Case Report

Dysgerminoma: diagnostic impasse

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ABSTRACT

Dysgerminoma is a rare malignant ovarian tumour in women of reproductive age group, featuring lower abdominal pain and abdominal mass with elevated tumour markers. The tumour grows rapidly and diagnosed at an earlier stage. We present a short-statured 20 years girl with intermittent abdominal pain and distention for 4 months, on examination huge abdominal mass of size 20x15 cm, firm in consistency was palpable, lower limit not ascertained. Routine blood investigations normal, thyroid function test showed hypothyroidism. A plain abdominal radiograph shows a large homogenous mass fitting mid part of abdomen with displaced bowel loop. Contrast-enhanced computed tomography abdomen and pelvis show up 20x18.5x9.5 cm well defined heterogeneous mass, with areas of necrosis suggesting mesenchymal tumour or germ cell tumour. CA 125, alpha-fetoprotein, lactate dehydrogenase, beta human chorionic gonadotropin were raised. Magnetic resonance imaging abdomen and pelvis show 10x16x17cm heterointense lesion, bilateral ovary normal. After tumour board discussion image-guided biopsy was done, suggestive of epithelioid gastrointestinal stromal tumour. Laparotomy displayed a large mass 22x16x10 cm arising from left ovary, no enlarged lymph node, completed with left salpingo-oophorectomy. Histopathology examination and immunohistochemistry definitive of dysgerminoma. In this clinical scenario, we narrate the importance of clinical examination and increased dependence on imaging modalities in diagnosing the patient. The treatment is based on the international federation of gynaecology and obstetrics staging with surgical treatment, adjuvant chemotherapy and radiotherapy.

Keywords: Dysgerminoma, GIST, Malignant ovarian tumour, Huge abdominal mass

INTRODUCTION

Germinoma originates from germ cells found in the ovary, pineal region and mediastinum.1 Dysgerminoma is one of the most common malignant ovarian tumours, arises from undifferentiated primordial germ cells in the ovary. It compromises 1-2% of all malignant ovarian tumours, occurs mainly in the second and third decade.2,3 Generally, unilateral disease also bilateral in 10-15% of cases. Clinical features include abdominal pain and distention, ascites and vaginal bleeding. The tumour grows rapidly and diagnosed at an earlier stage. In old age patients usually diagnosed at a later stage. Serum levels of lactate dehydrogenase (LDH), beta human chorionic gonadotropin (HCG), Ca-125, AFP are usually raised. If the tumour is too large, it is important to take biopsy and treatment is either surgical resection leading to conservative surgery or radical procedure with post-operative chemoradiation.

CASE REPORT

A 20 years old girl with intermittent vague abdominal pain for 6 months, which increased intensity for the past one week presented to our emergency room. History of abdominal distention for 4 months, no history of nausea,
vomiting and obstipation. She was short-statured with developmental delay; she has not attained menarche with no prior significant gynaecological history. On examination, tense distended abdomen with a huge abdominal mass of size 20x15 cm, firm in consistency was palpable, whose lower limit was not ascertained.

Routine blood investigations were within normal limits, TFT showed hypothyroidism. Karyotyping revealed a normal female; 46XX and Swyer syndrome was ruled out. A plain abdominal radiograph shows a large homogenous mass fitting mid part of abdomen with displaced bowel loop. Contrast-enhanced computed tomography (CECT) abdomen and pelvis show up 20x18.5x9.5 cm well defined heterogeneously enhancing mass, below greater omentum to bladder dome, displacing bowel laterally, with few areas of necrosis suggesting mesenchymal tumour or germ cell tumour (Figure1). Ascitic fluid cytology showed inflammatory pathology. With raised tumour markers Ca-125; 275 (30.5 U/ml), AFP; 36.9 (0.5-5.5 IU/ml), LDH; >700 (120-246 U/L), beta HCG; 468.73 (<10 mIU/ml) but magnetic resonance imaging (MRI) abdomen and pelvis show 10x16x17cm hetero intense lesion noted in the abdominal cavity, bilateral ovary normal, possibility of mesenchymal tumour (Figure 2).

Tumour board discussion concurred image-guided biopsy, biopsy resulted as a polyhedral cell with pleomorphic nuclei possibility of poorly differentiated carcinoma, (mesenchymal tumour) with immunohistochemistry (IHC) study CD-117 positive suggestive of epithelioid gastro-intestinal stromal tumour (GIST) (Figure 3).

After pre-operative optimisation with L-thyroxine, the patient was taken for elective laparotomy, on proceeding shows a large mass 22x16x10 cm arising from left ovary, no enlarged paraaortic and retroperitoneal lymph nodes were appreciated. Omentum was unremarkable, no abnormality or nodularity in any other intraabdominal organs. Uterus and right ovary found hypoplastic, completed with left salpingo-oophorectomy (Figure 4 and Figure 5).

Figure 1: Heterogeneous mass, suggesting mesenchymal tumour or germ cell tumour.

Figure 2: Hetero intense lesion noted in the abdominal cavity, possibility of mesenchymal tumour.

Figure 4: Intraoperative findings of a large mass arising from left ovary.

Figure 5: Resected left ovarian mass.

Figure 3: Image guided biopsy, IHC study CD-117 positive.
The resected specimen on gross was nodular and bosselated, the cut surface shows greyish white fleshy areas with no evidence of necrosis and haemorrhage. Histopathology examination (HPE) showed sheets and clusters of polyhedral cells with moderate eosinophilic cytoplasm, vesicular nuclei and prominent nucleoli. Immunohistochemistry showed Oct 3; negative, CD 30; negative, placental alkaline phosphatase (PLAP); 80% of cell membrane positive, definitive of dysgerminoma (Figure 6).

**DISCUSSION**

Ovarian tumours are classified into three broad types depending on their cell type of origin namely epithelial, sex cord, germ cell tumours. Germ cell tumours are further classified into dysgerminoma, immature teratoma, endodermal sinus (yolk sac) tumour, mixed germ cell tumour, embryonal carcinoma, polyembryoma and choriocarcinoma. Germ cell tumours form 30% of all ovarian neoplasm and 3% of all malignant ovarian tumours. Dysgerminoma compromises 1-2% of all malignant ovarian tumours. It is a counterpart of seminoma in males. It is one of the most common ovarian malignancy to occur during pregnancy since 10-20% of dysgerminoma is diagnosed during pregnancy. Dysgerminoma has a greater predilection to occur in females with chromosomal abnormalities such as 46 XY (swyer/testicular feminisation syndrome) and 45/46 XY mosaicism.

Usually, a young female presents with features of abdominal pain, pelvic or abdominal mass and distension with or without loss of weight and appetite. Some have also reported lower abdominal pain with recurrent culture-negative urinary tract infection. These patients can be easily overlooked as a case of acute appendicitis or ectopic pregnancy in an emergency room.

Ultrasound shows large solid multilobulated heterogeneous mass separated from the uterus. Exhibit hypoechoic fibrous septa or anechoic areas of necrosis and haemorrhage. CECT and MRI abdomen and pelvis shows well defined heterogeneously enhancing mass, with its extension and relation to other intraabdominal organs. Generally, dysgerminomas are well vascularised and colour doppler demonstrate flow in hypoechoic septa. Ascitic fluid can be made out in cul de sac. Quantification of tumour markers such as LDH, Beta HCG, Ca-125, alpha-fetoprotein at diagnosis and useful in the follow-up.

In this case after tumour board discussion, ultrasound-guided biopsy resulted in CD-117 (c-kit) positivity which was suggestive of GIST. But HPE and IHC examination of resected specimen proved it to be a dysgerminoma with PLAP positivity. It also expresses c-kit, Oct 3/4 immunoreactivities. It has been reported that 87% of dysgerminoma has shown c-kit positivity. C-kit is expressed in a wide variety of tumours such as gastrointestinal tumours, chronic myeloid leukaemia (CML), seminoma, adenoid cystic carcinoma and malignant melanoma.

Treatment of dysgerminoma is based on the International federation of gynaecology and obstetrics (FIGO) staging. Stage Ia, unilateral, encapsulated, the unruptured tumour is proceeded with conservative unilateral salpingo-oophorectomy, for stage Ib it is bilateral salpingo-oophorectomy with or without total abdominal hysterectomy followed by 3 cycles of BEP protocol (bleomycin, etoposide, cisplatin), stages II, III, IV are managed by the radical surgical procedure followed by 4 cycles of chemotherapy. Tumour usually spread through the lymphatic system into paraaortic lymph nodes. Haematogenous spread occurs in later stages of disease.

**CONCLUSION**

Dysgerminoma is peculiar in characteristics from other ovarian tumours, making accurate diagnosis vital for patient care. There is an increase in reliance on imaging modalities for the diagnosis of tumours, but it may also lead to pitfall in some cases. In our case, even though CECT was suggestive of germ cell tumour but MRI being a better investigation suggested mesenchymal tumour with bilateral ovaries normal and image-guided biopsy showed c-kit expression suggesting epithelioid GIST and both turned out to be false. Adjuvant chemotherapy show profitable outcome and thanks to the emergence and success of c-kit directed therapies in CML and GIST, c-kit expression in dysgerminoma can be targeted to provide new conservative fertility-preserving therapies.

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**REFERENCES**
