Pancreatic desmoid-type fibromatosis with beta-catenin gene mutation—Report of a case and review of the literature

Yoshitane Tsukamoto a,⁎, Masami Imakita b, Akiko Nishitani c, Toshikazu Ito c, Masaaki Izukura c, Seiichi Hirota a

a Department of Surgical Pathology, Hyogo College of Medicine, 1-1 Mukogawa-cho, Nishinomiya 663-8501, Hyogo, Japan
b Department of Pathology, Rinku General Medical Center, 2-23 Rinku-Ourai Kita, Izumisano-City 598-8577, Osaka, Japan
b Department of Surgery, Rinku General Medical Center, 2-23 Rinku-Ourai Kita, Izumisano-City 598-8577, Osaka, Japan

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A B S T R A C T

We experienced a rare case of pancreatic desmoid-type fibromatosis (DTF) in a 75-year-old Japanese woman. She was asymptomatic but routine examination including ultrasonography revealed a mass in the abdomen. For precise examination, she was referred to the regional hospital. Computed tomography showed that the mass was protruding anteriorly from the left-sided pancreas. Because of the enlargement of the mass lesion, distal pancreatectomy with splenectomy was performed after about 3 months. Macroscopically, the mass was encapsulated and approximately 8 cm in diameter. Histological examination revealed that spindle or blunt stellate cells were proliferating in parallel or storiform fashion with myxoid and fibrous background. The tumor cells did not show prominent atypia and mitoses were rarely seen, suggesting that the tumor was low grade or borderline. Immunohistochemistry showed obvious nuclear staining of beta-catenin. Furthermore, analysis of beta-catenin gene revealed that the tumor had a typical missense mutation of threonine to alanine at codon 41 (T41A) in exon 3. These findings confirmed the pathological diagnosis of DTF of the pancreas. To the best of our knowledge, 18 cases of pancreatic DTF have been reported in the English literature and beta-catenin gene mutation had been examined in only one case among them. Thus, our case is the 19th pancreatic DTF and the second case with confirmed beta-catenin gene mutation.

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1. Introduction

Desmoid-type fibromatosis (DTF), also known as desmoid tumor or aggressive fibromatosis, is a locally aggressive low grade fibroblastic tumor without metastatic potential. The tumor usually arises from the deep soft tissue and is characterized by infiltrative growth and high rate of recurrence after removal [1]. The estimated incidence in the general population is 2–4 per million per year [2]. Recurrence rates have been reported to be 30 to 40% in the major published series [3]. Surgical resection is the main choice of treatment, but the local recurrence is recognized to be unrelated to adequacy of surgical margin [4]. DTF is generally divided into two categories, abdominal and extra-abdominal. Abdominal fibromatosis is characterized by the localization in the abdominal wall, and its occurrence is closely related to reproductive women during or following pregnancy. On the other hand, extra-abdominal fibromatosis usually arises from the connective tissue within the muscle and its adjacent structure. It mainly affects the muscles of the shoulder, pelvic girdle and thigh of young adults. Head and neck, mesentery and pelvis are also affected [1]. From another point of view, DTF can be classified into two types, sporadic or familial adenomatous polyposis (FAP)-associated. FAP with DTF is called Gardner syndrome which has inactivating mutation of APC gene [5]. In contrast, up to 85% of sporadic DTF cases have mutations in the beta-catenin gene [6]. Both of the beta-catenin and APC gene mutations result in intranuclear accumulation of beta-catenin protein [7]. Although several mesenchymal tumors are included in the pathological differential diagnoses for DTF, detection of both nuclear localization of beta-catenin protein by immunohistochemistry (IHC) and beta-catenin gene mutation by gene analysis can distinguish DTF from other mesenchymal tumors. We experienced a pancreatic mesenchymal spindle cell neoplasm, and confirmed both nuclear localization of beta-catenin protein and a common beta-catenin gene mutation (T41A). These findings led us to the definite pathological diagnosis of DTF of the pancreas.
2. Case report

2.1. Clinical presentation

A 75 year-old Japanese woman with no familial history of FAP, who underwent hysterectomy for uterine leiomyoma, right hemi-colectomy for non-neoplastic disease and left hip replacement for femoral neck fracture, was pointed out an abdominal mass lesion in a routine examination by ultrasonography. The mass was located behind the stomach and adjacent to the pancreas. At that time, the mass was 5.5 cm in diameter. For precise examination, she was referred to the Department of Surgery, Rinku General Medical Center. Abdominal computed tomography (CT) showed an anteriorly protruding mass from the pancreas with low enhancement (Fig. 1A and B). Because of enlargement of the mass lesion to 8 cm in diameter for about 3 months, the operation was performed. Since the mass appeared to be encapsulated and protruding from the left-sided pancreas (mainly from the body of the pancreas), distal pancreatectomy with splenectomy was done (Fig. 1C and D). For the definite diagnosis, we performed pathological investigation, IHC and gene analyses.

3. Materials and methods

3.1. Histological and immunohistochemical examinations

Surgical materials were fixed in 10% buffered formalin and embedded in paraffin. Three-micrometer-thick sections were cut and stained with hematoxylin and eosin (H&E). IHC for pankeratin (AE1&AE3, Leica, Wetzlar, Germany, 1:200 dilution), EMA (GP1.4, Leica, 1:500), alpha-SMA (1A4, DAKO, Glostrup, Denmark, 1:500), Desmin (DE-R-II, Leica, 1:50), CD34 (QBend/10, Leica, 1:2000), CD117/c-kit (polyclonal, DAKO, 1:200), STAT6 (s-20, Santa Cruz Biotechnology, Santa Cruz, CA, 1:1000), MUC4 (8G7, Santa Cruz, 1:1000), beta-catenin (14/Beta-catenin, Becton Dickinson, Franklin Lakes, NJ, 1:400) and anaplastic lymphoma kinase (ALK) (clone 5A4, Abcam, Cambridge, UK, 1:50) was performed using Bond Polymer Refine Detection (Leica). IHCs without primary antibodies were performed as negative controls.

3.2. Mutational analysis of beta-catenin gene

Direct sequencing of PCR products using genomic DNA extracted from paraffin embedded materials was performed as
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