

Swyer Syndrome with Gonadoblastoma: A Rarity of Two Case Reports

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Introduction

Pure gonadal dysgenesis describes conditions with normal sets of sex chromosomes (46XX, 46XY). 46XX pure gonadal dysgenesis also termed as Swyer syndrome. First described by doctor Swyer in 1955. The incidence of Swyer syndrome is 1:100.000 [1]. They are phenotypically females with unambiguously female genital appearance from birth and normal Müllerian structures. The patients usually presents at puberty with primary amenorrhoea. Germ line tumors (gonadoblastoma, dysgerminoma, seminoma) can be found in one third of cases with 10% risk of metastasis [2]. Here, we report two cases of dysgerminoma in patients with Swyer syndrome.

Case Report

Patient X, 20 years presented to us with primary amenorrhoea and absent secondary sexual characteristics. There was no history of cyclical pain abdomen. On examination she was tall, thin built with height-163 cm and weight-42 kg. Breast and pubic hair were Tanner stage I-II. On per abdomen examination there was no mass palpable. Local examination revealed normal external genitalia but with blind vagina of 4 cm. A firm mass of 5x6 cm was felt anteriorly, on per rectal examination which was non tender and mobile.

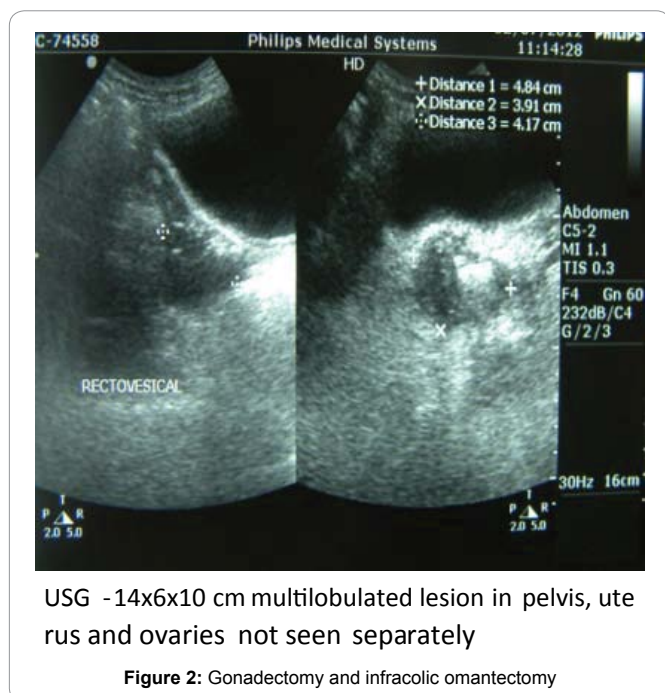
On ultrasound a 7x5 cm gonadal mass was seen but uterus and ovaries could not be visualized. Contrast CT revealed a well differentiated solid-cystic lesion of 6x9x9 cm extending into the right adnexal region, uterus and ovaries were not seen separately. All the tumor markers [α fetoprotein (1.08), CA-125 (7.9) and β Human Chorionic Gonadotrophin (1.6)] were negative. Hormone profile suggested a hypergonadotrophic hypogonadism profile with Follicle Stimulating Hormone of 89 and Leutinizing Hormone of 22IU/L. To support the diagnosis karyotype was done, which was 46 XY. With informed and written consent she underwent exploratory laparotomy with peritoneal wash cytology with bilateral salpingectomy with bilateral gonadectomy along with infracolic omentectomy (Figure 1).

Intraoperatively there was right gonadal tumor of size 7x5 cm with left streak gonad and rudimentary tubes and uterus. There was no ascitis, organ involvement or enlarged lymph nodes. The disease was stage Ia carcinoma ovary. On cut-section gonadal tumor did not show areas of haemorrhage, necrosis or calcification. Final histopathology report came out as, gonadal tumor showing features of dysgerminoma and the other sent as streak gonad had scant ovarian stroma along with fibroadipose tissue, rest was free of tumor. She did not require chemotherapy further. Patient is under regular follow-up and has been put on oestrogen-progesterone combination, doing well presently.



Figure 1: Intraoperative appearance of right gonadal tumour.

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Tumor markers:

- LDH-1673 U/L
- α FP-2.24 ng/ml
- bhcg-5.88 miu/ml
- CA-125-159 U/L

Hormone profile – FSH-107, LH-26.1

Karyotype – 46 XY

Figure 3: Ultrasonography finding of 14x6x10 cm of multiloculated lesion in pelvis.

Another patient Y, 24 years presented with hypomenorrhoea, underdeveloped secondary sexual characteristics and pain right lower abdomen. She was 162 cm tall, thin built (44 kg) with breast and pubic hair – Tanner stage I-II. There was a solid mass of 5x4 cm felt in the right iliac fossa arising from pelvis, non tender and with restricted mobility. Same mass was felt on per-vaginal examination also, otherwise external genitalia was normal and rectal mucosa was free (Figures 2 and 3).

Patient underwent exploratory laparotomy with ascitic fluid cytology with total abdominal hysterectomy with bilateral salpingectomy with bilateral gonadectomy and infracolic omentectomy in view of stage II A carcinoma ovary. Intraoperatively there was ascitic fluid 100 ml, right ovarian

tumor 15x12 cm (capsule ruptured, adherent to uterus). Uterus was small size with a left streak gonad. Histopathology report showed gonadal tumor with features of dysgerminoma with serosal tumor deposits on uterus invading myometrium as well, streak gonad shows no ovarian stroma, rest all specimens were free of tumor. In view of advanced disease she received 6 cycles chemotherapy (PACLITAXEL + CARBOPLATIN) post-operatively. Presently doing fine and is under regular follow-up.

Discussion

The patients with 46, XY gonadal dysgenesis usually present in adolescence with primary amenorrhoea and delayed pubertal development. The etiology described is a defect in the SRY (putative testicular-determining factor gene), located on the short arm of Y chromosome. SRY gene mutations prevent production of the sex-determining region Y protein, resulting in a fetus who will develop as a female despite having a Y chromosome. The gonads fail to differentiate into testes inspite of Y chromosome rather resembles ovarian stroma, although non-functional. Without testes, no testosterone or antimüllerian hormone is produced. Hence, the external genitalia fail to virilize, and instead of wolffian ducts (without AMH) Müllerian ducts develop into normal internal female organs (uterus, fallopian tubes, cervix, vagina).

Gonadoblastomas are almost always associated with dysgenetic gonads but only 30% of dysgenetic gonads have germ line tumors [3,4]. Due to this risk of developing a germ cell tumor gonadectomy is advisable in these cases at the earliest. About 65% of dysgerminomas are stage I. About 10-15% are bilateral. The treatment of patient with early dysgerminoma is primarily surgical, including resection of the primary lesion and proper surgical staging, as was done in the present case. Post operative chemotherapy and/or radiotherapy only in patients with metastatic disease (18). Since our patient was IA, only surgery was sufficient and patient is presently under regular follow-up. If the contralateral is left, some disease can develop in 5 ~ 10% of the retained gonads over the next 2 years [5].

Thus, in an adolescent patient with primary amenorrhea, karyotype analysis and investigation of gonads is recommended. The prophylactic early surgical resection of gonads is recommended.

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