Case Reports

Journal of INTERNATIONAL MEDICAL RESEARCH

Journal of International Medical Research
50(5) 1–7

DOI: 10.1177/03000605221100768
journals.sagepub.com/home/imr
SAGE

A case of primary hepatic stromal tumour misdiagnosed as a liver cyst

Fei Liu[1], Si-Hua Wu[1], Qiang-Qiang Sun[1], Sheng-Hai Liu[1] and De-Fa Hou[2]

Abstract

Primary hepatic stromal tumours are very rare and there are only sporadic reports in the literature. Due to the lack of specificity in their clinical manifestations and imaging features, these tumours are easily misdiagnosed. This current report presents a case of primary liver stromal tumour that was misdiagnosed as a liver cyst. The 72-year-old male patient was admitted to the hospital due to right upper abdomen fullness and discomfort for more than 2 weeks. Colour Doppler ultrasonography and enhanced computed tomography examinations revealed a cystic mass in the right lobe of the liver. The preoperative diagnosis was a liver cyst and the laparoscopic fenestration was performed. The pathological examination demonstrated that it was a primary hepatic stromal tumour. Gastroenteroscopy was performed postoperatively and no lesions were found in the gastrointestinal tract. Imatinib mesylate was given orally as the salvage therapy and a radical operation was planned at the patient's request. This current case serves as a reminder that clinicians should consider the possibility that it could be a primary hepatic stromal tumour rather than a hepatic cyst. A multidisciplinary team is necessary for the diagnosis and treatment of patients with a primary hepatic stromal tumour.

Keywords

Primary hepatic stromal tumour, misdiagnosis, case report

Date received: 22 December 2021; accepted: 22 April 2022

Introduction

Gastrointestinal stromal tumours are the most common mesenchymal tumours, which mainly occur in the gastrointestinal tract, being found in the stomach (60–70%), small intestine (20–25%), colon and rectum (5%) and oesophagus (<5%).[1–3]

[1]Department of General Surgery, Fuyang Tumour Hospital, Fuyang, Anhui Province, China
[2]Department of Pathology, Fuyang Tumour Hospital, Fuyang, Anhui Province, China

Corresponding author:
De-Fa Hou, Department of Pathology, Fuyang Tumour Hospital, 146, Hebin East Road, Yingzhou District, Fuyang, 236000, Anhui Province, China.
Email: defa.75@163.com

Gastrointestinal stromal tumours have diverse clinical manifestations and an accurate diagnosis is mainly based on histological and immunohistochemical criteria. Among them, cluster of differentiation (CD) 117 is a specific marker in the immunohistochemical examination of gastrointestinal stromal tumours.[3]Endoscopic ultrasound-guided tissue biopsy is also an important diagnostic method.

Primary stromal tumours that originate from the liver are extremely rare and there are only sporadic reports in the literature.[4,5] The disease is easily misdiagnosed because of a lack of specificity in clinical manifestations and imaging features.[4] This current report presents a case of primary hepatic stromal tumour that was misdiagnosed as a liver cyst. The patient underwent laparoscopic fenestration and the final pathological examination revealed a primary hepatic stromal tumour. A systematic literature review was undertaken to comprehensively identify the characteristics of primary hepatic stromal tumours.

Case report

In April 2021, a 72-year-old male patient was admitted to the Department of General Surgery of Fuyang Tumour Hospital due to right upper abdomen fullness and discomfort for more than 2 weeks. On admission, his abdomen was flat, with no intestinal pattern or peristaltic, and no varicose veins in the abdominal wall. A mass of approximately 16×12 cm in the right-side costal margin was palpable, with a smooth surface, and without tenderness. A colour Doppler ultrasound examination of the abdomen revealed a cystic mass of 16.4×12.8 cm in the right lobe of the liver, with a thin cyst wall, smooth border and uniform echo. The enhanced computed tomography (CT) examination of the abdomen showed a huge cystic low-density shadow on the right lobe of the liver, with clear margins, and the largest cross-section was about 16.7×12.0 cm. There was no obvious enhancement and the CT impression was that of a liver cyst (Figure 1). The patient's routine blood analyses, liver function and liver tumour markers were all normal. The clinical diagnosis of a liver cyst was made based on the patient's symptoms and imaging examinations. It was decided to perform laparoscopic fenestration and drainage of the liver cyst under general anaesthesia. Upon laparoscopy, a huge cyst in the right lobe of the liver was seen, with high tension, and the rest of the liver was soft with no obvious nodules. No abnormalities were found in the gallbladder, common bile duct, spleen, gastrointestinal tract and pelvic cavity. The cyst was penetrated and approximately 2000 ml of coffee-like fluid was drawn out. A laparoscopic fenestration was created, part of the cyst wall was removed for pathological examination and bleeding from the cyst edge was stopped by an ultrasonic knife and suture. After washing and checking, there was no obvious bleeding or bile leakage, so a drainage tube was placed in the cyst cavity. The patient recovered uneventfully after the surgery. Pathological examination confirmed it to be a primary hepatic stromal tumour, more specifically a rare type of extragastrointestinal stromal tumour. The immunohistochemistry results were as follows: CKp–, Vim+, desmin focal+, SMA vascular+, S-100–, CD34 vascular+, BCL-2+, STAT6 nuclear+, CD117+, DOG-1+ and Ki67 + 2% (Figure 2). Gastroenteroscopy was performed postoperatively and no lesions were found in the gastrointestinal tract. Imatinib mesylate was given orally at a dosage of 400 mg/day for 12 months as salvage therapy. A radical operation was planned at the patient's request.

All procedures performed in this case were undertaken according to the Declaration of Helsinki (as revised in 2013). The reporting

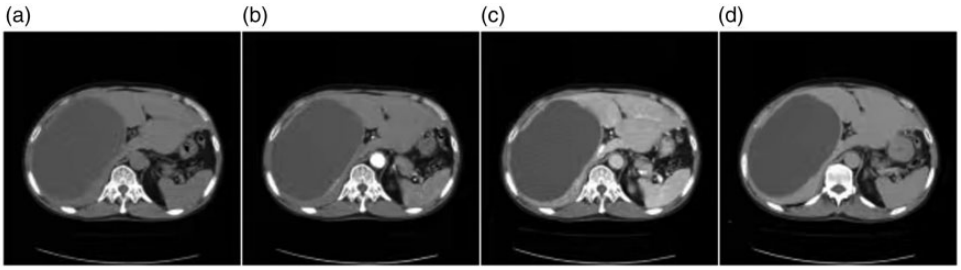

Figure 1. Preoperative enhanced computed tomography (CT) images of a 72-year-old male patient that presented with right upper abdomen fullness and discomfort for more than 2 weeks: (a) a CT scan showed a huge cyst in the right lobe of the liver, approximately 16.7 × 12.0 cm in size, with clear margins; (b) there was no obvious enhancement during the arterial phase; (c) the normal liver parenchyma in the venous phase was enhanced and (d) there was no obvious enhancement in the delayed phase.

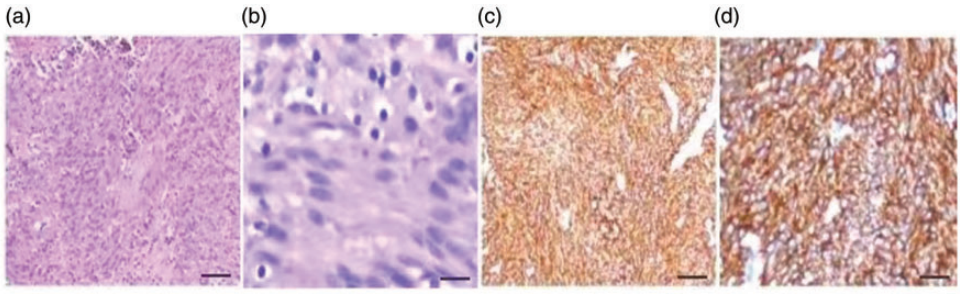

Figure 2. The postoperative pathological examination of the tumour removed from a 72-year-old male patient that presented with right upper abdomen fullness and discomfort for more than 2 weeks: (a) the spindle tumour cells were arranged in bundles and the nuclei were deeply stained (haematoxylin and eosin, scale bar 50 μm); (b) the tumour cells were arranged in a palisade (haematoxylin and eosin, scale bar 50 μm); (c) immunohistochemical staining showed cluster of differentiation 117 in the cell membrane and cytoplasm (scale bar 50 μm) and (d) immunohistochemical staining showed DOG-1 in the cell membrane and cytoplasm (scale bar 50 μm). The colour version of this figure is available at: http://imr.sagepub.com.

of this case conforms to CARE guidelines.[6] Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Discussion

Gastrointestinal stromal tumours are a type of tumour that originate from the mesenchymal tissue of the gastrointestinal tract.[7]They account for 1–2% of all gastrointestinal tumours.[8] This type of tumour originates from the interstitial cells of Cajal (ICC) in the gastrointestinal tract.[9] Cajal cells mainly play a role in triggering gastrointestinal activity.[7] This tumour usually occurs in the digestive tract including the stomach, small intestine, colorectal and oesophagus.[8]

Primary hepatic stromal tumours are extremely rare and there are only sporadic cases reported in the literature.[4,5] A previous study reported that ICC-like cells were observed in the portal vein of humans and animals by immunohistochemical staining, which were similar in function and structure to ICC cells.[10] Further research confirmed that ICC-like cells exist in the human

embryonic liver as single precursor or progenitor cells.[11] Therefore, it can be inferred that primary hepatic stromal tumours originate from these ICC-like cells.

Primary hepatic stromal tumours lack typical clinical manifestations and laboratory test findings, Imaging usually show a very large tumour mass, occasionally accompanied with abdominal discomfort and gastrointestinal symptoms.[5,12] It has also been reported that the this type of tumour was discovered when a patient sought medical advice due to rapid weight loss.[5] Slightly elevated levels of alpha-fetoprotein and glycoproteins 19-9 and 125 often indicate that the disease may be malignant, but there are no tumour markers that are sensitive and specific to this disease.[4] Primary hepatic stromal tumours also have no typical features on imaging and it is not easy to differentiate between primary hepatic stromal tumours and other liver diseases before surgery. The imaging findings of this type of tumour are mostly round cysts, occasionally lobulated. The cyst wall is usually thick, and necrosis and intratumoral haemorrhage may appear in the central area.

As primary hepatic stromal tumours are extremely rare, there is no consensus on how to treat them. The management is mainly based on what would be used for gastrointestinal stromal tumours. R0 resection or partial resection combined with postoperative targeted therapy could benefit patients.[13]Patients with tumours that are not resectable or those that cannot tolerate surgery might require cytoreductive surgery.[13] Imatinib can be used as adjuvant therapy.[13] Researchers have also used radiofrequency and microwave ablation for local treatment, and the initial effects were relatively satisfactory.[14,15]

This current report describes a case of primary hepatic stromal tumour that presented as a huge cystic lesion in the right lobe of the liver. The male patient presented with right upper abdomen fullness and discomfort. Colour Doppler ultrasonography and enhanced CT examination revealed a cystic mass in the right lobe of the liver. The preoperative diagnosis was a liver cyst and the laparoscopic fenestration was performed. However, the pathological examination demonstrated that it was a primary hepatic stromal tumour. This was an unusual case of misdiagnosis. Perhaps due to its rarity and the lack of specific clinical manifestations and imaging features, the possibility of a primary hepatic stromal tumour was not considered during the preoperative diagnosis. As a consequence, the treatment continued based on it being a hepatic cystic lesion.

A review of the published case reports of primary hepatic stromal tumours over the past 15 years was undertaken (Table 1), which included 13 males and 10females with a median age of 59 years.[2,4,5,14–32] The common features were that the primary hepatic stromal tumour lacked typical clinical manifestations and patients usually presented as an accidental liver mass. The tumour size ranged from 2.4 to 23 cm with a median size of 15 cm. Among them, 65.2% (15 of 23 patients) patients had a tumour size of >10 cm.[4,5,16–19,21,23,24,26,28–32] Tumours were mostly located in one lobe of the liver, but in a minority of patients, the tumour was multilobular in the liver. Regarding their treatment, more than half of the patients underwent liver resection.[2,4,5,17–23,25–32] The remaining received chemotherapy or other treatments.[5,14,15,24] One patient received no treatment.[16]The risk grade was high in all patients.

In conclusion, primary hepatic stromal tumours are sporadic and remain challenging in terms of accurately identifying the liver as the primary site of the tumour. The first-line treatment of primary hepatic stromal tumours is surgical resection. Additionally, imatinib can be used as

Table 1. Case reports of primary hepatic gastrointestinal stromal tumours reported between 2003 and 2019.

Author	Country	Age, years	Sex	Location	Size, cm	Treatment	Cell type	Risk grade	Recurrence
Hu et al. 2003[17]	USA	79	F	R/I	15.0	HR	S	High	16/Met
De Chiara et al. 2006[18]	Italy	37	M	R/I	18.0	HR	S	High	14/Met
Luo et al. 2009[14]	China	17	M	R/I	5.1	RFA	S	High	3/DFS
Ochiai et al. 2009[26]	Japan	30	M	R + L/I	20	HR	S + E	High	24/Rec + Met
Yamamoto et al. 2010[31]	Japan	70	M	L/I	20	HR	E	High	NA
Li et al. 2012[16]	China	53	M	R/I	20.0	Untreated	NA	High	NA
Louis et al. 2014[19]	India	55	F	R + L/3	18.0	HR	S	High	7/DFS
Zhou et al. 2014[20]	China	56	M	R + L/I	10.0	HR	S	High	12/DFS
Bhoy et al. 2014[21]	India	41	F	R/2	15.0	HR	S	High	5/DFS
Kim et al. 2014[22]	South Korea	71	M	L/I	7.0	HR	S	High	19/DFS
Mao et al. 2015[23]	China	60	F	R + C/I	19	HR + AT	S	High	12/DFS
Lin et al. 2015[24]	China	65	M	L/I	12	Chemo	S	High	13/Met
Su et al. 2015[25]	China	67	F	R/I	7.4	HR	NA	High	14/Rec
Nagai et al. 2016[2]	Japan	70	F	L/I	6.8	HR	S	High	10/DFS
Liu et al. 2016[15]	China	56	F	L/I	2.4	MW.	S	High	17/Met
Wang et al. 2016[27]	China	61	M	C/I	7.3	HR	NA	High	11/DFS
Cheng et al. 2016[28]	China	63	M	R/I	13	HR	S	High	60/DFS
Losada et al. 2016[29]	Chile	61	M	L/I	15	HR	NA	High	NA
Carrillo Colmenero et al. 2017[30]	Spain	41	M	R + L/I	20	HR	S	High	18/DFS
Lok et al. 2017[32]	China	50	F	R/I	15	HR	S	High	6/Met
Joyon et al. 2018[5]	France	56	M	R/I	10	TACE + HR + T + Chemo	S	High	12/Rec
Joyon et al. 2018[5]	France	59	F	R + L/I	23	Chemo	S + E	High	18/DFS
Xu et al. 2019[4]	China	64	F	R/I	15	HR	S	High	5/DFS

F, female; R, right lobe of liver; HR, hepatic resection; S, spindle; Met, metastasis; M, male; RFA, radiofrequency ablation; DFS, disease-free survival; L, left lobe of liver; E, epithelioid; Rec, recurrence; NA, not available; C, caudate lobe of liver; AT, autotransplantation; Chemo, chemotherapy; MW, microwave ablation; TACE, transcatheter arterial chemoembolization; T, transplantation.

adjuvant therapy for locally advanced tumours and as the first-line treatment for unresectable tumours, recurrence and metastases. A multidisciplinary team is necessary for the diagnosis and treatment of a patient with a primary stromal tumour. The misdiagnosis in this current case serves as a reminder that clinicians should consider the possibility that it could be a primary hepatic stromal tumour rather than a hepatic cyst.

Acknowledgements

We thank the editor Dr Gordon Mallarkey and the reviewers for valuable comments concerning this manuscript.

Author contributions

F.L., S.H.W. and D.F.H. contributed to the study design. F.L., S.H.W., Q.Q.S., S.H.L. and D.F.H. participated in the treatment of this patient. F.L., S.H.W. and D.F.H. participated in drafting this manuscript. F.L. and D.F.H. revised the manuscript.

Declaration of conflicting interest

The authors declare that there are no conflicts of interest.

Funding

This clinical research received no specific grant from funding agency in the public, commercial, or not-for-profit sectors.

ORCID iD

Fei Liu https://orcid.org/0000-0002-4986-8500

References

1. Miettinen M and Lasota J. Gastrointestinal stromal tumors–definition, clinical, histological, immunohistochemical, and molecular genetic features and differential diagnosis. *Virchows Arch* 2001; 438: 1–12.
2. Nagai T, Ueda K, Hakoda H, et al. Primary gastrointestinal stromal tumor of the liver: a case report and review of the literature. *Surg Case Rep* 2016; 2: 87.
3. Mantese G. Gastrointestinal stromal tumor: epidemiology, diagnosis, and treatment. *Curr Opin Gastroenterol* 2019; 35: 555–559.
4. Xu L, Zhang M and Xu M. Primary hepatic gastrointestinal stromal tumor with right adrenal gland invasion: a case report and systematic literature review. *Medicine (Baltimore)* 2019; 98: e15482.
5. Joyon N, Dumortier J, Aline-Fardin A, et al. Gastrointestinal stromal tumors (GIST) presenting in the liver: diagnostic, prognostic and therapeutic issues. *Clin Res Hepatol Gastroenterol* 2018; 42: e23–e28.
6. Gagnier JJ, Kienle G, Altman DG, et al; The CARE guidelines: consensus-based clinical case reporting guideline development. *Headache* 2013; 53: 1541–1547.
7. Kindblom LG, Remotti HE, Aldenborg F, et al. Gastrointestinal pacemaker cell tumor (GIPACT): gastrointestinal stromal tumors show phenotypic characteristics of the intestinal cells of Cajal. *Am J Pathol* 1998; 152: 1259–1269.
8. Menge F, Jakob J, Kasper B, et al. Clinical presentation of gastrointestinal stromal tumors. *Visc Med* 2018; 34: 335–340.
9. Taşkın OÇ, Armutlu A, Adsay V, et al. Clinicopathologic and immunohistochemical characteristics of upper gastrointestinal leiomyomas harboring interstitial cells of Cajal: a potential mimicker of gastrointestinal stromal tumor. *Ann Diagn Pathol* 2020; 45: 151476.
10. Rusu MC, Pop F, Hostiuc S, et al. Extrahepatic and intrahepatic human portal interstitial Cajal Cells. *Anat Rec (Hoboken)* 2011; 294: 1382–1392.
11. Rusu MC, I Dută, Didilescu AC, et al. Precursor and interstitial Cajal cells in the human embryo liver. *Rom J Morphol Embryol* 2014; 55: 291–296.
12. Liu Z, Tian Y, Liu S, et al. Clinicopathological feature and prognosis of primary hepatic gastrointestinal stromal tumor. *Cancer Med* 2016; 5: 2268–2275.
13. Fernandes MR, Ghezzi CLA, Grezzana-Filho TJ, et al. Giant hepatic extra-gastrointestinal stromal tumor treated with cytoreductive surgery and adjuvant systemic therapy: a case report and review of literature. *World J Gastrointest Surg* 2021; 13: 315–322.
14. Luo XL, Liu D, Yang JJ, et al. Primary gastrointestinal stromal tumor of the liver: a case report. *World J Gastroenterol* 2009; 15: 3704–3707.
15. Liu L, Zhu Y, Wang D, et al. Coexisting and possible primary extra-gastrointestinal stromal tumors of the pancreas and liver: a single case report. *Oncol Lett* 2016; 11: 3303–3307.
16. Li ZY, Liang QL, Chen GQ, et al. Extra-gastrointestinal stromal tumor of the liver diagnosed by ultrasound-guided fine needle aspiration cytology: a case report and review of the literature. *Arch Med Sci* 2012; 8: 392–397.
17. Hu X, Forster J and Damjanov I. Primary malignant gastrointestinal stromal tumor of the liver. *Arch Pathol Lab Med* 2003; 127: 1606–1608.
18. De Chiara A, De Rosa V, Lastoria S, et al. Primary gastrointestinal stromal tumor of the liver with lung metastases successfully treated with STI-571 (imatinib mesylate). *Front Biosci* 2006; 11: 498–501.

19. Louis AR, Singh S, Gupta SK, et al. Primary GIST of the liver masquerading as primary intra-abdominal tumor: a rare extra-gastrointestinal stromal tumor (EGIST) of the liver. *J Gastrointest Cancer* 2014; 45: 392–394.
20. Zhou B, Zhang M, Yan S, et al. Primary gastrointestinal stromal tumor of the liver: report of a case. *Surg Today* 2014; 44: 1142–1146.
21. Bhoy T, Lalwani S, Mistry J, et al. Primary hepatic gastrointestinal stromal tumor. *Trop Gastroenterol* 2014; 35: 252–253.
22. Kim HO, Kim JE, Bae KS, et al. Imaging findings of primary malignant gastrointestinal stromal tumor of the liver. *Jpn J Radiol* 2014; 32: 365–370.
23. Mao L, Chen J, Liu Z, et al. Extracorporeal hepatic resection and autotransplantation for primary gastrointestinal stromal tumor of the liver. *Transplant Proc* 2015; 47: 174–178.
24. Lin XK, Zhang Q, Yang WL, et al. Primary gastrointestinal stromal tumor of the liver treated with sequential therapy. *World J Gastroenterol* 2015; 21: 2573–2576.
25. Su YY, Chiang NJ, Wu CC, et al. Primary gastrointestinal stromal tumor of the liver in an anorectal melanoma survivor: a case report. *Oncol Lett* 2015; 10: 2366–2370.
26. Ochiai T, Sonoyama T, Kikuchi S, et al. Primary large gastrointestinal stromal tumor of the liver: report of a case. *Surg Today* 2009; 39: 633–636.
27. Wang Y, Liu Y, Zhong Y, et al. Malignant extra-gastrointestinal stromal tumor of the liver: a case report. *Oncol Lett* 2016; 11: 3929–3932.
28. Cheng X, Chen D, Chen W, et al. Primary gastrointestinal stromal tumor of the liver: a case report and review of the literature. *Oncol Lett* 2016; 12: 2772–2776.
29. Losada H, Villaseca M, Vivallo C, et al. Gastrointestinal stromal tumor as cause of hepatic mass. *Hepatobiliary Surg Nutr* 2016; 5: 388–389.
30. Carrillo Colmenero AM, Serradilla Martín M, Redondo Olmedilla MD, et al. Giant primary extra gastrointestinal stromal tumor of the liver. *Cir Esp* 2017; 95: 547–550.
31. Yamamoto H, Miyamoto Y, Nishihara Y, et al. Primary gastrointestinal stromal tumor of the liver with PDGFRA gene mutation. *Hum Pathol* 2010; 41: 605–609.
32. Lok HT, Chong CN, Chan A, et al. Primary hepatic gastrointestinal stromal tumor presented with rupture. *Hepatobiliary Surg Nutr* 2017; 6: 65–66.