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

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A R T I C L E   I N F O

Article history:
Received 16 May 2015
Received in revised form 20 June 2015
Accepted 23 June 2015

Key words:
Desmoid tumor
Hemihyperplasia
Hemihypertrophy
Pancreas
CTNNB1

A B S T R A C T

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 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 mm²/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).
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 $4.5 \times 3.0 \times 2.5$ 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
more frequent in female patients and occur at any age, with a peak incidence in the third decade, and are rare in patients younger than 10 years old [7]. A pancreatic desmoid tumor in a 4-year-old boy is thus highly atypical, and the pancreas is a very rare site for desmoid tumors, especially in children [6].

Desmoid tumors, also known as musculoaponeurotic fibromatoses, are locally aggressive soft tissue neoplasms that are histologically characterized by fibroblastic proliferation within a collagen matrix. Three CTNNB1 gene variants (41A, 45F, and 45P) are known to be specific for sporadic desmoid tumors [8], and the tumor in the present case was positive for β-catenin and harbored a 41A CTNNB1 gene mutation. However, at initial presentation, a pancreatic epidermoid cyst or lymphangioma was suspected, and a desmoid tumor was not considered. Desmoid tumors do not usually show cystic features, and their appearance depends on the relative degree of fibroblast proliferation, fibrosis, collagen content, and vascularity. Although some studies have reported CT or MRI findings for these tumors [9,10], they have no specific imaging features, and it is therefore difficult to distinguish desmoid tumors from other solid masses [11]. Most cystic lesions of the pancreas are benign and the most frequent pancreatic cystic lesion is a pancreatic pseudocyst, accounting for 70–90% of such lesions, while primary cystic neoplasms account for 10–15% of cases [12].

The underlying etiology for desmoid tumor development is probably multifactorial. Hemi hypertrophy may be associated with the development of the pancreatic desmoid tumor in the present case. However, hemihypertrophy is diagnosed only on the basis of clinical manifestations, and consequently includes various diseases such as Beckwith-Wiedemann syndrome and familial adenomatous polyposis. The tumor in our case may have had a different etiology from those in other patients with hemihypertrophy.

3. Conclusion

Although preoperative diagnosis is difficult, desmoid tumors should be included in the differential diagnosis if there are pancreatic cystic findings. Mutational analysis should be performed to identify CTNNB1 gene mutations, which has both diagnostic and prognostic relevance. This case highlights the need to consider desmoid tumors as a possible etiology of pancreatic cystic lesions in patients with hemihypertrophy.

Conflict of interest and source of funding

The authors declare that they have no financial associations or any other conflicts of interest. This work was supported in part by a Grant of the National Center for Child Health and Development (25–2–1).

Acknowledgment

We would like to thank Dr. Hajime Okita for his invaluable assistance with the pathologic analysis.

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