ORIGINAL ARTICLE

Transjejunal fine-needle biopsy using a forward-viewing echoendoscope for diagnosis of a rare hereditary paraganglioma

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CASE PRESENTATION

A 32-year-old man with a family history of neck paraganglioma presented with mild left abdominal pain. A 90-mm tumor adjacent to the left kidney was found, which exhibited significant vascular proliferation and was accompanied by spherical calcification at its center on contrast-enhanced CT (Fig. 1). Although paraganglioma was suspected, a definite diagnosis could not be made because serum catecholamines were in the normal range and ^{123}I-meta-iodobenzylguanidine (MIBG) did not clearly accumulate in the tumor on scintigraphy and single-photon emission CT/x-ray CT (Fig. 2). Around the tumor, an arteriovenous shunt formed, raising concerns about potential future chronic heart failure. Surgical resection was considered, but the tumor had invaded blood vessels and multiple organs, causing us to anticipate a highly invasive procedure. To justify the resection, a preoperative histological diagnosis was deemed necessary. We attempted both CT-guided punctures and EUS-guided FNA (EUS-FNA) using an oblique-viewing echoendoscope (GF-UCT260; Olympus, Tokyo, Japan) for diagnosis, but even slight respiratory fluctuations resulted in blood vessels entering the puncture line (Fig. 3A), and it proved unfeasible.

Abbreviations: EUS-FNA, EUS-guided FNA; MIBG, meta-iodobenzylguanidine; SDHB, succinate dehydrogenase complex subunit B.

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PROCEDURE

As the tumor was situated in the retroperitoneum, we sought to puncture from the ligament of Treitz using a forward-viewing echoendoscope (TGF-UC260J; Olympus) (Video 1, available online at www.videogie.org). A forward-viewing echoendoscope was carefully inserted, shortening the intestine several times using the fluoroscopically visible calcification as a guide. The tumor was clearly seen ahead of the endoscope from the ligament of Treitz. After aligning the endoscope straight, we proceeded with insertion of a 22-gauge SharkCore needle (Covidien, Waltham, Mass, USA). Stable and reliable puncture of the solid lesion was accomplished (Fig. 3B), yielding an adequate tissue sample without adverse events.

OUTCOME

The EUS-FNA specimen revealed that the tumor was positive for synaptophysin, GATA binding protein 3, and paired mesoderm homeobox protein 2B, while negative for cytokeratin AE1/3. As a result, it was indeed diagnosed as a paraganglioma. Furthermore, it tested negative for succinate dehydrogenase complex subunit B (SDHB), leading to the diagnosis of hereditary paraganglioma due to SDHB mutation (Fig. 4). We discussed 2 treatment options with the patient: surgery or chemotherapy with cyclophosphamide, vincristine, and dacarbazine. The patient refrained from selecting either of the treatments due to concerns regarding adverse events and opted for observation; subsequently, the patient decided to discontinue visiting our facility.

DISCUSSION

EUS-FNA is a valuable tool for diagnosing abdominal tumors.[1] Forward-viewing echoendoscopes offer a narrower and more anterior perspective compared with oblique scopes. However, they also provide a forward endoscopic view and enable deeper insertion. Even in challenging situations like the present case, in which EUS-FNA with an oblique-viewing echoendoscope is difficult, switching to a forward-viewing echoendoscope has been reported to enable safe and accurate EUS-FNA.[2]

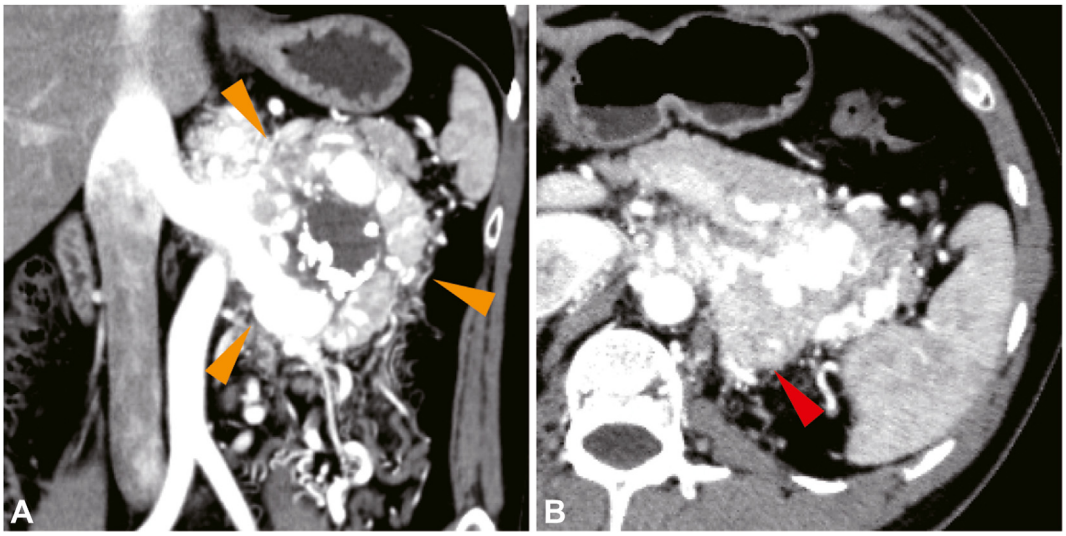

Figure 1. A, A 90-mm tumor (*orange arrow*) exhibiting significant vascular proliferation and calcifications was observed adjacent to the left kidney on contrast-enhanced CT. **B,** A few distinct solid lesions (*red arrow*) are observed on the cephalic and dorsal aspects of the tumor.

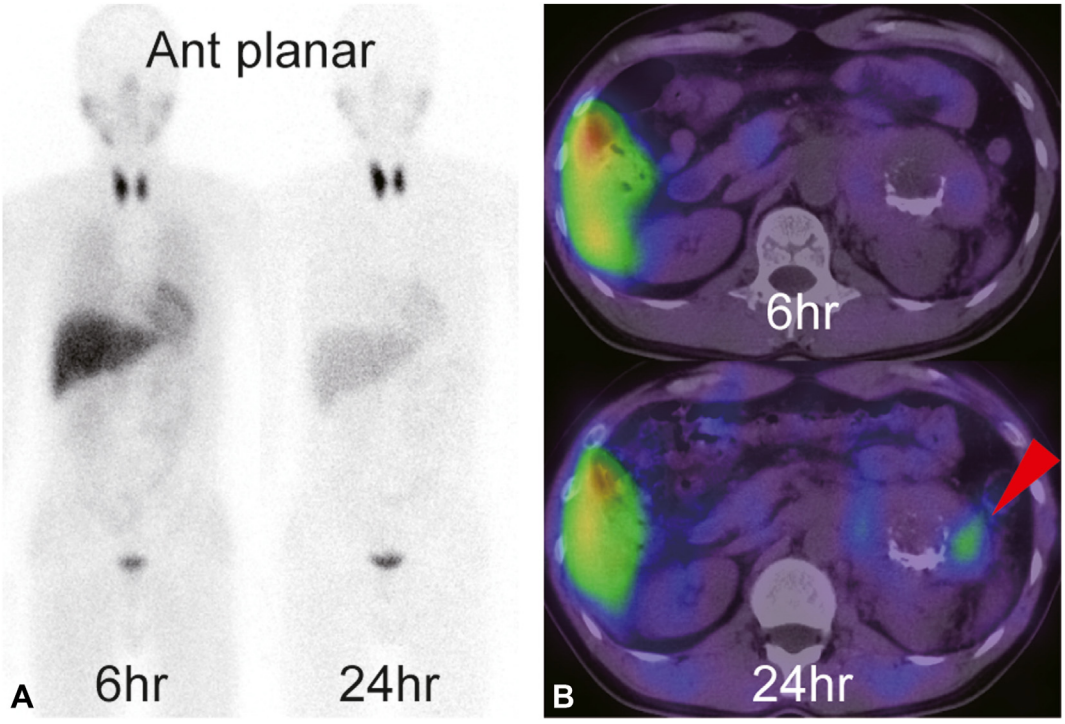

Figure 2. A, ^{123}I-meta-iodobenzylguanidine scintigraphy showing no uptake in the tumor at 6 and 24 hours after injection. **B,** ^{123}I-meta-iodobenzylguanidine single-photon emission CT/X-ray CT demonstrates no uptake at 6 hours and only insignificant accumulation (*red arrow*), lower than hepatic physiologic uptake at 24 hours after injection. Overall, the results of nuclear medicine studies did not confirm the diagnosis of paraganglioma.

In this case, EUS-FNA diagnosed hereditary paraganglioma with a SDHB mutation.[3] In recent years, numerous genes associated with pheochromocytoma and paraganglioma have been identified.[4] Tumors arising from these genetic factors are collectively referred to as hereditary pheochromocytoma/paraganglioma syndrome. This syndrome with SDHB mutations is typically reported to develop in the abdomen, exhibiting non-accumulation on MIBG scintigraphy,[5,6] which is consistent with this case. Additionally, it is associated with distant metastases and poor prognosis.[7] If surgery is not an option, chemotherapy with cyclophosphamide, vincristine, and dacarbazine is chosen. It has a 26% response rate in malignant cases, with responders experiencing a progression-free survival period of about 8 years.[8]

When MIBG accumulation of the tumor is not detected, even when paraganglioma is suspected, consideration should be given to the possibility of hereditary paraganglioma with a poor prognosis. In case of suspicion, ensuring a pathological

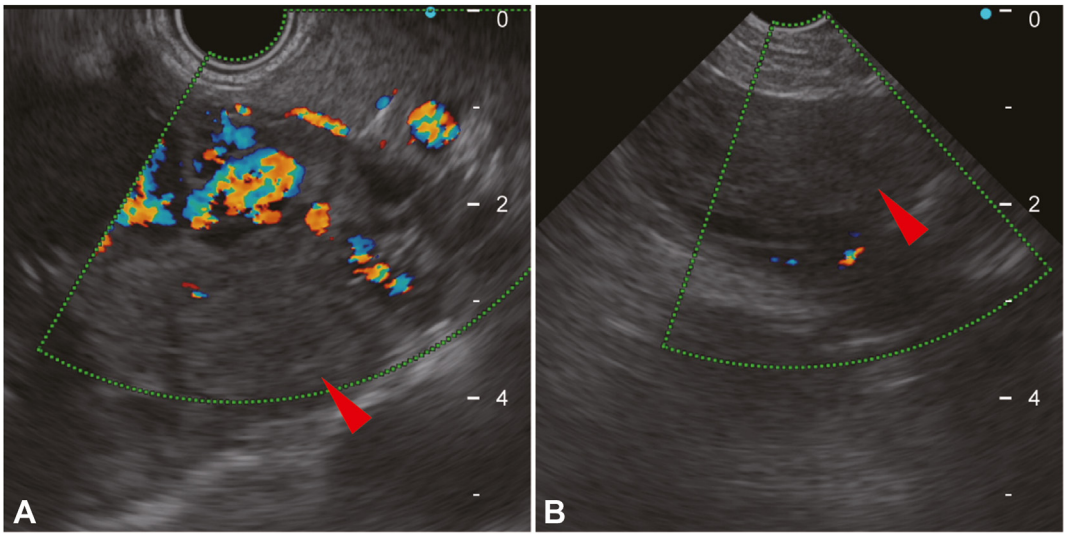

Figure 3. A, Using an oblique-viewing echoendoscope, we clearly observed the tumor from the stomach, but we could not puncture the distinct solid lesion (*red arrow*) due to notable vascular proliferation. **B,** The solid lesion (*red arrow*) with reduced blood flow was clearly observed from the vicinity of the ligament of Treitz using a forward-viewing echoendoscope.

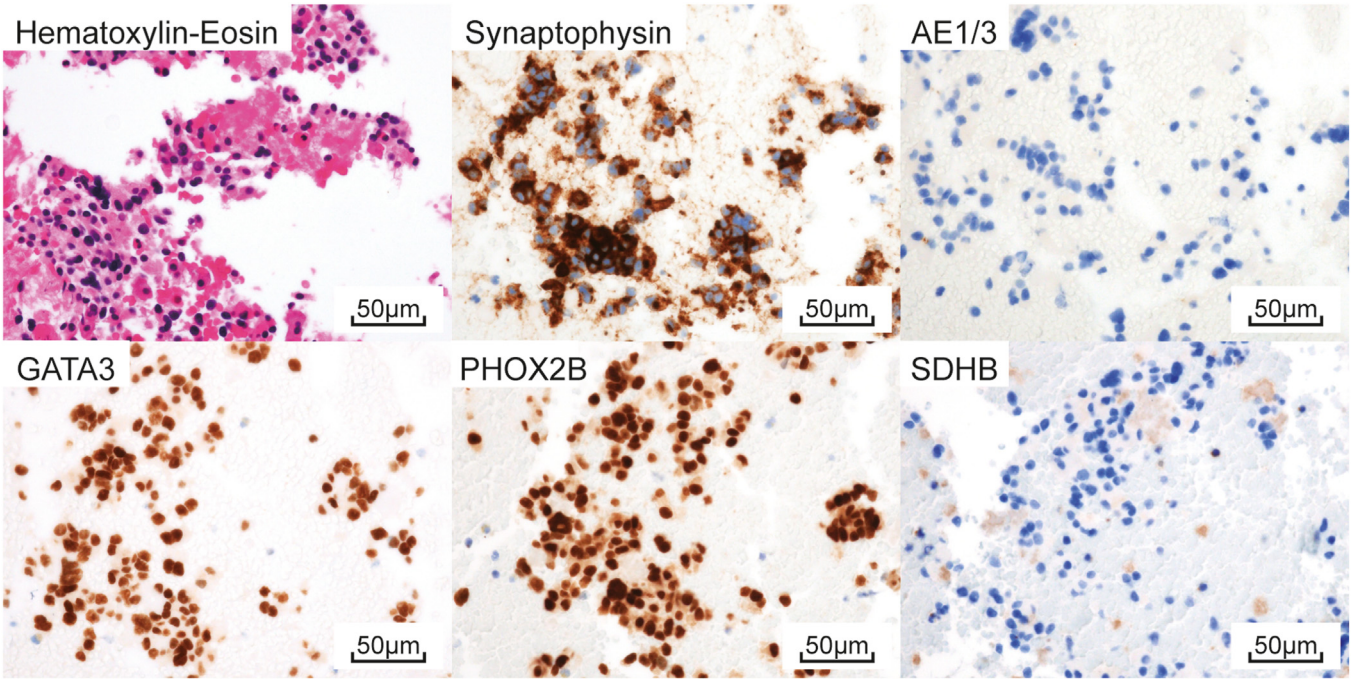

Figure 4. The tumor was positive for synaptophysin, GATA binding protein 3, paired mesoderm homeobox protein 2B, and negative for cytokeratin AE1/3; hence, it was diagnosed as paraganglioma. Furthermore, it tested negative for succinate dehydrogenase complex subunit B (SDHB) and was diagnosed as hereditary paraganglioma due to SDHB mutation.

diagnosis is crucial, even considering techniques such as EUS-FNA with a forward-viewing echoendoscope.

DISCLOSURE

The authors disclosed no financial relationships relevant to this publication.

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