Uncommon Laparoscopic Findings in a Sexually Immature Agonadal XY Phenotypic Female

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Abstract – A defect of intrauterine development occurring early in male embryogenesis results in a broad range of XY agonadal phenotypes. A 16-year-old girl was referred for evaluation of primary amenorrhea and failure of sexual development. Gynecological examination revealed infantile but otherwise normal female external genitalia with clitoral hypotrophy, absence of hymen, and a tight vaginal introitus. The vagina was 4 cm long and ended in a blind pouch. No masses were detected in inguinal canals or in labia majora. Ultrasonography, computerized tomography, and magnetic resonance imaging failed to demonstrate internal genitalia or gonads. Endocrine evaluation showed elevated plasma concentrations of Follicle Stimulating Hormone and Luteinizing Hormone with undetectable estradiol levels. The karyotype from peripheral blood cultures of peripheral leucocytes was 46, XY. At laparoscopy no uterus or ovarian structures were found. Only two small fibrous oval masses with a thin peduncle were found along each lateral pelvic wall and were excised. Histological examination of these masses revealed that they resembled oviduct tissue. Therefore, the present agonadal XY phenotypic female is the third of such cases that have been reported to have only rudiments of fallopian tubes but is the first of these three cases that has been explored entirely by laparoscopy.

Keywords - Disorders of Sex Development, Embryonic Testicular Regression, Laparoscopy, Male Pseudohermaphroditism, Vanishing Testis

I. INTRODUCTION

Testis regression is one of the causes of 46, XY disorders of sex development (DSDs).1 In 1956, Overzier and Linden2 first described what they called “true agonadism” in two siblings with female phenotype, rudimentary genital structures, male karyotype, and absence of gonads. Thereafter, there were other reports of patients with similar characteristics that were called by different authors “pure gonadal dysgenesis”, “Swaer’s syndrome”, “XY gonadal agenesis syndrome”, “agonadism”, “embryonic testicular dysgenesis”, “vanishing testis”, or “early testicular dysgenesis”.3

In 1977, Edman et al.4 postulated that a defect of development occurring early in male embryogenesis could result in a broad phenotypic range of XY agonadal persons and suggested the term “embryonic testicular regression” to include the whole clinical spectrum of XY agonadal phenotypes.5 Since the loss of testicular function may occur at different times during male intrauterine development, agonadism in 46,XY phenotypic females presenting with sexual infantilism may be associated with only remnants or total absence of Müllerian and/or Wolffian derivatives.3,4 So far only two cases have been reported that had nothing more than rudiments of the fallopian tubes.6,7

Surgical exploration for gonadectomy is indicated in patients with 46, XY DSDs to reduce the risk of cancer estimated as high as approximately 30% in dysgenetic gonads.8 Laparoscopic approach to gonadectomy has been demonstrated to be both effective and safe for pediatric and adolescent patients with DSDs.9,10 However, only two cases of agonadal XY phenotypic females were surgically explored entirely by laparoscopy whereas the majority of such cases underwent laparotomies after a brief “diagnostic” laparoscopy.9,10

The present case of agonadal XY phenotypic female is the third of such cases having nothing more than rudiments of the fallopian tubes but is the first of such cases that has been explored entirely by laparoscopy.

II. CASE REPORT

A 16-year-old white female was referred to our department for evaluation of primary amenorrhea and failure of sexual development. The girl was the only child of her parents and she was born at term after an uncomplicated pregnancy. She had been reared as a female; growth and development were considered normal until puberty, when secondary sex characteristics failed to develop. No other developmental anomalies were known in her family.

Physical evaluation revealed a sexually immature female. The height was 150 cm, the weight was 40 kg and the blood pressure was 120/80 mm Hg. Breast development was absent. She had only sparse axillary and pubic hair.

Gynecological examination revealed infantile but otherwise normal female external genitalia with clitoral

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hypotrophy, absence of hymen, and a tight vaginal introitus. The urethra was normally positioned in the vestibule. The vagina had a 4 cm length and ended in a blind pouch. No cervix was identified. Rectovaginal examination failed to demonstrate the presence of uterus and/or adnexal. No masses were detected in both inguinal canals or in labia majora or perineum. Perianal condylomas were present.

Both transabdominal and transrectal pelvic ultrasonography, as well as computerized tomography and magnetic resonance imaging, failed to demonstrate internal genitalia or gonads.

Basal hormones evaluation showed elevated plasma concentrations of both Follicle Stimulating Hormone (FSH, 144 IU/l) and Luteinizing Hormone (LH, 35 IU/l) and undetectable estradiol levels. Total and free testosterone, prolactin, 17-hydroxyprogesterone, cortisol, and sex hormone binding globulin were within the normal adult range.

The analysis from cultures of peripheral leucocytes revealed a 46, XY karyotype. A diagnosis of male pseudohermaphroditism (or 46, XY disorder of sex development or 46, XY DSD) was made and an exploratory laparoscopy was proposed to the patient and her parents to search for gonadal remnants.

At laparoscopy no uterus or ovarian structures were found. Only two small fibrous oval masses with a thin peduncle were found along each lateral pelvic wall and were excised. A thorough search of both inguinal canals and the anatomic pathways of testicular descent failed to identify any evidence of testicular structures. The postoperative stay was normal and the patient was discharged from the hospital two days after laparoscopy.

Histological examination of the excised fibrous masses revealed that they were tubular and resembled oviduct tissue with fibrous stroma but without gonadal elements.

III. DISCUSSION

Approximately one hundred cases of agonadism have been reported since 1564, when bilateral absence of the testis was first described. 2-5,8-9 The loss of testicular function may occur at different times during male intraterine development and a broad range of XY agonadal persons, phenotypic males or females, have been described. 2-5,8-9 Agonadism in 46, XY phenotypic females has been reported to be associated with only remnants of Müllerian or Wolffian derivatives as well as normal female or ambiguous external genitalia. 3,4

The patient described in the present report is the last of the only three reported so far, who presented with infantile, but normal, female external genitalia, clitoral hypotrophy, absence of hymen, tight vaginal introitus, short vagina, and cord-like folds of tissue that resembled tubal remnants along each pelvic wall. The previous two cases have been both reported in 1977. 4,5 One report was in German. 4

This is the first time since 1977 4,5 that a sexually immature agonadal XY phenotypic female having nothing more than rudiments of the fallopian tubes successfully underwent only laparoscopy to define the diagnosis of “embryonic testicular regression” occurring during early gestation. Interestingly, only another case of agonadal XY phenotypic female without internal genitalia but with enlarged clitoris was surgically explored entirely by laparoscopy in 1976. 8 Thereafter, all reported cases of XY agonadal patients underwent surgical exploration only by laparotomy or by a laparoscopy which followed a previous laparoscopy. 3,4,9

In 1979 Coulam 3 reported the case of a phenotypic female patient who underwent a laparoscopy when she was 16-year-old, which failed to demonstrate the presence of rudimentary internal genitalia, and subsequently underwent a laparotomy when she was 21-year-old, which revealed rudimentary uterine horn with fimbriae on either side of the pelvis. One could hypothesize that in this case laparoscopy failed to demonstrate uterine horns because of the primitive technology available at that time. 3 However, twenty years later a diagnostic laparoscopy failed again to demonstrate the presence of rudimentary internal genital organs in a different case of “embryonic testicular regression” syndrome, but remnants of epididymis, vas deferens, and seminiferous tubules were uncovered during exploratory laparotomy. 9 This last report suggests that surgeon experience in such delicate cases probably plays a major role in making laparoscopy able to identify remnants of internal genitalia.

The rarity of cases having testis regression among all cases of 46, XY DSDs may explain the difficulties encountered by laparoscopic surgeons in such diagnose. However, laparoscopic management of these patients could reach the same efficacy of laparotomy by increasing laparoscopic surgeons’ experience.

REFERENCES