A first report of endoscopic ultrasound-guided biopsy in the diagnosis of desmoid-type fibromatosis

A 65-year-old woman presented with recurrent sporadic mesenteric desmoid-type fibromatosis following a primary margin-positive surgical resection. The disease relapsed with new mesenteric nodules and a splenic hilar mass. Serial computed-tomography (CT) scans in the 4 months after initiation of tamoxifen revealed that the splenic hilar mass was gradually increasing in size (Fig. 1). A tissue diagnosis was necessary because radiological interpretation was not entirely consistent with desmoid-type fibromatosis. In the absence of safe access for a CT-guided biopsy, the patient was referred for an endoscopic ultrasound (EUS)-guided fine needle aspiration (FNA). Esophagogastroduodenoscopy revealed an approximately 3-cm subepithelial compression (Fig. 2).

EUS revealed a 3.5 × 2.36-cm, hypoechoic, heterogeneous splenic hilar mass with scattered hyperechoic foci (Fig. 3). Initially attempts were made at FNA with three passes of a 25-gauge needle (Echotip; Cook Medical Inc., Bloomington, Indiana, USA), all of which were nondiagnostic. Subsequently, an attempt was made at EUS-guided fine needle core biopsy (FNCB) with two passes of a 22-gauge needle (Echotip ProCore; Cook Medical). Cytopathology revealed fragments of dense collagenous tissue with bland spindle cells, compatible with desmoid-type fibromatosis (Fig. 4).
The patient was considered unresponsive to tamoxifen, therefore imatinib monotherapy was initiated. Desmoid-type fibromatosis is a rare fibroblastic proliferative disease, which is usually sporadic and very occasionally associated with Gardner’s syndrome [1]. Primary surgical resection is the standard treatment, but if resection margins are positive, local recurrence rates are approximately 80%. Treatment options for recurrence include repeat surgery, radiotherapy, tamoxifen, doxorubicin-based regimens, and more recently therapy with imatinib [2,3]. These tumors have a dense fibrous stroma, therefore FNA may not yield diagnostic material [4]. EUS-FNCB with a 19-gauge Quick-Core needle (Cook Medical) can be technically challenging. The new 22-gauge EchoTip ProCore needle features a core-trap and a reverse bevel for tissue-core procurement [5]. We performed FNCB with the aim of providing better cytology than had been obtained on the FNA specimens. Tissue procurement for histopathology can also be accomplished. To our knowledge, this is the first report of the use of EUS in the imaging and diagnosis of sporadic desmoid-type fibromatosis. Given this experience, we recommend EUS-guided FNCB instead of FNA when sampling suspected lesions with dense fibrous stroma such as desmoid tumors.

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Competing interests: None

S. G. Krishna, R. Suzuki, M. S. Bhutani
Department of Gastroenterology, Hepatology and Nutrition, University of Texas MD Anderson Cancer Center, Houston, Texas, USA

References


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Corresponding author
M. S. Bhutani, MD
Department of Gastroenterology, Hepatology and Nutrition
University of Texas MD Anderson Cancer Center
1515 Holcombe Blvd.
Houston, Texas 77030-4008
USA
Fax: +1-713-563-4408
Manoop.Bhutani@mdanderson.org

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