

CASE REPORT

Adenocarcinoma and neuroendocrine tumor arising within presacral teratoma associated with Currarino syndrome: A case report

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Abstract

We present a case of a 59-year-old woman with a malignant tumor arising within presacral teratoma associated with Currarino syndrome (CS). A characteristic crescent-shaped sacrum was detected on preoperative image examination and the presacral mass was pathologically diagnosed as a malignant tumor associated with CS. To our knowledge, this is the first case report of presacral teratoma associated with CS coexisting with both adenocarcinoma and a neuroendocrine tumor.

Key words: Adenocarcinoma; Currarino syndrome; neuroendocrine tumor; teratoma

Introduction

Currarino syndrome (CS) is a hereditary disorder characterized by the classical triad of sacral agenesis, anorectal malformation, and a presacral mass. It was first described by Currarino *et al.* and considered the result of abnormal endoectodermal adhesions and notochordal defects in early fetal life.^[1] The phenotypic expression of CS is variable.^[2-4] Some patients are asymptomatic and incidentally diagnosed late in life, whereas the illness is fatal in infancy in other cases because of severe complications such as bacterial meningitis and urinary tract infections. Malignancy in CS is rare, with a reported frequency of approximately 1%.^[2,5] Concerning malignancy in adults with CS, only six cases have been

described to date [Table 1].^[6-11] This case report is the first documented instance of presacral teratoma associated with CS coexisting with both adenocarcinoma and a neuroendocrine tumor.

Case Report

A 59-year-old Japanese woman with hypertension and diabetes mellitus was admitted to our hospital to undergo pelvic surgery. She had a presacral mass that was incidentally detected on CT before cholecystectomy for cholelithiasis, and adenocarcinoma was detected in the mass via endoscopic ultrasound-guided fine needle aspiration

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through the rectum in a prior hospital. Complete blood count, liver function, basic metabolic panel, and urinalysis were unremarkable. CEA, CA19-9, and CA125 were within their normal ranges.

A pelvic radiograph revealed a scimitar sacrum with a right-sided defect [Figure 1]. Contrast-enhanced CT revealed two presacral masses in the pelvis. The upper mass had cystic and solid components containing subtle calcification, and the second mass was a simple cyst through an enlarged neural foramen [Figure 2]. Pelvic MRI illustrated the upper mass with cystic and solid components with various intensities on T1- and T2-weighted images (T2WI). The other presacral cystic lesion displayed high intensity on T2WI, and it had no mural nodule, which was suggestive of an anterior meningocele [Figure 3]. The patient was diagnosed with a malignant tumor associated with CS preoperatively, and



Figure 1: Pelvic X-ray revealed a scimitar sacrum with a right-sided defect

she underwent surgical resection of the presacral mass in the pelvis. The tumor was completely resected, although the lower cystic lesion was accidentally ruptured because of adhesion.

Pathologic examination confirmed that the cystic and solid tumor contained adenocarcinoma and neuroendocrine tumor (NET) components (G2) [Figure 4]. The resected tumor included many cysts covered by stratified columnar epithelium, smooth muscle tissues, and a glomus tumor. Subtle fat tissue was present in the resected tumor, although it was not detected on preoperative MRI. Histologically, the tumor contained several different tissue types, suggesting a teratomatous nature. Most cysts inside the tumor that exhibited varied intensity on MRI included necrotic tissues. Adenocarcinoma was found in the cyst-like structure, and it was not distinguishable from other cysts on preoperative image examinations [Figure 5]. The adenocarcinoma component exhibited ductal and papillary structures, and a diagnosis of moderately differentiated adenocarcinoma was rendered. In the NET component, which was close to the adenocarcinoma lesion, immunohistochemical staining was positive for synaptophysin and chromogranin A. Residual NET tissue was observed along the surgical margins, and postoperative adjuvant radiotherapy (50.4 Gy/28 fr) was administered without adjuvant chemotherapy. After 8 months of follow-up, contrast-enhanced CT and pelvic MRI revealed no tumor recurrence, but genetic testing has not been performed in line with the patient's preference.

Discussion

CS is a rare autosomal dominant disorder, and mutations in the coding sequence of HLXB9 located at 7q36 have been identified in nearly all cases of familial CS, as well as approximately 30% of patients with sporadic CS.^[2] Sacral defects in CS vary from lateral deviation of the coccygeal bone to a hemisacrum or the absence of the lower sacral segments.^[2-4] Presacral masses contain anterior myelomeningocele, malignant or benign teratoma, dermoid or epidermoid cysts, leiomyoma, leiomyosarcoma, lipoma, or combinations of



Figure 2: Contrast-enhanced CT revealed two presacral masses in the pelvis. One mass had cystic and solid components containing subtle calcification (arrowhead). The other mass was a cystic lesion through an enlarged neural foramen (arrow)

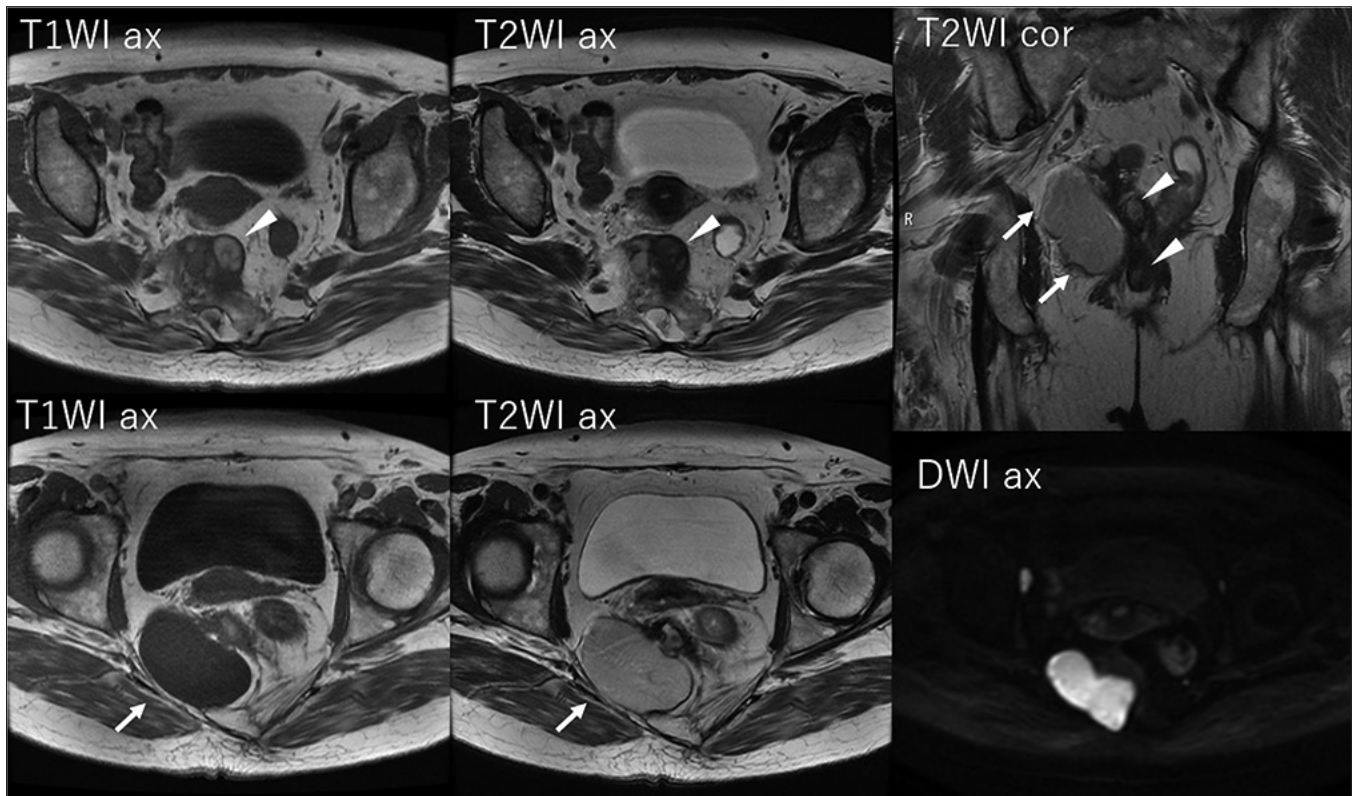


Figure 3: Pelvic MRI revealed cystic and solid components with various intensities on T1- and T2-weighted images (arrowheads). The other presacral cystic lesion exhibited high intensity on T2-weighted images, and it featured no mural nodule (arrows)

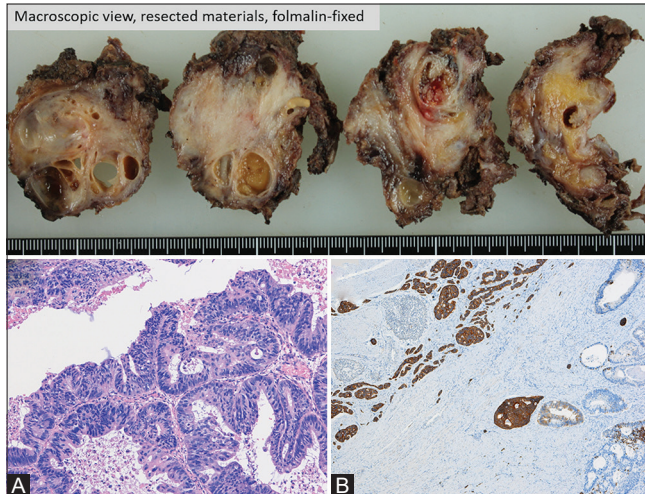


Figure 4 (A and B): (A) Moderately differentiated adenocarcinoma, H and E stain. (B) Neuroendocrine tumor, synaptophysin stain. The histological presence of a neuroendocrine tumor, smooth muscle tissue, cysts covered by stratified squamous epithelium, and glomus tumor suggested a teratomatous nature (not shown)

these lesions. More than 80% of patients with the classic triad are diagnosed within the first decade of life because of the symptoms of anorectal malformation.^[12] However, one-third of patients are asymptomatic at the time of diagnosis.^[2] The combination of all three defects is not necessary for a diagnosis of CS because the clinical presentation of the syndrome is variable. Although genetic tests have not been performed, the

present case featured a hemisacrum and a presacral mass with anterior meningocele, and no anorectal malformation was present, suggesting the mild type of CS in the classification by Martucciello *et al.*^[14] Therefore, the present case was diagnosed incidentally at older age than typical cases.

Malignancy in CS is rare. Only a small number of patients of CS with malignancy have been reported. The risk of malignant transformation of presacral teratomas was estimated in previous reports at around 1% of CS cases.^[2,5] On top of that, as for malignancy in adults with CS, only six cases have been described until now [Table 1].^[6-11] The median age of diagnosis for CS with malignancy is 38 years (range, 22–59). Among the previously reported cases, HLXB9 mutations were detected in two cases, and the remaining cases were diagnosed as CS clinically. NETs from presacral teratoma were reported in three cases, and malignant tumors were associated with presacral teratoma in all but one case of leiomyosarcoma. Surgical resection of a presacral mass carries risks of severe complications related to adhesion of the anterior meningocele such as bacterial meningitis and cerebrospinal fluid leakage.^[13] In our case, although the anterior meningocele was accidentally ruptured during surgery, no severe postoperative complication occurred.

In general, somatic malignant transformation of mature cystic teratoma is uncommon, and the risk of transformation is estimated at 1%–3%.^[14,15] Although any tissue component of a

Table 1: Malignancy in adults with Currarino syndrome

Author, year	Age, sex	CS diagnosis	Malignant tumor	Treatment
Ashcraft <i>et al.</i> , 1974	32, female	Clinical	Malignant presacral teratoma	Not described
Yates <i>et al.</i> , 1983	54, male	Clinical	Malignant presacral teratoma	Surgery
Norum <i>et al.</i> , 1991	59, female	Clinical	Presacral leiomyosarcoma	Surgery, radiotherapy
Urioste <i>et al.</i> , 2004	22, male	HLXB9 mutation	Neuroendocrine tumor from presacral teratoma	Surgery, chemotherapy
Pendlimari <i>et al.</i> , 2010	22, female	Clinical	Neuroendocrine tumor from presacral teratoma	Surgery
Ciotti <i>et al.</i> , 2011	44, female	HLXB9 mutation	Neuroendocrine tumor from presacral teratoma	Surgery, chemotherapy
Present case	59, female	Clinical	Adenocarcinoma and neuroendocrine tumor from presacral teratoma	Surgery, radiotherapy

Table 2: Clinical characteristics of adenocarcinoma and neuroendocrine tumor arising from mature cystic teratoma

Author, year	Age, sex	Symptoms	Lesion site	Treatment	Follow-up
Kim <i>et al.</i> , 2003	62, female	Lower abdominal pain	Ovary	Hysterectomy with BSO, chemotherapy, radiotherapy	Alive without evidence of disease at 13 months after surgery
Armah <i>et al.</i> , 2009	50, female	Low back and right hip pain	Horseshoe kidney	Partial nephrectomy	Alive without evidence of disease at 6 months after surgery
Hinshaw <i>et al.</i> , 2012	74, female	Incidental finding on CT	Ovary	Hysterectomy with BSO	Not described
Agarwal <i>et al.</i> , 2015	24, female	Chest pain	Mediastinum	Surgical resection	Alive without evidence of disease at 1 month after surgery
Higgins <i>et al.</i> , 2017	66, male	Epigastric pain	Horseshoe kidney	Partial nephrectomy, chemotherapy	Died 7 months after surgery from metastatic disease
Present case	59, female	Incidental finding on CT	Presacral teratoma associated with CS	Surgical resection, radiotherapy	Alive without evidence of disease at 6 months after surgery

BSO=Bilateral salpingo-oophorectomy; CS=Currarino syndrome

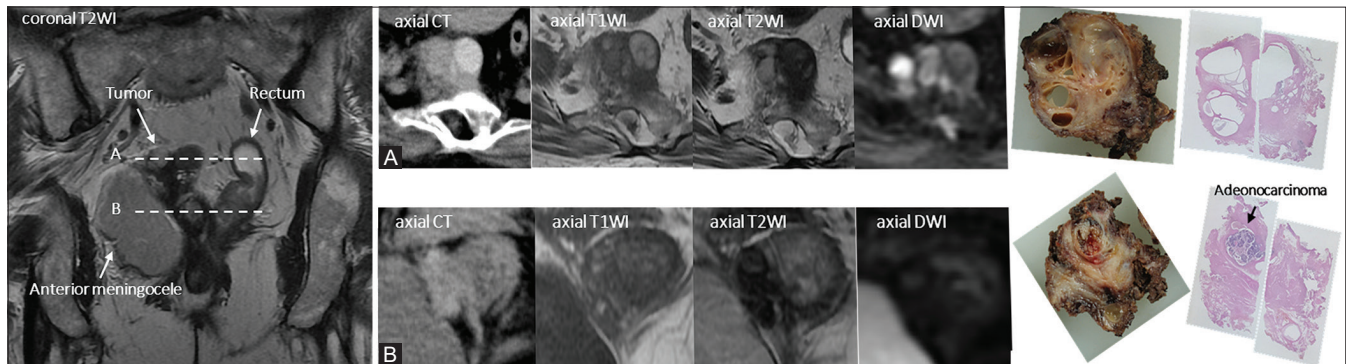


Figure 5 (A and B): (A and B) The presacral tumor near the rectum contained cysts with varied intensity on MRI. One of these cysts included adenocarcinoma

teratoma can potentially undergo malignant transformation, squamous cell carcinoma is the most commonly associated malignancy. Other reported malignant tumors arising within a mature cystic teratoma include adenocarcinoma, basal cell carcinoma, adenosquamous carcinoma, thyroid carcinoma, sebaceous carcinoma, malignant melanoma, sarcoma, carcinoid tumor (NET), and neuroectodermal tumor.^[16,17] A synchronous primary neuroendocrine tumor and primary adenocarcinoma arising within mature cystic teratoma is extremely rare, and only five cases have been reported [Table 2].^[18-22] To the best of our knowledge, no prior report described a mature cystic teratoma in CS coexisting with both adenocarcinoma and NET.

Conclusion

CS is a rare congenital disease with variable phenotypic expression, thereby producing a range of symptoms. The

occurrence of synchronous primary adenocarcinoma and NET arising within a teratoma is rare. The present patient was diagnosed with CS incidentally in middle age, and this is the first reported case to our knowledge of a mature cystic teratoma coexisting with adenocarcinoma and NET in a patient with CS.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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