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Pancreatic desmoid tumor in a 4-year-old male with hemihypertrophy

Ken Saida [a,*], Osamu Miyazaki [a], Kentaro Matsuoka [b], Toshihiko Watanabe [c], Akihiro Fujino [c,d], Shunsuke Nosaka [a]

[a] Department of Radiology, National Center for Child Health and Development, Tokyo, Japan
[b] Department of Pathology, National Center for Child Health and Development, Tokyo, Japan
[c] Department of Surgery Specialties, National Center for Child Health and Development, Tokyo, Japan
[d] Department of Pediatric Surgery, Keio University School of Medicine, Tokyo, Japan

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ABSTRACT

We report the first case of a pancreatic desmoid tumor detected during follow-up for hemihypertrophy in a 4-year-old boy. Hemihypertrophy is a rare disorder in which one side of the body grows more than the other, causing asymmetry, and well-known complications include embryonal tumors. However, there has been no report of desmoid tumors in patients with hemihypertrophy, and these tumors are rare and poorly characterized in the literature, especially the cystic variant. For this patient, the lesion was diagnosed as a desmoid tumor based on immunostaining positive for beta-catenin and mutation of the beta-catenin gene (*CTNNB1*). This case suggests that desmoid tumors should be considered a possible etiology of pancreatic cystic lesions in patients with hemihypertrophy.

Hemihypertrophy, also called hemihyperplasia, is a rare disorder in which one side of the body grows more than the other, causing asymmetry [1]. The incidence of hemihypertrophy is reported to be 1 in every 13,200 to 86,000 births [2,3], and well-known complications are kidney malformation, cryptorchidism, and embryonal tumors. The overall incidence of embryonal tumors in patients with hemihypertrophy is 5.9%, and Hoyme et al. found that, amongst 168 children with isolated hemihyperplasia who were followed for 10 years, there were 6 Wilms tumors, 1 hepatoblastoma, 2 adrenal cell carcinomas, and 1 small bowel leiomyosarcoma [4].

Desmoid tumors, or musculoaponeurotic fibromatoses, are non-metastatic, locally aggressive neoplasms with a high rate of postoperative recurrence, and represent approximately 0.03% of all tumors and 3% of soft tissue tumors [5]. Pancreatic desmoid tumors are exceedingly rare, and only a few cases have been reported to date [6]. There has been no report of desmoid tumors in patients with hemihypertrophy.

1. Case report

A 4-year-old boy with hemihypertrophy was undergoing routine follow-up with periodical ultrasonography (US). The child was born normally after 39 weeks of gestation at another hospital. Two months after birth it was noted that his feet were of different lengths, and he was diagnosed with hemihypertrophy. There was no remarkable family history. The patient was referred to our department at the age of 1 year and was being followed up with semiannual US. At the age of 4 years, US revealed an asymptomatic cystic lesion in the left upper quadrant of the abdomen (Fig. 1a and b). The mass was located in the tail of the pancreas. Abdominal computed tomography (CT) showed a low density mass (Fig. 2a) and magnetic resonance imaging (MRI) showed a low intensity mass in a T1-weighted image (WI) and a high signal mass in a T2-WI (Fig. 2b and c). There was no signal abnormality on diffusion WI and the apparent diffusion coefficient (ADC) value was 3.26×10^{-3} mm^2/s, which indicated a non-malignant pattern. No distant metastasis was detected. Laboratory analysis including tumor markers showed values within the normal range (carcinoembryonic antigen, 0.6 ng/mL; alpha-fetoprotein, 2.7 ng/mL; and carbohydrate antigen 19-9, 12.1 U/mL).

* Corresponding author. Department of Radiology, National Center for Child Health and Development, 2-10-1 Okura, Setagaya-ku, Tokyo 157-8535, Japan. Tel.: +81 33 416 0181; fax: +81 33 416 2222.
E-mail address: kensai524@gmail.com (K. Saida).

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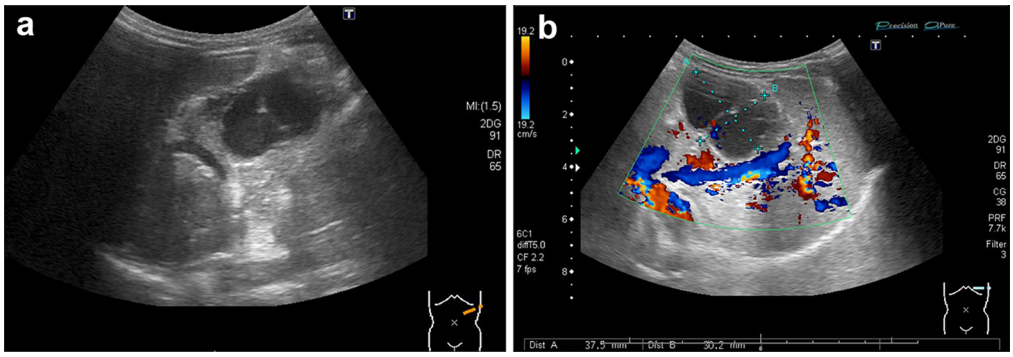

Fig. 1. (a and b) Ultrasonography (US) showing a cystic mass in the abdomen. The mass was located in the tail of the pancreas and contained a cystic component with multiple septation and a minimal solid component.

A pancreatic epidermoid cyst was suspected preoperatively and differential diagnosis included lymphangioma, solid pseudopapillary neoplasm, malignant pancreatic tumor (pancreaticoblastoma), and mucinous cystadenoma. The lesion was surgically resected by distal pancreatectomy. The resected specimen measured 4.5 × 3.0 × 2.5 cm. On the cut surface, a white solid lesion with multilocular cysts was seen in the pancreas (Fig. 3a). The lesion was well demarcated from the pancreas and had invaded the stomach wall. Histologically, the tumor was composed of proliferating spindle cells with fine cytoplasm. The cyst walls were covered with a low columnar epithelium mimicking the dilated pancreatic duct (Fig. 3b). Immunohistochemical analysis revealed that these spindle cells were partially positive for vimentin, B-cell lymphoma 2, alpha smooth muscle actin, and Muscle actin antibody (HHF35), and the nuclei of the spindle cells were positive for beta-catenin (Fig. 3c). The spindle cells were negative for S-100 protein, CD34, CD117, and pan keratin AE1/3. The MIB1 labeling index was less than 1%. A mutation in exon 3 of the beta-catenin gene (*CTNNB1*) was detected, and was predicted to encode the 41A CTNNB1 protein. Based on these histological findings and the *CTNNB1* mutation, we diagnosed the lesion as a desmoid tumor. The postoperative course was uneventful, and no recurrence has been detected 3 years after surgery.

2. Discussion

To the best of our knowledge, this is the first report of a pancreatic desmoid tumor in a patient with hemihypertrophy. Intra-abdominal desmoid tumors have been occasionally reported, although desmoid tumors in the pancreas are exceptionally rare with only 11 reported cases [6]. Abdominal desmoid tumors are

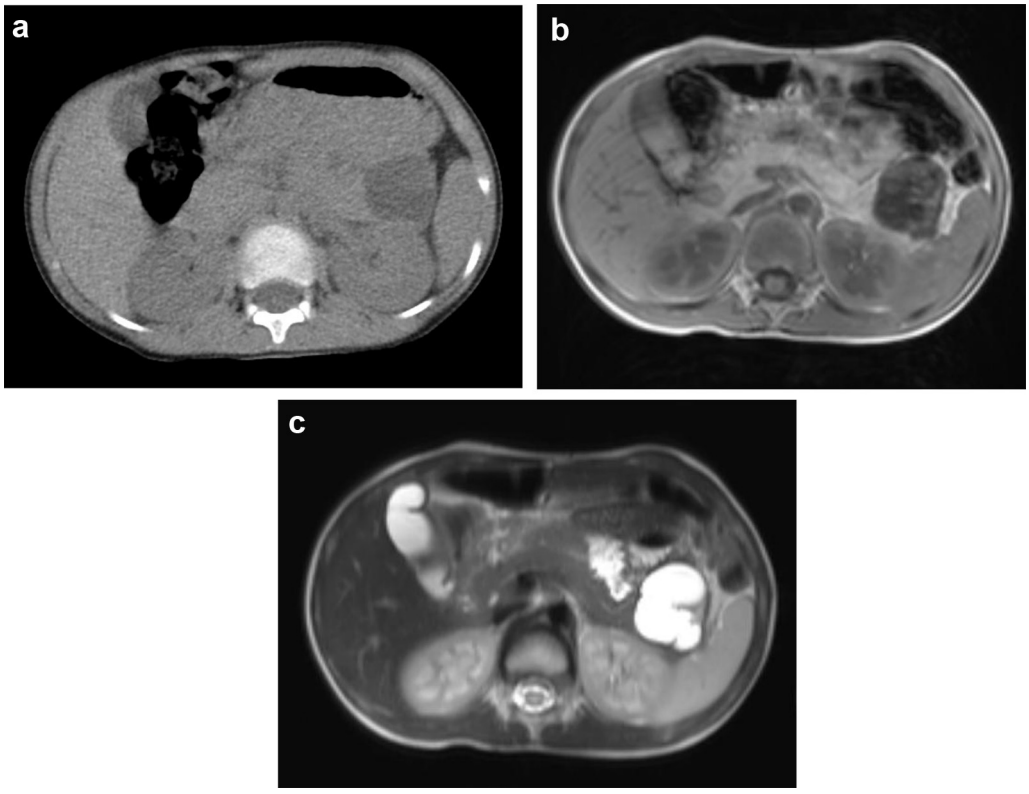

Fig. 2. (a) A plain abdominal computed tomography image revealed a low-density area at the pancreatic tail without obvious calcification and a high intensity signal on a T2-weighted image (WI). (b) Magnetic resonance imaging (MRI) showed mainly cystic masses that originated from the pancreatic tail with low signal intensity on T1-WI. (c) T2-WI showing a high intensity mass.