes are expected to provide precise targeted therapy for patients with locally advanced or metastatic tumors and allow for the confident diagnosis of gastroblastoma.

Among the 13 gastroblastoma cases with fusion gene, 11 cases have GLI1 gene fusions. Four intestinal-based neoplasms were found to have clinicopathologic, immunohistochemical, and molecular features similar to gastroblastoma [27, 28]. Three of them harbored GLI1 gene fusions. Because of the indolence of the tumor, the term "blastoma" may be not inappropriate. Recently, Given the spectrum of morphologic and immunohistochemical features of these tumors, and the variability in GLI1 fusion partners, Jessurun et al. propose that tumors sharing this constellation of attributes be classified as GLI1-rearranged enteric tumors [29].

As for a definite diagnosis of gastroblastoma, comprehensive evaluation of clinical and pathological features, including tumor site, histomorphology, IHC patterns, and mutations, should be considered. In this case, the biphasic morphology, the tumor location, immunohistochemical features, special glands or luminal structure supports the diagnosis of gastroblastoma.

The differential diagnosis for GB is a number of biphasic malignant tumors, including synovial sarcoma, GIST, carcinosarcoma and sarcomatoid carcinoma.

Most reported gastric synovial sarcomas were the monophasic type [30]. The vast majority of synovial sarcomas carry a t(X;18)(p11.2;q11.2) chromosomal translocation, involving SSX1, SSX2, very rarely SSX4 and SS18L [31–33]. In our case, depending on the fluorescence in situ hybridization and NGS, the specific SS18 gene rearrangement is absent.

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal tract [34]. Histologically, most GISTs consist of spindle cells or epithelioid cells, only 10% of GISTs are mixed type with spindle and epithelioid cells [35]. GISTs include a variety of molecular entities with usually mutually exclusive activating oncogene mutations, mostly KIT or PDGFRA mutations [36]. The commonly expressed IHC markers include CD117 (95% of GIST), DOG-1 (95% of GIST), CD34 (70% of GIST), smooth muscle actin (SMA; 25% of GIST) and desmin (<5% of GIST). Focally positive for CD117, negativity for DOG1 by immunohistochemistry and no KIT or PDGFRA mutations of our case eliminate this diagnosis.

With highly atypical squamous, adenocarcinomatous, or undifferentiated epithelial elements [37] and older patients, carcinosarcoma and sarcomatoid carcinoma in the stomach has high mortality, and 50% of patients die within 6 months after surgery [38]. In our case, the relative uniformity of neoplastic cells, low mitotic activity and Ki-67 index exclude the diagnosis of carcinosarcoma and sarcomatoid carcinoma.

All of the patients underwent surgeries, except for one's treatment and follow-up information was not available. One patient had the preoperative chemotherapy and another one had radiation after the surgery. The follow-up positron emission tomography scan showed no response of the tumor to 6 weeks of chemotherapy [5]. Surgical resection with clear margins has been the preferred treatment of choice [13]. Most of patients underwent gastrectomy, while 2 ESD and 1 EFTR. Due to the Gastroblastoma mostly centers in the muscularis propria with various degrees of infiltration, ESD might not be an appropriate treatment. For subepithelial tumors (SETs) and epithelial neoplasia extending deeper than the mucosa or associated with significant fibrosis, EFTR is emerging as a therapeutic option [39]. The tumor size suitable for retrieval of solid SETs by EFTR is <30 mm in the view of resection completeness and safety [40]. Therefore, for GB with a maximum diameter less than 30 mm, EFTR can be considered.

Among the 27 cases reported so far, except 4 cases(Case #5, #8, #10, #12) had metastases, others had no recurrent or metastatic disease during the reported monitoring period. Case #12, for whom most information was not available showed a liver metastasis. As the only case with multiple metastases, case #5, a 28 year-old man, had a transmural tumor measuring 3.8×3.3×2.5 cm with a cystic hemorrhagic focus. After 6 weeks of chemotherapy and partial gastrectomy, the patient was alive with disease for 3 months. The mitotic index was 2 mitoses per 10 high-power fields. The only one dead case was case #8, a 29 year-old lady, who had a tumor, measuring 7×4×4 cm, located on the posterior gastric wall with encroachment of the splenic hilum. Partial gastrectomy with splenectomy was performed. The transmural tumor with focal areas of necrosis and hemorrhage showed invasion of the splenic hilum and lymph node metastasis. Six months after the sugery, the patient developed loco-regional recurrence, thus, underwent surgical debulking and died one month after debulking due to massive pulmonary embolism. Case #8 had the most frequent mitoses, 21 mitoses per 10 high power fields. Case #12 showed a 74 year-old man who was the oldest patient ever found. It was excised a 9 cm tumor with haemorrhagic. Neither mitotic activity nor necrosis was detected. The patient developed loco-regional recurrence 51 months after the surgery. The recurrent tumor consisted of three masses, with sizes of 0.3 cm, 11 cm, and 0.5 cm, and was removed through partial gastrectomy. Both case #8 and #12 had peri-tumoral vascular emboli. Because of the limited number of cases reported, the risk factors that affect the prognosis or grade the malignancy have not been clearly defined. Based on the characterized biphasic morphology consisting of both epidermal and stromal components, the risk factors of gastroblastoma could be inferred by gastric cancer and GIST, including the tumor size, mitoses, invasion depth, age, and so on.

Conclusion

Our case reports a gastroblastoma with a characteristic histopathology and IHC patterns but without any fusion genes. Due to the limited cases reported, the diagnostic standard still should be the characterized biphasic morphology and IHC patterns. Whether other factors could be the diagnostic standard needs more research. The treatment involves EFTR and gastrectomy. The epidemiological studies, pathogenesis, and prognosis factors have not been clearly defined.

The present report is, to our knowledge, the first one to summarize all the gastroblastoma cases ever reported through various aspects thoroughly. Our aim in presenting this exhaustive literature review is to increase awareness of gastroblastoma, better characterize the disease, and provide a reference point for gastroblastoma research in the future. It is hoped that more cases can be found, which is expected to deepen the understanding of the disease and avoid missed diagnosis and misdiagnosis.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12957-024-03534-y.

Supplementary Material 1

Author contributions
Zijin Luo contributed to the preparation of the manuscript, Figures and Tables. Jian Cui, Fuhai Ma, Zijian Li, Shishu Yin contributed to the data collection. Zheng Wang performed the immunohistochemical staining. Gang Zhao contributed to the final approval of the version to be published.

Funding
This study was supported by the National High Level Hospital Clinical Research Funding (BJ-2024-095).

Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethical approval
Our study comply with the Ethical Standards.

Competing interests
The authors declare no competing interests.

Data transparency statement
The study materials will not be made available to other researchers.

Received: 29 June 2024 / Accepted: 9 September 2024
Published online: 27 September 2024

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