A Case of Turner's Syndrome Complicated with Desmoid Tumor of the Transverse Colon

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Summary: The association of Turner's syndrome and endometrial carcinoma has been previously established, but has never been described in conjunction with a desmoid tumor of the colon. A case is described of a colonic desmoid tumor developing in a 38-year-old female with Turner's syndrome. The association has not been previously reported. In this report, we describe a 38-year-old woman who has Turner's syndrome with a colonic desmoid and review the literature.

Key words Turner's syndrome, desmoid tumor, gastrointestinal tumor

INTRODUCTION

Turner's syndrome is a result of a variety of abnormalities of the X chromosome. There are several reports that the risk of malignancy, both gonadal and nongonadal, is increased in patients with such chromosomal defects. The desmoid tumor is a virtually non-metastasizing tumor composed of fibrous tissue, with no apparent histologic features of malignancy but with significant potential for local invasiveness and recurrence. Presented here is a patient with Turner's syndrome who developed a desmoid tumor of the colon at 38 years of age.

CASE REPORT

A 38-year-old female patient was admitted to our hospital because of a palpable mass in the upper abdomen since that had been present for three years. She had neither abdominal pain nor gastrointestinal complaints. She had never had a menstrual period. Clinical examinations revealed a short girl who was 129.6 cm tall with scoliosis. The patient's head had no deformities, but the neck was short and mildly webbed with a low hairline in the back. Her chest was broad and shield-like with lateralization and

hypoplasia of the nipples. The external genitalia were female in character, but immature, with no growth of axillary or pubic hair. Chromosome analysis was not performed in our hospital because she had been diagnosed with Turner's syndrome by an outside gynecologist. She had been treated for a dislocation of the right hip joint and a horseshoe kidney. There was a huge abdominal mass in her abdomen which she had palpated herself 3 years previously. It was hard and was not fixed. The abdominal tumor has become larger in size without any symptoms. Laboratory data revealed no abnormal findings; serum cortisol, adrenocorticotrophic hormone, thyroxine levels were within normal range. Serum alpha fetoprotein was also at a normal level.

Abdominal computed tomography (CT) showed a large mass lesion which revealed a slightly lower attenuation than muscle tissue (Fig. 1a). Barium enema of the large intestine revealed an oppressive finding of the transverse colon (Fig. 1b).

The clinical diagnosis was Turner's syndrome with an abdominal tumor possibly arising from the transverse colon.

At laparotomy, there was a 15×12×11 cm tumor arising from the transversal colon. Her genital organs such as the uterus and ovary were very small in size

Fig. 1. a: Abdominal computed tomography showed a large mass lesion in the abdominal cavity. b: Barium enema of the large intestine revealed an oppressive finding of the transverse colon.

Fig. 2. a: Cut surface of the resected specimen showed a large solid tumor with no areas of necrosis or bleeding. b: Histopathological finding was desmoid tumor of the colon. The tumor cells are composed of a proliferation of elongated slender spindle cells with minimal atypia and bundles of collagen between them. (H. & E. $\times 50$)

and atrophic. The mass was resected combined with partial transversal colectomy. Macroscopically, the resected specimen showed a large solid tumor without necrosis or bleeding (Fig. 2a). Pathological diagnosis was intraabdominal fibromatosis (intraabdominal desmoid). Microscopically, the ill-defined tumor was composed of a proliferation of elongated slender spindle cells with minimal atypia and bundles of collagen between them, arranged in sweeping fascicles, involving the colonic wall. On immunohistochemical study, some of the spindle cells were positive for actins (HHF35, alpha-smooth muscle actin), while desmin, CD34, c-kit and S-100 protein were negative. Although relatively hypercellular areas and occasional mitotic figures were seen, there was no evidence of malignancy (Fig. 2b). Her postoperative course was uneventful and she has no recurrent tumor at this time.

DISCUSSION

Turner's syndrome is the result of a variety of abnormalities of the X chromosome, varying from complete absence to a 45, XO karyotype to minimal alterations such as mosaicism. The typical features in a patient with Turner's syndrome are short stature, infantilism, streak gonads with several associated congenital malformations. The etiology of the chromosomal aberration could be due to nondisjunction of the sex chromosome or deletion of varying portions of the X or Y chromosome.

The structural abnormalities of the internal organs include coarctation of the aorta, pulmonary stenosis, renal abnormalities, such as the horseshoe kidney in our case, rotation of the kidney, duplication of the

TABLE 1.
Reported cases of Turner's syndrome associated with gastrointestinal neoplasm

No	Author	Year	Age	Organs	Histologic type	Reference
1	Sternlieb	'54	38	Colon	Adenocarcinoma	N Y J Med 54:1663-1665
2	Engel	'65	44	Colon	Adenocarcinoma	Medicine 44:135-164
3	Wertelecki	'70	13	Cecum	Carcinoid	Cancer 26:485-488
4	Wertelecki	'70	38	Sigma	Adenocarcinoma	
5	Wertelecki	'70	44	Sigma	Adenocarcinoma	
6	Wertelecki	'70	38	Sigma	Adenocarcinoma	
7	Herrera-Ornelas	'84	43	Colon	Adenocarcinoma	J Surg Oncol 27:251-254
8	Cheng	'93	49	Colon	Adenocarcinoma	J Formos Med Assoc 92:580-582
9	Present case	'99	39	Colon	Desmoid tumor	
10	Pich	'37	49	Stomach	Adenocarcinoma	Beitr Pathol Anat 98:218-263
11	Engel	'65	63	Stomach	Adenocarcinoma	Medicine 44:135-164
12	Wertelecki	'70	63	Stomach	Adenocarcinoma	Cancer 26:458-488
13	Wertelecki	'70	49	Stomach	Adenocarcinoma	
14	Siegler	'75	34	Stomach	Adenocarcinoma	Postgrad Med J 51:411-412
15	Cayla	'84	64	Stomach	Adenocarcinoma	Rev Rhum 51:281-282
16	Yata	'94	64	Stomach	Leiomyosarcoma	Jpn J Gastroenterol 91:1328-1332

renal pelvis and ureter, and rarely gastro-intestinal telangiectasia and Hirschsprung's disease [1,2]. As for the high percentage of tumors among cases of Turner's syndrome, it is known that dysgenetic gonads give rise to neoplastic change [3]. This is related to the presence of Y chromosomal components [3-5]. The most common tumor which arises from the dysgenetic gonad is gonadoblastoma [5]. According to the report on a multi-hospital survey of 289 cases by Wertelcki et al. [6], 8 patients were found to have nongonadal tumors. Thus, gastro-intestinal tumors associated with Turner's syndrome are relatively rare, especially in the form of colonic desmoids (Table 1). There were 16 cases of gastro-intestinal tumor, including our case. Seven of them were colonic cancer and only one case was carcinoid tumor of the cecum. Six of 7 gastric malignant tumors were gastric cancer, and leiomyosarcoma occurred in 1 patient. Patients ranged in age from 13 to 64 years (mean, 45 years).

Intraabdominal desmoid tumors are infrequently encountered in surgical practice and, when seen, are most often genetically determined tumors associated with Gardner's syndrome or familial adenomatous polyposis syndrome (FAP).

Trauma and endocrine and genetic factors seem to be implicated in the pathogenesis of desmoid tumors; however, the exact pathogenesis is unclear [7].

The mortality associated with desmoid tumors was reported to be approximately 11% of all deaths in patients with FAP and 31% of deaths after colectomy, making it the second most common cause of death once colorectal cancer is excluded [8]. Therefore, alternative treatment strategies, including radiation therapy [9-11], nonsteroidal antiinflammatory agents [12], antiestrogen compounds [13-17], and cytotoxic chemotherapy [18,19] have been employed.

Since metastasis almost never occurs, aggressive surgical removal of the tumors is the first treatment of choice. A surgical approach aiming at clear margins is presently the best treatment option. When this cannot be accomplished without severe disfigurement or functional impairment, partial resection is an acceptable alternative. Adjuvant strategies, preoperatively and/or after partial resection, should be considered in the context of an actively growing tumor with potential for mortality or significant morbidity.

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