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CASE REPORT
ΕΝΔΙΑΦΕΡΟΥΣΑ ΠΕΡΙΠΤΩΣΗ

Inguinal hernia containing fallopian tube and ovary in a 38-year-old woman with Mayer-Rokitansky-Küster-Hauser syndrome

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome has an incidence of 1/4,500 female newborns. It is characterized by congenital aplasia or hypoplasia of the uterus, cervix and upper part of the vagina in females showing normal development of secondary sexual characteristics and a normal 46, XX karyotype. Can a hernia be the initial diagnostic sign? We report a case where an inguinal hernia containing fallopian tube and ovary in a 38-year-old woman led to further diagnostic investigations which established the diagnosis of MRKH. A literature review of the MRKH syndrome is also included.

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ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2020, 37(2):247–250

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Βουβωνοκήλη με περιεχόμενο σάλπιγγα και ωοθήκη σε γυναίκα 38 ετών με σύνδρομο Mayer-Rokitansky-Küster-Hauser

Περίληψη στο τέλος του άρθρου

Key words

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In women, inguinal hernia with a sac containing fallopian tube and ovary is an uncommon finding, with an incidence of only 2.9% in operable cases of inguinal hernia.[1]

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome describes women with normal female external development and a normal female karyotype (46, XX), but internally with absent paramesonephric (Müllerian) ducts, resulting in congenital aplasia or hypoplasia of the uterus, cervix and upper two-thirds of the vagina.[2] It may occur as a purely genital malformation (type 1), but may also be associated with other malformations (type 2 or MURCS association).

MURCS association is the combined occurrence of Müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia. The incidence of the syndrome is 1/4,500 female newborns.[3,4] Although the syndrome is usually revealed when primary amenorrhea is observed or attempts at coitus fail, here we report a rare case where the hernia was the primary diagnostic sign.

CASE PRESENTATION

A 38-year-old woman with a right inguinal hernia and an umbilical hernia was admitted for elective repair of the hernias. Her past medical history included scoliosis and congenital aplasia of the left kidney. Concerning her gynecological history, menstruation started at the age of eleven, with an irregular menstrual cycle; dyspareunia was reported, but, most importantly, she had a successful pregnancy at the age of 36 years. Her family history included no known medical issues.

On physical examination, her height was 1.50 m, and there was normal development of secondary sex characteristics with normal pubic hair. She had a palpable inguinal hernia in the right groin and an umbilical hernia. The rest of the physical examination was unremarkable and she had a normal preoperative encephalogram (ECG).

Surgical exploration of the right inguinal canal revealed an indirect inguinal hernia, and during palpation of the hernial sac an unusual content was suspected. The hernial sac was therefore opened anteriorly, where the ovary and part of the right fallopian

tube were identified as the hernial contents (figures 1, 2). With careful handling, the ovary and the right fallopian tube were reduced into abdominal cavity. The sac then was closed with continuous sutures vicryl 2.0 and the hernia repair was completed using the Lichtenstein technique.[5] Surgical repair of her umbilical hernia was also performed.

The patient was discharged on the second post-operative day and was seen in the clinic 10 days after the procedure with no post-operative complications, at which time the sutures were removed.

Magnetic resonance imaging (MRI) of the abdomen and pelvis revealed a solitary left kidney, a thickened junctional zone in uterus, which was suggestive of possible adenomyosis, an indistinct right ovary and a normal left ovary (figures 3–5). The spleen, liver and adrenal glands were normal. These findings, along with her scoliosis, were consistent with the diagnosis of MRKH syndrome.

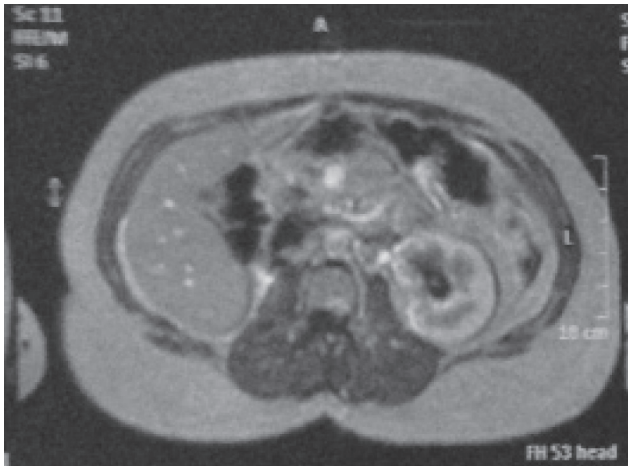
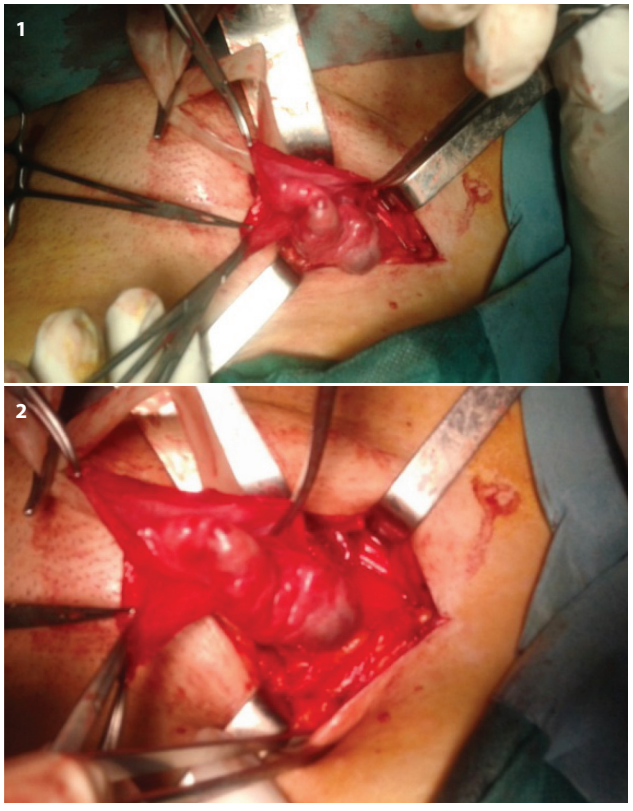


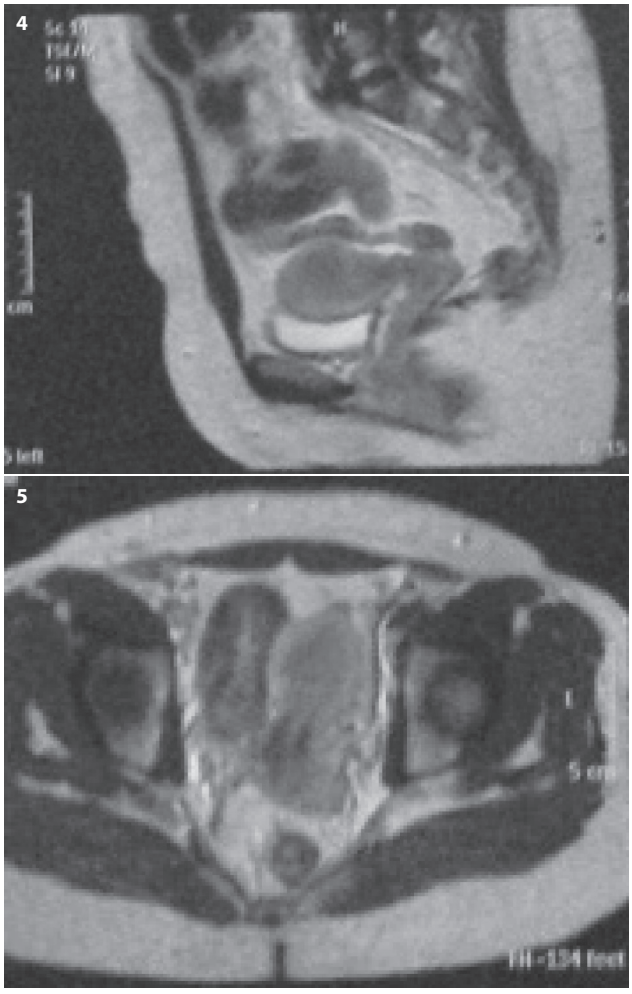
Figure 3. Mayer-Rokitansky-Küster-Hauser syndrome in a 38-year-old female. Abdominal computed tomography (CT) scan showing the solitary left kidney.

DISCUSSION

The MRKH syndrome is characterized by congenital aplasia or hypoplasia of the uterus, cervix and upper part (2/3) of the vagina in women who show normal development of secondary sexual characteristics and a normal



Figures 1, 2. Mayer-Rokitansky-Küster-Hauser syndrome in a 38-year-old female. Intra-operative photograph showing the right ovary and part of the right fallopian tube in the right inguinal hernial sac.



Figures 4, 5. Mayer-Rokitansky-Küster-Hauser syndrome in a 38-year-old female. Pelvic computed tomography (CT) scan showing thickened junctional zone in the uterus suggestive of possible adenomyosis, indistinct right ovary and normal left ovary.

46, XX karyotype. Type 2 MRKH syndrome and MURCS association show additional malformations which includes renal abnormalities (i.e., unilateral agenesis, ectopia of the kidneys or horseshoe kidney), skeletal and vertebral malformations (Klippel-Feil anomaly; fused vertebrae, mainly cervical; scoliosis); hearing defects, such as auditory problems or deafness are present in 10–25% of MURCS patients, and more rarely, cardiac and digital anomalies (syndactyly, polydactyly).[6]

The incidence of the syndrome is 1/4,500 female newborns. The precise pathogenetic mechanism is still unknown, but MRKH syndrome may be attributed to an initial defect in the intermediate mesoderm, leading, by the end of the fourth week of fetal life, to an alteration of the blastema of the cervico-thoracic somites and the pronephric ducts (which subsequently induce the differentiation of the mesonephron and the Wolffian and Müllerian ducts).[1,4]

A single case of the syndrome was first described by the German anatomist and physiologist Mayer in 1829. Later, in 1910, Küster published the first review of the syndrome. In 1938 Rokitansky published a case report of the syndrome and it was only 1961 that a gynecologist named Hauser first described the syndrome with its current name.[7] In 1979, Duncan and colleagues associated malformations of the renal system and the skeleton with the MRKH syndrome and proposed the term MURCS association.[8]

In 2005, the Vagina, Cervix, Uterus, Adnexa-associated Malformation (VCUAM) classification was introduced to permit an accurate description of the genital and associated malformations. This classification was the product of a study based on the clinical evaluation of 290 women with the MRKH syndrome. According to this study, complete atresia of vagina and bilateral atresia of cervix were found in all of the patients, and bilateral rudimentary or aplastic uterine horns were found in 84.2% of women. Normal adnexae were found in 87.3% and associated malformations were found in 126 of 282 women evaluated (44.7%), and in particular, 84 women (29.6%) had malformations of the renal system.[6]

The diagnosis is based on identification of characteristic symptoms, a detailed patient history, physical examination, ultrasound (U/S), diagnostic laparoscopy and magnetic resonance imaging (MRI). Most patients present with primary amenorrhea, infertility, dyspareunia and possible cyclic abdominal pain. External examination reveals completed puberty, with normal secondary female sexual characteristics and normal external genitalia. Anatomical examination is important. MRI is a non-invasive technique that provides a more sensitive and more specific means of diagnosis than U/S. MRI allows accurate evaluation of the uterine aplasia and clear visualization of the rudimentary horns and ovaries, and can be used at the same time to search for associated renal and skeletal malformations. Typical MRI findings are bilateral uterine buds connected by fibrous band-like structures, which converge in the midline triangular soft tissue lying above the bladder dome.[9,10]

Karyotyping may be performed, on which females with MRKH syndrome have a karyotype of 46, XX with no visible chromosomal modification. Additional tests for investigating associated malformations include spine X-ray, audiography, and heart echography.[11]

ΠΕΡΙΛΗΨΗ

Βουβωνοκήλη με περιεχόμενο σάλπιγγα και ωοθήκη σε γυναίκα 38 ετών με σύνδρομο Mayer-Rokitansky-Küster-Hauser

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Το σύνδρομο Mayer-Rokitansky-Küster-Hauser (MRKH) είναι ένα κληρονομικό σύνδρομο με επίπτωση 1 στις 4.500 γεννήσεις και χαρακτηρίζεται από απλασία ή υποπλασία της μήτρας, του τραχήλου και του ανώτερου τμήματος του κόλπου σε γυναίκες με φυσιολογικό φαινότυπο και δευτερογενή χαρακτηριστικά φύλου, καθώς και με φυσιολογικό καρυότυπο 46 XX. Είναι δυνατόν το αρχικό διαγνωστικό εύρημα του συνδρόμου να είναι μια βουβωνοκήλη; Παρουσιάζεται περιστατικό όπου η ανεύρεση ωοθήκης και σάλπιγγας εντός βουβωνοκήλης σε γυναίκα 38 ετών μάς οδήγησε σε περαιτέρω διαγνωστικό έλεγχο ώστε να θέσουμε τη διάγνωση του συνδρόμου MRKH. Επί πλέον, περιλαμβάνεται σύντομη βιβλιογραφική έρευνα σχετικά με το σύνδρομο MRKH.

Λέξεις ευρετηρίου: MRKH, MURCS, Ωοθήκη-βουβωνοκήλη

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