CASE REPORT

Ersen Eraydın[1]
Şevki Göksun Gökulu[2]
Ahmet Karataş[3]
Seyhan Sönmez[1]

[1] Bitlis State Hospital, Department of Obstetrics and Gynecology, Bitlis.
[2] Ministery Of Health, Aegean Maternity Education And Research Hospital, Izmır
[3] Beyhekim State Hospital, Department Of Obstetrics And Gynecology, Konya.

Corresponding address:
MD, Ahmet Karataş
Beyhekim State Hospital, Department of Obstetrics and Gynecology, 42250, Selçuklu, Konya-Turkey
Tel: 0090 505 638 1344
email:akaratas1973@hotmail.com

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konuralptipdergi@duzce.edu.tr
konuralpgeneltip@gmail.com
www.konuralptipdergi.duzce.edu.tr

Laparoscopic Gonadectomy for Complete Androgen Insensitivity Syndrome: A Case Report

ABSTRACT
Objective: Our aim was to present and discuss a case with androgen insensitivity syndrome who underwent laparoscopic gonadectomy.
Case: A 21 year-old women admitted to our clinic with the complaint of primary amenorrhea and infertility. She had sufficient breast maturation but have scanty pubic and axillar hair. In gynecologic examination vagina was 5 cm in length and ended blindly. In ultrasonographic examination uterus was absent, there were bilateral masses each 3 cm in diameter located near the internal os of the inguinal canals. Her karyotype was 46 XY.
Results: The patient underwent laparoscopy. Pelvic inspection revealed no internal genitalia except bilateral gonads appearing as testis. The pedicle of gonads were coagulated with bipolar diathermy and cut with laparoscopic scissors and removed with endobags.
Conclusions: Androgen insensitivity syndrome should be suspected in cases with primary amenorrhea and laparoscopic gonadectomy should be performed after puberty
Key words: Laparoscopy, primer amenorrhea, androgen insensitivity

Komplet Androjen Duyarsızlık Sendromunda Laparoskopik Gonadektomi: Olgu Sunumu

ÖZET
Amaç: Laparoskopik gonadektomi uygulanan androjen insensitivite sendromlu bir hastayı sunmayı ve tartışmayı amaçladık.
Olgu: Yirmi beş yaşında bir kadın primer amenore ve infertilite şikayetiyle kliniğimize başvurdu. Meme gelişimi uygun fakat pubik ve aksiller kıllanması yetersizdi. Jinekolojik muayenede vajina 5 cm ve kör sonlanmaktaydı. Ultrasonografik muayenede uterus yok, inguinal kanal iç kısmında yerleşimli ikisi de üç cm çapında iki taraflı kitleleri vardı. Karyotipi 46 XY idi.
Bulgular: Hasta laparoskopiye alındı. Pelvik inceleme iki taraflı testis görünümlü gonad dışında iç genital organ olmadığını açığa çıkardı. Gonadlar pediküllerinden bipolar diyatermi ile koagüle edilerek ve laparoskopik makasla kesilerek endo-bag içinde çıkartıldı.
Sonuç: Primer amenoreli hastalarda androjen insensitivite sendromundan şüphelenilmeli ve laparoskopik gonadektomi puberteden sonra uygulanmalıdır.
Anahtar kelimeler: Laparoskopi, primer amenore, androjen duyarsızlığı

INTRODUCTION

Androgen insensitivity syndrome (AIS), previously called 'testicular feminization syndrome' is a X linked recessive disease due to a mutation at Xq 11-q 12 localization on the androgen receptor gene (1). AIS is the most common cause of male pseudohermaphroditism, patients usually present a female phenotype with a normal 46 XY karyotype, primer amenorrhea and impaired development of the secondary sexual characteristics (2). The prevalence of the syndrome is estimated to be at least 1 in 20,000 births (3). The complete form is a more common variety, occurring in 1 in 20,000 to 64,000 male births. The importance of this syndrome, testicular tumors, especially seminomas, may develop after puberty (1).

Gonadal malignancies like sertoli cell tumor, yolk sac tumor, embryonic teratoma and unclassified sex cord stromal tumor are rare in these patients (1, 4). The risk for the occurrence of gonadal tumors is relatively late and rare before the age of 25 (5). The risk of developing testicular neoplasia is 3.6 % at an age 25 years and 33 % at 50 years (4). The diagnosis is often based on clinical findings such as presence of nondysplastic testis, absence of a cervix, uterus and fallopian tubes and a vagina of variable length (6). In this report we present and discuss a case with complete androgen insensitivity syndrome who underwent laparoscopic gonadectomy.

CASE

A 21-years old woman admitted to our clinic with the complaint of primary amenorrhea and infertility. The patient was 175 cm tall and 60 kg weight. Her breast maturation was Tanner's stage 5 and scanty pubic and axillar hair was present. She had no pathological finding in her physical examination. In gynecological examination, the vulva and perineum appeared normal; vagina was 5 cm in length and ended blindly. The transabdominal pelvic ultrasonography confirmed the absence of uterus. There were bilateral masses each 3 cm in diameter located near the internal os of the inguinal canals. She had no other pathological finding in her abdominal ultrasonographic investigation. The hormonal analysis were as follows: FSH:10.7 mIU/ml, LH:19.4 mIU/ml, Estradiol:38 pg/ml. Other hormonal parameters and tumor markers were in normal limits. We had no investigated testosterone levels before operation. In cytogenetic examination 46, XY karyotype was determined.

After standard preoperative preparation, operative laparoscopy was performed under general anesthesia. Pelvic and abdominal inspection revealed no internal genitalia except, bilateral gonads appearing as testis attached near the internal os of the inguinal canals. The pedicle of gonads were coagulated with bipolar diathermy and cut with laparoscopic scissors. To prevent the spillage of cells and contamination gonads were placed in endobags and removed intact after extending the port. No complications occurred during the operation. The patient was discharged on the following day after surgery.

The histopathologic report revealed a testicle on the right 3x5x5 cm with 8x1 cm duct addicted to it (Figure 1). On the left a testicle 3x2x2 cm was reported (Figure 2). Histopathological diagnosis was bilateral sertoli cell adenoma, bilateral leyding cell hyperplasia and a wolffian ducts cyst next to right testicle. A long–term conjugated estrogen 0.625 mg per day was started.

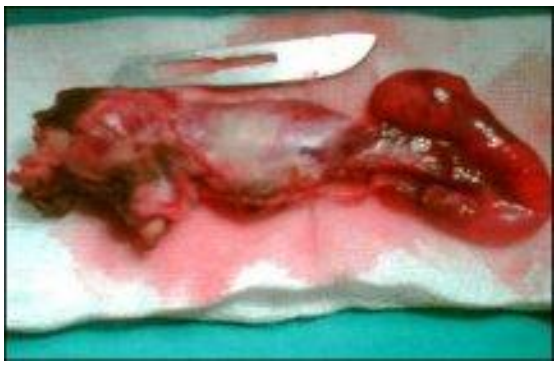

Figure 1. Right side gonad

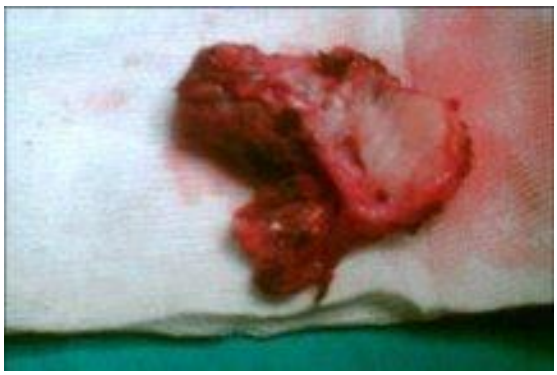

Figure 2. Left side gonad

DISCUSSION

AIS is the most common cause of the male pseudohermaprotidism and the third most common cause of primary amenorrhea (approximately 10% of the primary amenorrhea) (2,7). Different types of AIS have been reported (2,7). There are three AIS phenotype classifications: complete androgen insensitivity syndrome (CAIS), also known as testicular feminization. Partial androgen insensitivity syndrome (PAIS), and mild androgen insensitivity syndrome (MAIS) also known as under-virilized male syndrome (8). CAIS, the typical mode of presentation is in an adolescent female who has breast development with a pubertal

growth but has no menarche and absence or scanty growth of pubic and axillary hair. CAIS may also present in early infancy with the appearance of bilateral inguinal or labial swellings. Bilateral inguinal hernia are rare in girls and it has been estimated that 1-2% of such cases have CAIS. On the other hand if a female child shows inguinal hernia CAIS must be suspected every time (6). In the literature a case of CAIS in a 22-year-old female where the presence of testes, prostatic tissue and seminal vesicles was confirmed by ultrasonography, hormonal analysis, operative findings and histopathological study. She was second of the two sisters and her elder sister was also diagnosed with CAIS (9). The PAIS terminology implies a biological response to androgens in terms of immediate sex assignment impossible. PAIS is one cause of intersex. The prototypic phenotype for PAIS is characterized by perineo-scrotal hypospadias, micropenis and a bifid scrotum. The testes may also be undescended. The more severe form of PAIS, presents as isolated clitoromegaly (8).

MAIS is the category of AIS was realized following investigations for male factor infertility which suggested a defect in androgen action. Several large surveys of males with oligospermia and normal testosterone levels with increased LH concentrations have shown that a small percentage have AIS as based on finding a mutation in the androgen receptor gene (8,10). Diagnosis of CAIS is usually with absence of female internal genital organs on physical exam and pelvic ultrasonography, karyotyping, molecular genetic testing of the AR gene mutations (chromosomal locus Xq 11-q 12), and elevated testosterone and luteinizing hormone levels (6,11).

In our case the diagnosis of AIS was based on gynecologic examination, laparoscopy and the karyotype. Gonadal tissue can be located in the inguinal canal or anywhere in the abdomen, sites that are invisible during laparoscopic examination. Magnetic resonance imaging and laparoscopic examination have proven value for localization of nonpalpable undescended testes (12,13). Kim reported that inguinal hernia repair of 3-year-old female complete AIS was discovered during the operation. Hernias were repaired and gonadal biopsy was taken. The gonads were then placed into the subcutaneous space below the skin incision to facilitate a later orchiectomy (14).

There is a increased risk of dysgenetic gonads for developing malignancies which can be as high as 30%. In contrast to the other forms of gonadal disgenesis the incidence of tumors in AIS cases is rare before puberty and significantly higher after the age of 35 (2,8). Kriplani reported that two of seven male pseudohermaprodite cases (28.6%) who underwent laparoscopic gonadectomy had gonadal neoplasias (15). For patients with AIS prophylactic gonadectomy is necessary in the post pubertal period for the risk of malignant potential of the gonads. Gonadectomy is performed during the post pubertal period, for to allow development of the secondary sex characteristics during puberty (1,4). The laparoscopic removal of gonads has many advantages compared to laparotomy: minimal blood loss, rapid recovery, shorter hospital stays and minimum psychological trauma are the advantages. Laparoscopy has a better visualization of abdomen and pelvis compared to the laparotomy (15).

The operational time is similar in laparoscopy and laparotomy, but the recovery time and the duration of the hospital stay of the patients is much shorter with laparoscopy when compared with laparotomy (16,17). The patients with AIS should be treated with long–term hormonal replacement therapy after gonadectomy (2,7). The androgen supplementation treatment is not beneficial in these patients because of the absence of functional androgen receptors (18).

In conclusion androgen insensitivity syndrome should be suspected in cases with primary amenorrhea and laparoscopic gonadectomy should be performed after puberty.

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