



Presentation of Dysgerminoma and Gonadoblastoma in a Patient with Swyer Syndrome

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ABSTRACT

Introduction Swyer syndrome is determined by primary amenorrhea, normal external genitalia, and the presence of a vagina, uterus, and 46XY karyotype. The aim of this case report was to introduce a patient with Swyer syndrome referred with pain and an abdominal mass.

Patient Information This case study was done in Gynecology Clinic of Ghaem Hospital in Mashhad, Iran, in 2015. A single 18-year-old woman came to the clinic with complaints of primary amenorrhea, pain, and abdominal mass underwent laparotomy. Based on her histopathology report which indicated a left ovary dysgerminoma and a right ovary gonadoblastoma, a bilateral salpingo-oophorectomy, followed by chemotherapy, was conducted. The patient was under Bleomycin, Etoposide and Platinum (BEP) chemotherapy and has been living without evidence of recurrence.

Conclusion A genetic disorder in patients younger than 20 years with an ovarian mass and diagnosis of dysgerminoma should be rejected.

Keywords Dysgerminoma; Gonadoblastoma; Ovary; Swyer Syndrome

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Introduction

Swyer syndrome was first described in 1955 by Dr. Swyer in two tall women with primary amenorrhea characteristics, normal female external genitalia, and the presence of a vagina, cervix, and 46XY karyotype [1]. Since they were born, patients with Swyer syndrome have had female phenotypes and unambiguous female external genitals. These patients are usually referred with complaints of primary amenorrhea or delayed maturation due to gonadal loss of function. 25% of these patients develop dysgerminoma or gonadoblastoma [2]. Nearly 5% of dysgerminoma were seen in patients with female phenotypes, abnormal gonads, and 46XY karyotype [2].

The aim of this case report was to introduce a patient with Swyer syndrome that referred with pain and an abdominal mass.

Patient Information

This study was done in Gynecology Clinic of Ghaem Hospital in Mashhad, Iran in 2015.

A single 18-year-old woman came to the clinic with complaints of primary amenorrhea and abdominal pain in June 2015. The patient had a female phenotype and, based on her medical records, she did not have any problems except for primary amenorrhea and abdominal pain. Her height was 176cm and her weight was 50kg. In the examination of the breasts, her breasts were at Tanner stage II and pubic hair growth was at Tanner stage I. She had a female external genitalia and clitoris. In the vaginal examination, the vaginal length was 10cm and the vagina had a soft consistency. Moreover, the cervix was normal. In the rectovaginal examination, the uterus was palpable.

During the examinations performed by conducting the abdomen and pelvis sonography, the uterus was 22×54mm and its position and shape were normal. The myometrium had a natural parenchymal echo pattern and there was no focal lesion. The endometrium was atrophic and was 2mm thick. A hypoechoic lobulated lesion was seen in the pelvis which extended beyond the navel (170mm) and contained an internal echo. Due to its magnitude, determining its uterine or ovarian origin was not possible. Several other lesions with similar sizes (50 to 70mm) were seen in the left flank region of the pelvis. Abundant free fluid was visible in the pelvis and the space around the spleen and there was also pleural effusion.

In the performed spiral CT scan, a large lobulated mass with dimensions of 108×192mm was observed in the right side of the pelvis which extended to the abdominal cavity. Another 50mm similar mass was reported in the left adnexa and omental infiltration and the left pleural effusion. The liver, spleen, and pancreas had normal shapes. Mild bilateral hydronephrosis was also observed. Both adrenals were healthy.

In the patient's laboratory tests, the following results were revealed:

LDH: 3671U/L (NL<480), Beta HCG: 2.2mIU/ml, Serum CEA: <0.5ng/ml (NL<5) CA-125: >200U/ml (NL≤35), AFP: 1ng/ml (NL<8.5), HB: 11.3, PLT: 235, HCT: 35.2%, and Cr: 0.9 (mg/dl).

In the conducted laparotomy, a tumor with dimensions of 15×19cm which was solid with a tight consistency and a cauliflower shape without any adhesions to the surrounding tissues was observed in the left ovary. There was a creamy cut surface tumor with dimensions of 1×3.5cm in the right ovary. A bilateral salpingo-oophorectomy was carried out on the patient and the uterus was preserved. In the left pelvic retroperitoneum, a 6cm lymph node which had an iliac in the place of bifurcation was resected. In the para-aortic, a large lymphadenopathy was found as a mass with dimensions of 15×15cm around the aorta and the spinal cord and it was not possible to resect it.

The histopathology findings included a left ovary dysgerminoma and a right ovary gonadoblastoma. In the examination of the pelvic lymph nodes, an involvement with dysgerminoma was reported. The omental involvement was not observed and the abdominal free fluid was seen without malignancy. The patient was under Bleomycin, Etoposide, and Platinum (BEP) chemotherapy and has been living without evidence of recurrence.

Discussion

The aim of this case report was to introduce a patient with Swyer syndrome that referred with pain and an abdominal mass.

Primary amenorrhea refers to a condition which presents itself with the lack of menstruation by 16 years of age in the presence of normal secondary sexual characteristics or by the age of 14 years and the lack of presence of such secondary characteristics [3]. Swyer syndrome is a rare cause of primary amenorrhea. This syndrome is a gonadal dysgenesis classified as sexual abnormalities [4]. There is a complete gonadal dysgenesis in this syndrome. The Wolffian ducts regress and the Mullerian ducts develop into the uterus, fallopian tubes, and vagina, and as a result, testosterone and inhibitor deficiencies have become major contributors to Mullerian. Patients with Swyer syndrome have female phenotypes and the appearance of female external genitalia. Minimal breasts enlargement indicates the aromatization of the environment of androgens. Both ovaries have fibrous tissues and ambiguous ovarian stroma. However, there is no follicle. It is suspected that the cause of 46XY complete gonadal dysgenesis is a short arm Y chromosome deletion including SRY or a mutation in other genes that leads to inhabitation of SRY function or a mutation of SRY function [5]. In patients with gonadal dysgenesis, either its specific

or 46XY karyotype, 46XY related to the physical manifestation of Turner syndrome (46XO) of both gonads is determined by fibrous strip textures and ambiguous ovarian tissues [6, 7]. Testicular feminization and true hermaphroditism are among differential diagnosis for this syndrome. In the testicular feminization, there is a lack of sensitivity to androgens in the target tissue and a person's karyotype is 46XY. In the true hermaphroditism, gonads include both ovarian and testicular tissues (Ovotestis) [8, 9]. In the laboratory findings, these patients have high levels of gonadotropins, reduced estrogen, and normal female androgen levels [5]. In the past, it was believed that there was no possibility of developing tumors in these patients [10]. However, recent studies indicated that these patients are at high risk of developing gonadoblastoma and dysgerminoma, which may occur in the early adulthood [11, 2]. The risk of developing gonadoblastoma or dysgerminoma in patients with Swyer syndrome is 25% [11]. The synchronization of gonadoblastoma and dysgerminoma are found in 50% of these patients [12]. Histopathology view of gonadoblastoma may indicate hyalinization, calcification, or excessive growth of germ cell tumors, mainly dysgerminoma. Maleki *et al.* reported such a disease with dysgenic gonads and in the pathologic view, it had a bilateral neoplasm [13]. At the time of diagnosis, 65% of cases of dysgerminoma are at stage I 85 to 90% of cases have stage I tumors in one of their ovaries. 10 to 15% of cases are bilateral. Dysgerminoma is the only malignant that has this bilateral percentage. Other germ cell tumors are rarely bilateral.

In patients with Swyer syndrome, conducting prophylactic gonadectomy is necessary to prevent gonadal cancer. It should be carried out on patients with Y chromosome gonadectomy as soon as upon diagnosis [14]. Treatments for patients with dysgerminoma include removing primary lesions and determining the stage of the disease. A chemoradiotherapy is accompanied with indications in patients with metastasis. In patients with a preserved ovary, the risk of malignancy in the remaining ovary is 5 to 10% in the next two years. If the uterus is not malignant and preserved, in some countries, it is possible to have a pregnancy with an ovum donation.

Conclusion

A genetic disorder in patients younger than 20 years with an ovarian mass and diagnosis of dysgerminoma should be rejected.

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