A Rare Case of Rudimentary Uterus With Absence of Both Ovaries and 46,XX Normal Karyotype Without Mosaicism

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SUMMARY

Objective: We report an 18-year-old patient with bilateral ovarian agenesis, rudimentary uterus and normal fallopian tubes, and with normal 46,XX karyotype (without mosaicism).

Case Report: The patient was admitted to our clinic with primary amenorrhea. Secondary sexual characteristics (thelarche and pubarche) were both Tanner classification stage 1. With the help of vaginoscopy, vaginal depth was measured without distorting the hymenal ring and was found to be 8 cm. The laboratory findings were as follows: follicle-stimulating hormone 85 IU/L, luteinizing hormone 40 IU/L, and estradiol 14 pg/dL. Genetic investigation revealed a 46,XX karyotype without any mosaicism. Diagnostic laparoscopy was performed. During laparoscopic pelvic exploration, a rudimentary uterus without ovaries and normal bilateral fallopian tubes were observed. Bone mineral densitometry measurements were in the normal range. The patient was given oral contraceptives for hormone replacement.

Conclusion: If gonadal agenesis is thought to be the cause of primary amenorrhea in patients with defective secondary sexual characteristics, we believe that laparoscopic evaluation is the gold standard in diagnosis. [*Taiwan J Obstet Gynecol* 2008;47(1):84–86]

Key Words: diagnosis, follow-up, laparoscopy, ovarian agenesis, rudimentary uterus, treatment

Introduction

Primary amenorrhea may be due to a hypothalamic pituitary disorder, gonadal dysgenesis or müllerian duct malformations, with gonadal dysgenesis being the most common cause [1]. The Mayer-Rokitansky-Küster-Hauser syndrome is characterized by uterine aplasia or hypoplasia [2].

Gonadal agenesis is a rare clinical entity, usually observed in association with 46,XY karyotype and absence of secondary sexual characteristics. To the best of our knowledge, we report here for the first time in the literature the presence of a rudimentary uterus, together with gonadal agenesis but with a normal vagina and normal fallopian tubes, in a patient with a 46,XX karyotype and no other associated organ system anomaly.

Case Report

An 18-year-old virgin patient was admitted to our clinic with complaints of primary amenorrhea and absence of secondary sexual characteristics. Anamnesis revealed that she was the only child of phenotypically normal parents. She had been born by normal delivery, and her birth weight was 3,000 g. Her mother was 165 cm tall and had experienced her first period at 15 years of age. The father was 180 cm tall and was phenotypically normal.

The patient was 160 cm tall and weighed 65 kg. Physical examination revealed prepubertal features, thelarche and pubarche were both Tanner stage 1. The bone age was determined as 16 years by radiography. Genital

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Accepted: October 2, 2007

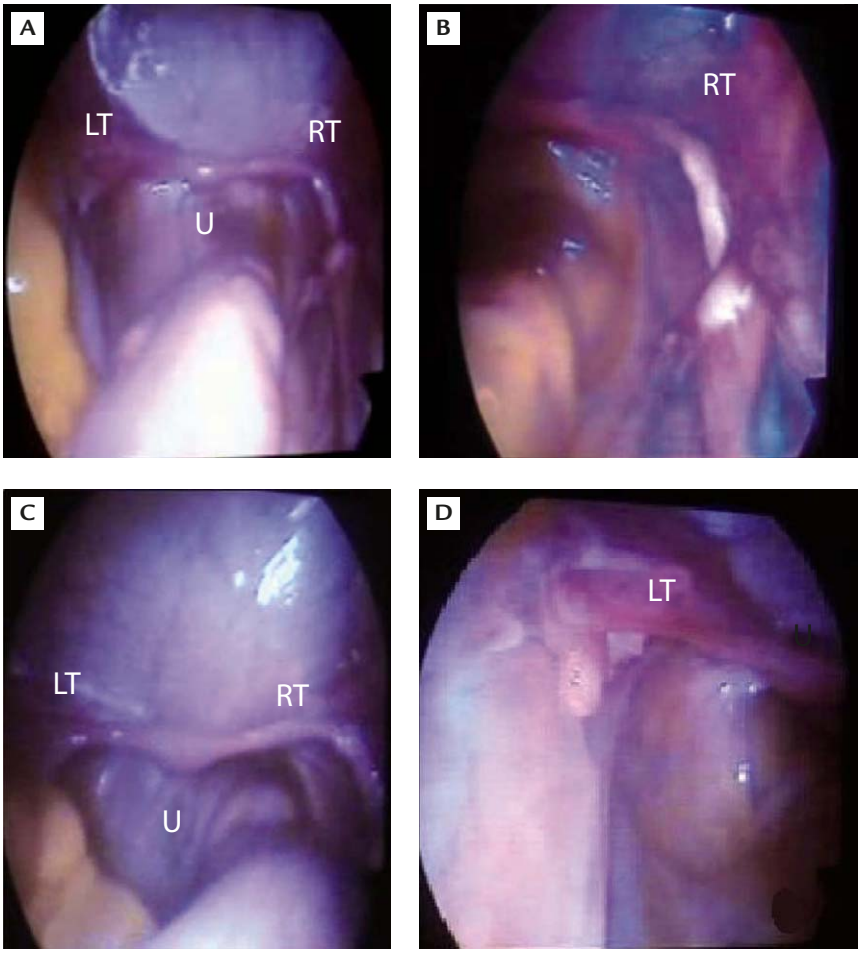

Figure. (A) Rudimentary uterus with normal fallopian tubes and absence of both ovaries. (B) Normal right fallopian tube, normal round ligament and no ovary. (C) Rudimentary uterus with normal fallopian tubes and absence of both ovaries. (D) Normal left fallopian tube, normal round ligament and no ovary. LT = left fallopian tube; RT = right fallopian tube; U = uterus.

inspection revealed the clitoris to be normal in size and shape, whereas the labia minora and majora were hypoplastic. Since virginity is culturally and traditionally important in our country, a bimanual pelvic examination cannot be performed. A vaginoscopy using office hysteroscopy optics was therefore planned to determine the presence and depth of the vaginal canal without distorting the hymenal ring. Vaginoscopy showed a vaginal length of 8 cm and a hypoplastic cervix.

The hormone laboratory results were: follicle-stimulating hormone 85 IU/L, luteinizing hormone 40 IU/L and estradiol 14 pg/dL, and the thyroid hormone panel was normal.

Bone mineral density, measured by bone mineral densitometry, and blood lipid levels were normal. In pelvic ultrasonography, the uterus was observed as hypoplastic and ovaries were not visible.

Using intravenous pyelography, both kidneys and ureters were observed to be normal. Hypophysial investigation by computerized brain tomography revealed no abnormalities. All other biochemical and hematologic parameters, as well as the results of urinalysis, were normal.

The result of karyotype analysis performed by the fluorescence *in situ* hybridization technique reported the absence of a Y chromosome and a 46,XX karyotype. Since this patient was initially suspected to have hypergonadotropic hypogonadism, diagnostic laparoscopy was performed. On laparoscopic pelvic exploration, the uterus was rudimentary and both ovaries were absent.

Bilateral fallopian tubes were normal (Figure). Oral contraceptive treatment was initiated in order to support secondary sexual characteristics and to prevent long-term complications.

Discussion

A review of the literature on the subject did not reveal any case of bilateral ovarian agenesis in association with rudimentary uterus, normal fallopian tubes and vagina in a patient with 46,XX karyotype [3].

The absence of a uterus and vagina, together with the absence of ovaries, is called Mayer-Rokitansky-Küster-Hauser syndrome. Our case is discriminated from this syndrome by the presence of a normal vagina and fallopian tubes in the absence of gonads.

Other organ anomalies coincident with müllerian duct agenesis are common. In approximately 30% of these cases, renal abnormalities (horse-shoe kidney, unilateral renal agenesis, etc) and in another 25%, skeletal anomalies (spina bifida, scoliosis, etc) are observed [4]. In our case, the absence of gonads and a rudimentary uterus were the only anomalies, and no other associated anomaly was detected.

Initially, undifferentiated gonads present as a mass in front of the coelomic epithelium at the fourth to fifth week of embryonic development. Later on, the cortex and medulla appear within the mass. In males, the cortex regresses, whereas the medullary part develops. In females, the cortex develops and the medulla regresses. The migration of primordial germ cells from the yolk sac to the gonads is one of the most important steps of embryonic development (fifth to sixth week). If this step fails, the gonads cannot develop completely, and gonadal agenesis occurs [5]. In our case, we could not detect any factor that might have led to gonadal agenesis and a rudimentary uterus, and we consider this to be a sporadic case.

The height of patients with Turner syndrome averages about 147.3 ± 6.6 cm [6]. Our patient's height was above this average. The absence of any mosaicism and the normal body constitutions of the parents led us away from this diagnosis.

In our opinion, good clinical practice in patients admitted with primary amenorrhea and defective secondary sexual characteristics should include a detailed general physical and pelvic examination. Hormonal and genetic laboratory tests should also be carried out.

In conclusion, we believe that if gonadal agenesis is thought to be the cause of primary amenorrhea, laparoscopic evaluation should be the gold standard for diagnosis.

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