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A case of 45, XO/46 X, dic (Y) (qter-p11:: p11-qter) mosaicism in Turner syndrome with mixed germ cell tumour

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Abstract

We report a case of a mixed germ cell tumor of the ovary in a patient with 45, XO/46, X, dic (Y)(qter-p11:: p11-qter) mosaicism. A 24-year-old girl was admitted to our Hospital because of bloating and abdominal pain for more than half a month. Physical examination showed short stature and webbed neck. Her lower abdomen was bulging. Chromosomal analysis showed 45, XO/46, X, dic (Y) (qter-p11:: p11-qter) mosaicism in karyotype ten years ago. Chemical examination found that the values of α-fetoprotein and CA-125 were elevated. CT scan of the whole abdomen and pelvis showed a large cystic solid mass in the lower abdomen and pelvis; bilateral ovaries and uterus were not visible. Pathological examination showed a mixed germ cell tumor. After operation, chemotherapy using bleomycin- etoposide-cisplatin was performed and the tumor marker went down. However, the a-Fetoprotein in the serum elevated again. CT scan of the abdomen showed a new mass in the right lobe of the liver. After 3 courses of Kealil-Cisplatin-Ancoda chemotherapy, the liver mass was resected and diagnosed as metastatic germ-cell tumor of the liver with yolk sac tumor component. The patient died at 6 months since the diagnosis of liver metastasis of tumor.

Keywords: Turner syndrome; 45; XO/46; X; dic (Y) (qter-p11:: p11-qter); Mixed germ cell tumor.

Introduction

Turner Syndrome (TS) refers to the abnormalities of growth and development, reproductive system, cardiovascular system and endocrine system caused by the complete or partial deletion of an X chromosome in female patients. Most of the TS patients have 45, X(32.7%) or 45, X/46, XX and other chimeric karyotypes (43%), and a small number of patients (8%-12%) have Y chromosome components [1,3]. The chimeric dicentromere Y chromosome is even rarer.

Clinic summary

The patient was 15year-old and had not yet menstruated and ultrasonic examination revealed a rudimentary uterus. The chromosomal analysis from peripheral blood lymphocytes revealed 45, XO(48%)/46, X, dic (Y) (qter-p11:: p11-qter) (52%), and she was diagnosed as Turner syndrome and admitted to our Hospital in February 2022 because bloating, abdominal pain and abdominal bulging were present. She was 150 cm in height and weighed 49.2 kg, had a webbed neck and poorly developed breasts but normal intelligence. No congenital cardiopathy was found. The external genitalia were female, but pubic hair was scant and the labia were underdeveloped. Her uterus and cervix couldn't be touched. Computerized Tomography (CT) revealed a huge cystic-solid mass within the lower abdomen and pelvic cavity. The mass invaded the sigmoid colon and left ureter, several enlarged lymph nodes also could be seen. CT scan also

showed the suspicious uterine dysplasia and bilateral unclear ovaries. Ultrasonic cardiogram was normal.Serum α-fetoprotein (AFP) and CA-125 concentrations were 597,195.94 ug/L (normal range: 0~25 ug/l) and 91.10 U/ml (normal range: 0~35 U/ml) before operation, respectively.

Laparotomy revealed a huge mass about 20 cm in diameter with rich vessels and connected with two fallopian tubes within the pelvic cavity and lower abdomen. The mass formed dense adhesion with part of the colorectal, abdominal wall, pelvic wall and peritoneum on the surface of the bladder and several enlarged lymph nodes were found. The immature uterus also could be seen. The mass, enlarged lymph nodes, bilateral fallopian tubes, uterus, partial rectum and omentum majus were removed.

Pathological findings: Grossly, tumor of pelvis appeared greyish-red with haemorrhage and necrosis. Microscopically, most of tumor presented coagulation necrosis; the surviving tumor mainly had a reticular/microcystic pattern, soild pattern and glandular pattern, tumor cells showed variable atypia. Schiller-Duval bodies could be found. Immunohistochemistry staining showed the tumor cells were positive for Glypican-3 but negative for OCT3/4. So the tumor was diagnosed as yolk sac tumor. Within an enlarged lymph node, the above-mentioned lesion also could be seen, and tumor cells were positive for CK, Glypican-3, AFP and SALL4, but negative for D2-40, OCT3/4 and CD117 (Figure 1). Whereas another area of tumor exhibited that the sheets and nests of monotonous tumor cells were separated by thin fibrous septa containing abundant lymphocytes and the nuclei of tumor cells exhibit angular or squared-off contours and prominent nucleoli besides above lesion. The tumor cells were positive for D2-40, OCT3/4, AFP, SALL4 and CD117 and negative for CK, Glypican-3. So the pathological diagnosis was a mixed germ cell tumor (60% yolk sac tumor and 40% dysgerminoma). The yolk sac tumor cells involved the uterus and bilateral fallopian tubes.

After the operation, the patient received a total of 6 courses of bleomycin-etoposide-cisplatin chemotherapy for 5 months. The serum CA-125 levels decreased to 9.10 U/ml which was within the normal range. The serum AFP levels fell to 1344.84 ug/l which was still high.

The serum AFP levels elevated to 2165.20 ug/l in July 2022. CT scan of abdomen showed a new mixed density mass in the right lobe of the liver, the size was 57 mm × 53 mm × 51 mm, the mass was highly suspected to be metastatic tumor of liver. After 3 courses of paclitaxel-cisplatin-bevacizumab chemotherapy, the liver mass was resected. Pathological examination showed there were several tumor nodules in the liver tissue. The tumor cells were arranged in a reticular /microcystic and soild patterns. It was confirmed by immunohistochemical examination that the lesions were liver metastatic germ cell tumor and the metastatic component was yolk sac tumor (Figure 2). The patient died of tumor at 6 months since the diagnosis of liver metastasis of tumor.

Discussion

Women with TS have an increased risk of basal cell carcinoma, colon and rectal cancer and meningioma. Furthermore, the present of Y-chromosome component will significantly increase

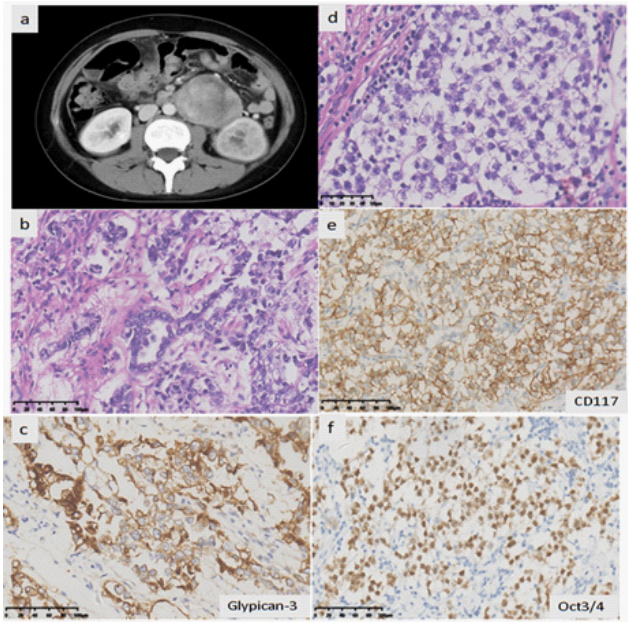

Figure 1: The CT scan, HE and immunohistochemical manifestations of the mass. **(a)** The CT scan showed a large cystic-solid mass in the lower abdomen and pelvic cavity. **(b-c)** One area of tumor was Yolk sac tumor. The tumor consisted of variable atypia cells which had a reticular /microcystic pattern. Tumor cells were positive for Glypican-3 (×200). **(d-f)** Another area was dysgerminoma. Sheets and nests of monotonous tumor cells were separated by thin fibrous septa containing abundant lymphocytes, the nuclei of tumor cells exhibit angular or squared-off contours and prominent nucleoli. Tumor cells were positive for CD117 and OCT3/4 (×200).

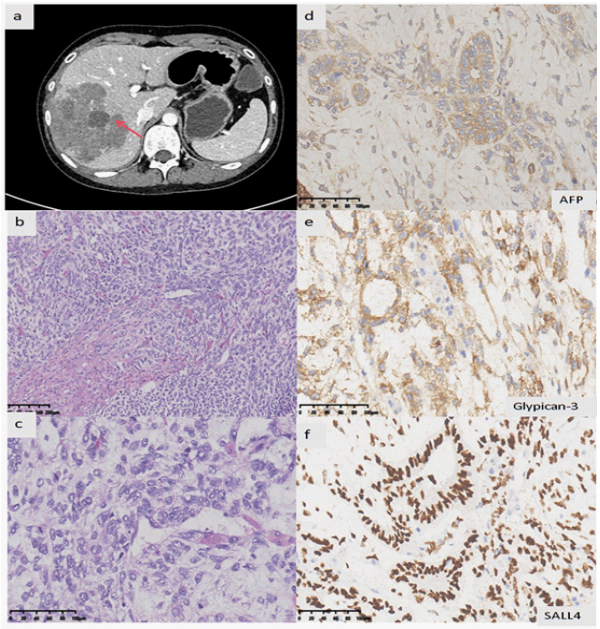

Figure 2: The CT scan showed a liver nodule and HE and immunohistochemical features of the nodule. **(a)** Abdominal CT scan showed a mixed density mass in the right lobe of the liver. **(b-f)** Tumor showed reticular /microcystic and soild patterns at low (×100) and high (×200) magnification power, and the tumor cells were positive for AFP, Glypican-3, and SALL4 (×200).

the risk of gonadoblastoma, a benign neoplasm, and approximately 3% of which will transform into malignant tumor, such as dysgerminoma, embryonal carcinoma, choriocarcinoma and yolk sac tumor [4,5]. There was only one case of 45, XO/46, X, dic (Y) karyotype combined with ovarian yolk sac tumor [6]. We first report 45, XO/46, X, dic (Y) (qter-p11:: p11-qter) in Turner Syndrome with mixed yolk sac tumor and dysgerminoma.

Declarations

Competing interests: None declared.

Ethics approval statement: The approval for this study was obtained the Ethics Committee of the First Affiliated Hospital, Sun Yat-sen University.

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