Case Report

Importance of Laparoscopic Assessment of the Uterine Adnexa in a Mayer-Rokitansky-Kuster-Hauser Syndrome Type II Case

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ABSTRACT: In the case reported, diagnosed with Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, the presence of normal ovaries proved to be challenging to confirm due to unusual high positioned (ectopic) ovaries. MRKH syndrome is a rare pathological condition characterized by a spectrum of the Mullerian duct abnormalities resulting in congenital aplasia of the uterus and of the upper part (2/3) of the vagina, developed during embryogenesis. At the same time, the mullerian development is interdependent with the Wolffian (mesonephric) duct and this explains the associated renal abnormalities (MRKH type II). Laparoscopic assessment was of great importance in defining the exact anatomic characteristics of MRKH syndrome.

KEYWORDS: *adnexal ectopy, laparoscopy, Mayer-Rokitansky-Kuster-Hauser syndrome, renal agenesis*

Introduction

The Mayer-Rokitansky-Kuster-Hauser syndrome represents a developmental anomaly of the female genital tract and should be suspected when a young female complaints are primary amenorrhea and the inability of sexual intercourse. The diagnosis is usually accessible using clinical information, laboratory results and ultrasonographic (US) findings. If these findings are inconclusive, magnetic resonance imaging (MRI) should be performed, since failure to clearly identify the uterus, the Mullerian rudiments or ovaries by US does not necessarily imply their absence. If MRI fails to define the abnormalities of the uterus and the precise anatomical location of the possible tubal remnants and the ovaries, laparoscopy is used for the diagnosis. We report a case of type II Mayer- Rokitansky-Kuster-Hauser syndrome associated with adnexal ectopy. In this case laparoscopy was instrumental in confirming the unilateral renal agenesis and presence of the morphological and functionally normal, but ectopic ovaries, missed on US scan and MRI examination.

Case report

An 18-year-old young woman presented to our clinic for primary amenorrhea and dyspareunia. She developed clinical signs of puberty at 12 years old (thelarche -11 years old, pubarche - 12 years old) and she became sexually active at 14 years. She had two previous surgical interventions, an appendectomy and an exploratory laparotomy. The physical exam noted an average stature and body mass index (165cm in height, 66 kilos in weight), normal developed breast (Tanner 5) (Fig 1.a) and normal body hair distribution, including arm span, pubic and axillary hair (Fig1.b.c). The clinic examination revealed normal clitoris, labia major and minor, and a short (2 cm) vagina, that ended in a blind pouch (Fig 1.d).

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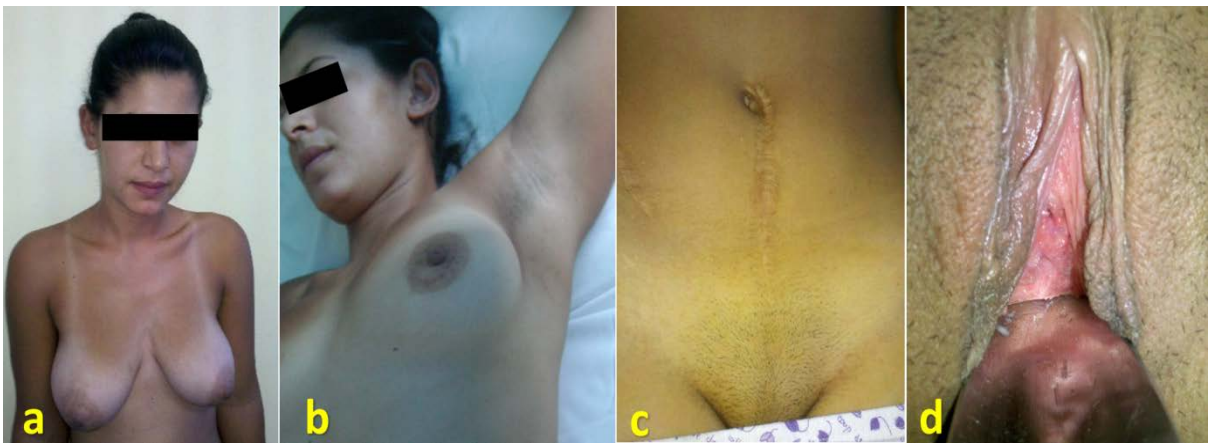

Figure 1. Images showing normal secondary sexual characteristics:
a.Normal development of breast (Tanner 5) and normal distribution of the facial hair.
b.Normal distribution of body hair, arm span.
c.Abdomen, anterior aspect, normal growth of pubic hair and postoperative scars.
d.Vulva and vagina exposed at the clinical evaluation. Normal clitoris, labia majora and minora and a small, 3cm, vagina ended in a blind pouch.

The US exam (Voluson Pro 730 GE Medical Systems, Zipf, Austria machine, equipped with 4-8-MHz and 5-9 MHz curvilinear transducers) found an ectopic right solitary kidney (with mild pyelectasis), located in the right low quadrant of the abdomen, and failed to identify the uterus and ovaries using both transabdominal and transvaginal approach (Fig 2.a.b).

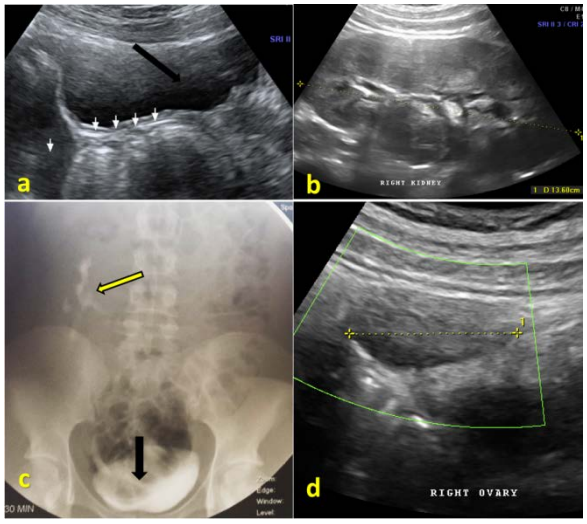

Figure 2. Paraclinical findings:
a.Ultrasonographic transabdominal sagittal view showing a normal urinary bladder (black arrow) and the interface between the urinary bladder and the rectum, with no uterus identified (white arrows).
b.Ultrasonographic transabdominal longitudinal view of the ectopic right congenital solitary kidney.
c.Intravenous pyelography showing normal secretion of the solitary right kidney (yellow arrow) and a normal filling of the urinary bladder (black arrow).
d.Ultrasonographic confirmation of the right ectopic ovary after the laparoscopic localization.

Due to the discordance between the clinical and laboratory findings (normal complete sexualisation, normal hormones) and ultrasound features (absent ovaries) we considered these findings incomplete. Thus, we performed MRI, which revealed normal liver, pancreas, confirmed the right solitary ectopic kidney and the absence of uterus, but failed to visualize any tubal remnants and the ovaries.

Blood routine and renal function were in normal limits. Hormone profile included measurement of follicular stimulating hormone (4.3mUI/ml), estradiol (840pmol/l) and testosterone (1.59 nmol/l), which were all

normal, indicating normal function of the hypothalamic-pituitary-ovarian axis.

Classic G banding is the elective genetic method used to differentiate the Mayer-Rokitansy-Kuster-Hauser syndrome from other genital tract development defects such as Turner syndrome (45, 0X) and androgen insensitivity syndrome (46, XY). The karyotype was normal (46, XX).

The cardiologic exam (ECG and echocardiogram) showed normal heart functionality with no evidence of morphological anomalies and the chest X-ray was also normal. Intravenous pyelography showed left renal agenesis, right hypertrophic kidney in ectopic ilio-pelvic position, with normal secretion and excretion function and normal urinary bladder (Fig 2.c).

After counselling, informed consent for laparoscopic exploration was obtained, in order to locate and describe the internal genitals. Laparoscopy confirmed normal urinary bladder, absent uterus (Fig 3.a), left renal agenesis and a right ectopic kidney (Fig 3.b). Both ovaries were found in an ectopic high position, in the right and left superior quadrants, with adjacent small fallopian tubes. The right ovary had a normal aspect (Fig 3.d) and the left ovary presented a haemorrhagic cyst that was removed (Fig 3.c). Complete inspection of the peritoneal cavity showed a congestive cecal appendix, also removed. After acknowledging the topography of the ovaries, we were able to identify them on later US scans (Fig 2.d). There were no postoperative complications and the patient was discharged after 3 days. Before discharge, another ultrasonographic evaluation was performed and revealed both high positioned ectopic ovaries. Further psychological support was provided and corrective dilatory surgery of the vagina was recommended.

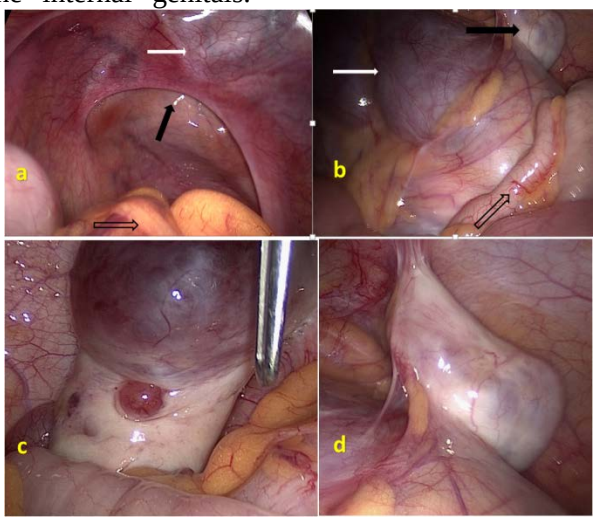

Figure 3. Laparoscopic findings, completing the diagnostic protocol of MRKH syndrome type II:
a.Laparoscopic image of the pelvis showing the absence of the uterus (black arrow), the urinary bladder with a Foley catheter balloon (white arrow) and bowels (open black arrow).
b.Laparoscopic image of the congenital solitary kidney (white arrow), right ectopic ovary (black arrow) and appendix (open black arrow).
c.Laparoscopic image of the left ovary with a haemorrhagic cyst.
d.Laparoscopic image of a macroscopically normal right ectopic ovary.

Discussion

The Mayer-Rokitansky-Kuster-Hauser syndrome diagnostic is usually clinically established [2], when the patient presents with primary amenorrhea and the inability of sexual intercourse due to vaginal aplasia/hypoplasia. It is the second most frequent cause of amenorrhea, after gonadal dysgenesis in Turner's syndrome [2]. The prevalence has been reported as 1 in 4.500 female births [3].

In the literature, MRKH syndrome is subdivided into type A (typical) having symmetric uterine remnants and normal fallopian tubes and type B (atypical) with asymmetric uterine buds and abnormally developed fallopian tubes and other organ system anomalies [4]. Another classification of the syndrome describes two types: type 1 or isolated or Rokitansky sequence and type II (the MURCS association- Mullerian duct aplasia, Renal dysplasia and Cervical Somite anomalies),

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that associates urologic (upper tract), vertebral, cardiac or otological abnormalities [5].

The etiology is still unknown, and it is not clear whether it is from genetic or environmental causes. Most cases are sporadic, although some authors suggest an autosomal dominant inheritance in some cases, with an incomplete penetrance and variable expressivity [6]. This patient does not have any affected relative.

Our case was a type II MRKH syndrome that necessitated laparoscopy to complete the diagnosis, confirming that this is the gold standard technique for such cases [7]. Despite the important role of imaging techniques (US, MRI) in providing a detailed map of the pelvic anatomy, which includes the presence and the extent of the anomalies of the pelvic structures [7], in our case these techniques failed to identify the high placed, anatomically normal ovaries.

The infertility remains the most significant problem in the MRKH syndrome. Assisted fertility techniques, including surrogacy, enable women without a uterus to have genetic offspring [3]. This is why the genetic characterization of the syndrome is of major importance for the exclusion of other syndromes that clinically mimic the MRKH, but because of the absence of normal gonads, is not compatible with assisted reproduction.

Conclusion

Suspicion is the key to diagnosing and in our case, the diagnosis of MRKH was suspected based on history, physical examination, US and MRI evaluation. Even if the secondary sexual characteristics and the endocrine status indicated the presence of ovarian tissue, we were unable to proper localize these genital organs using advanced imagistic techniques. Complete evaluation of these patients is advisable, including laparoscopy, as the normal sexual function is achievable following surgical treatment, and reproduction is possible using assisted techniques. Proper counselling and management would reduce the emotional impact related to this condition, that otherwise may have psychologically devastating consequences.

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