Case Report

DOI: 10.4274/tjem.2365

Swyer Syndrome: A Case Report

Swyer Sendromu: Bir Olgu Sunumu

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Abstract

Swyer syndrome is a pure gonad dysgenesis associating with 46.XY karyotype, primary amenorrhea and presence of female internal genital tract and bilateral streak gonads in a phenotypic female. The diagnosis is usually made at adolescence when the primary amenorrhea is investigated. A 23-year-old female presented with primary amenorrhea. On physical examination, she had female external genitalia. Laboratory analyses revealed serum follicle-stimulating hormone and luteinizing hormone levels compatible with hypergonodotropic hypogonadism. Pelvic ultrasonography and magnetic resonance imaging showed a hypoplastic uterus and absent ovaries. Chromosome analysis revealed 46.XY karyotype. Prophylactic bilateral gonadectomy was performed and histological examination showed pure gonadoblastome in the left ovary. In conclusion, the presence of Y chromosome in patients with 46.XY gonadal dysgenesis may increase the risk of gonadal tumors. A prophylactic bilateral salpingo-gonadenectomy should be advised to those patients. *Turk Jem 2014; 2: 56-57*

Key words: Gonadoblastome, Swyer syndrome, primary amenorrhea

Özet

Swyer syndrome dişi fenotipinde, dişi iç genital organ ve 2 taraflı çizgi gonadlar varlığında primer amenore ve 46,XY karyotipi ile ilişkili saf gonadal disgenezidir. Tanı genellikle adolesan dönemde primer amenore ile araştırılırken konulur. Yirmi üç yaşında kadın primer amenore ile başvurdu. Fizik muayenede kadın dış genital organına sahipti. Laboratuvar tetkiklerinde folikül stimüle edici hormon ve lüteinizan hormon düzeyleri hipergonodotropik hipogonadizm ile uyumluydu. Pelvik Ultrasonografi ve manyetik rezonans görüntülemelerinde hipoplastik uterus görüldü ve overler izlenmedi. Kromozom analizinde 46,XY karyotipine sahipti. Proflaktik bilateral gonadektomi yapıldı ve histolojik değerlendirmede sol overde saf gonadoblastom gözlendi. Sonuç olarak 46,XY gonadal disgenezili hastalarda Y kromozom varlığı gonadal tümör riskini artırabilir. Bu hastalarda proflatik bilateral salpingo-gonadektomi tavsiye edilmelidir. *Turk Jem 2014; 2: 56-57*

Key words: Gonadoblastom, Swyer sendromu, primer amenore

Introduction

Swyer syndrome was first identified in 1955 by Doctor Swyer in 2 women with primary amenorrhea, normal female external genitals, vagina and cervix, tall and, a 46.XY karyotype (1). Patients with Swyer syndrome have female phenotype. They usually present with delayed puberty or primary amenorrhea due to absence of functional gonads and reproductive potential. 25% of cases develop gonadoblastoma or dysgerminoma (2). Here, we present a 23-year-old patient with Swyer syndrome who had gonadoblastom in both dysgenetic gonads.

Case Report

A 23-year-old female was admitted to our clinic with the complaint of primary amenorrhea. On physical examination, she had female phenotype, 174 cm height, 65 kg weight, Tanner stage 3 breast development and tanner stage 5 pubic and axillary hair growth. The patient had female external genitalia and oversized clitoris. Pelvic ultrasonography (USG) showed 29x14 mm uterus and absent ovaries. No ovaries were observed on pelvic magnetic resonance imaging as well. Laboratory findings were consistent with hypergonadotropic hypogonadism (Table 1). Testosterone and cortisol levels were within normal limits. Diagnostic laparoscopy, which revealed rudimentary uterus and bilateral infantile fallopian tubes, was performed. Testis-looking gonads were observed in place of ovaries. Both gonads were removed by laparotomy for prophylactic purposes. Histopathological analysis revealed gonadal dysgenesis and pure gonadoblastoma on the left side. SRY gene analysis showed no mutation. 2 mg estradiol hemihydrate treatment was initiated. After ensuring adequate levels of estrogen, combined estrogen and progesterone hormone replacement therapy was given and menarche was provided.

Discussion

Primary amenorrhea is a condition with the absence of menstruation by age 16 years in the presence of normal

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Turkish Journal of Endocrinology and Metabolism, published by Galenos Publishing.

secondary sexual development or by age 14 in the absence of secondary sexual characteristics (3). Swyer syndrome is a rare cause of primary amenorrhea. Patients with Swyer syndrome are phenotypically female with female genital appearance. These individuals have 46.XY karyotype, incomplete sexual development, and clinically, they are with primary amenorrhea. Individuals with 46.XY gonadal dysgenesis are usually diagnosed in early adolescence due to incomplete pubertal growth. Female external genitalia, uterus and fallopian tubes are present. In laboratory tests, high levels of gonadotropin, decreased estrogen and normal female androgen levels are detected (4). In our case, gonodatropin levels were close to that in postmenopausal women, and estrogen levels were low for reproductive age.

Deletion of the testis determining factor gene (SRY) is detected in 10%-20% of patients. SRY gene is normal in the rest of the patients (80 to 90%) (5). SRY gene mutation was not found in our case. Testicular feminization and true hermaphroditism are considered in the differential diagnosis. In testicular feminization, androgen insensitivity at the level of the target organ is present and the karyotype is 46.XY. In real hermaphroditism, gonads containing both ovarian and testicular tissues (ovotestis) are observed (6,7). Our case did not have ovary tissue, thus, the differential diagnosis was made based on real hermaphroditism, besides,the presence of hypergonadotropic hypogonadism separated our case from testicular feminization. There is a risk of developing gonadoblastoma or dysgerminoma in 25% of patients with Swyer syndrome (2,8). In patients diagnosed with Swyer syndrome gonadectomy is required. Due to the risk of malignant gonadal tumor development, patients with female phenotype, who are carrying the Y chromosome, gonadal tissue should be removed following the diagnosis (9). In our case, histopathological evaluation after bilateral gonadectomy showed pure gonadoblastoma in the left ovary. Management of Swyer syndrome is done similar to that of hypogonadism due to other causes. To ensure the development of secondary sexual characteristics, estrogen should be given to stimulate puberty. Then the combination of estrogen and progesterone is given in the long-term (10). In our case, 2 mg of estradiol was initiated and the treatment was followed by estrogen and progesterone combination after we provided secondary sexual development and endometrial mucosal thickening.

As a result; in adolescence period, karyotype analysis should be performed in patients who present with primary amenorrhea and hypergonadotropic hypogonadism. Gonadectomy should be performed in those with gonadal dysgenesis due to the high risk of gonadal neoplasia.

Table 1. Hormonal tests for the etiology of primary amenorrhea

Hormones	Results	Normal value
FSH (mIU/ml)	34.7	3.03-8.08
LH (mIU/ml)	13.6	2.39-6.6
Estradiol (pg/ml)	<10	21-251
DHEA-S (mcg/dl)	423	134.2-407.4
Cortisol (mcg/dl)	19.3	3.7-19.4
ACTH (pg/ml)	37.5	10-46
17α OH Progesterone (ng/ml)	0.6	0.23-1.36
Androstenedion (ng/ml)	2.76	0-1

Conflicts of Interest

There are no conflicts of interest.

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