Case 2894

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: Ultrasound findings in two cases

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Patient: 18 years, female

Clinical History:

Two female patients presented with primary amenorrhea.

Imaging Findings:

This report pertains to two cases. CASE 1 concerned an 18-year-old woman who was referred to the Radiologic Department of Aretaieion Hospital, University of Athens, for primary amenorrhea. A clinical examination when done revealed a blind 3 cm vaginal pouch with normal external genital organs. No other abnormalities were identified. An abdominal ultrasound of the pelvis revealed a 12 cm pelvic kidney behind the urinary bladder, with its main artery originating from the RT common iliac artery (Fig. 1) and normal Doppler waveforms both in the renal and intrarenal arteries. The contralateral kidney, the uterus and the cervix were absent (Fig. 2). Both the ovaries were identified to be normal in appearance. No other cause for amenorrhea was revealed (pituitary or hypothalamic dysfunction) and a normal karyotype (46, XX) was identified. The MRKH syndrome was considered to be the correct diagnosis. CASE 2 concerned a 42-year-old nun who was referred to our department for bland pelvic pain that had start over the past two months. From the patient's history, it was discovered that she suffered from a hearing loss in the right ear and primary amenorrhea, but she had never been referred to a gynecologist before. On presenting to our institution, a gynecologic examination was done, which revealed a blind 2 cm vaginal pouch with normal external genital organs and no other obvious abnormalities. An abdominal ultrasound that was performed revealed a 10.5 cm extrarotated right-sided pelvic kidney with the absence of the contralateral one. Its main arterial supply was from the right common iliac artery (Fig 3). Behind the urinary bladder, a solid mass of a 74.3 mm maximum diameter was found, which was considered to be a residual component of the uterus (Fig. 4). Only the right ovary was identified to be normal in appearance. Laboratory findings were also normal. The patient underwent an operative laparotomy for resection of the mass. A pathologic examination of the specimen revealed a solid mass of leiomyomas derived from the uterus anlage.

Discussion:

Many congenital malformations of the female genital tract may not manifest themselves until adolescence and they appear with various clinical symptoms during the second decade of life, particularly following the onset of puberty. The Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome occurs in 1 in 4000–5000 female births. It is defined by a maldevelopment of the Mullerian ducts, at any time between its origin and fusion with the urogenital sinus, which results in the congenital absence of the vagina and the uterus or the presence of residual components such as a blind vaginal pouch and a rudimentary anlagen of the uterus (nonfunctioning myometrial tissue), which are noted in 25% and 90% of these patients, respectively (1). Many studies give controversial results trying to explain the way of
inheritance of the MRKH syndrome (2). The simultaneous presence of other congenital abnormalities and the fact that these abnormalities have a recurrence risk of 1% to 2%, leads to the conclusion that the cause of the syndrome is polygenic and multi-factorial. Women are usually referred for evaluation due to primary amenorrhea. They present with normal secondary sexual characteristics, ovarian function, distal fallopian tubes, endocrine status and karyotype (46, XX) (3). Clinical and imaging findings show that there are two forms of the MRKH syndrome. The first group consists of patients with a congenital absence of the vagina and a uterus, while the second group of patients present with the congenital absence of the vagina and uterus and also with renal (pelvic kidney, horse-shoe kidney, unilateral renal agenesis, polycystic kidney disease), ear (unilateral or bilateral hearing loss) and skeletal abnormalities (scoliosis, spina bifida, Klippel- Feil syndrome). As far as the skeletal and ear abnormalities are concerned, they accompany 10%–12% and 10% of patients, respectively (2). Other abnormalities that may associate with the MRKH syndrome are disorders in galactose metabolism, congenital heart disease, inguinal and femoral hernias, cleft palate, situs inversus-lateral transposition of the organs of the thorax and abdomen, leiomyomas of the uterine remnants and endometriosis (3, 4). The therapeutic approach of these patients deals with the management of their congenital anatomical abnormality with the need to be sexually active and fertile, and the psychological impact of the knowledge that these individuals have no vagina or uterus (2). The American College of Obstetricians and Gynecologists emphasizes the primary role of the non-surgical approach. Vaginal dilators are considered to be the first treatment of choice. The technique involved requires passive and graduated dilatation of the vaginal pouch. The results are very satisfying; thus, a well-motivated, well-supported patient can achieve a vaginal length which is totally satisfactory for intercourse in 85% of cases (2). In case of failure of a non-surgical approach, various surgical techniques offer good results. Surgical creation of a neovaginal space, vulvo-vaginoplasty, bowel vaginoplasty and Vecchietti’s operation are the surgical techniques which aim to create a functional vagina (5). Psychological input is of great importance, as it is extremely difficult, in a young patient, to deal with the diagnosis and treatment of the MRKH syndrome. The loss of their sexual identity and the ability to fulfill the female role in the future; creates a great emotional stress in these patients. Additionally, an adequate psychological assessment leads to greater success in any kind of treatment (2).

**Differential Diagnosis List:** Mayer–Rokitansky–Kuster–Hauser syndrome.

**Final Diagnosis:** Mayer–Rokitansky–Kuster–Hauser syndrome.

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