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Mayer-Rokitansky-Küster-Hauser syndrome presenting as vaginal atresia: report of two cases

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SUMMARY: **Mayer-Rokitansky-Küster-Hauser syndrome presenting as vaginal atresia: report of two cases.**

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Mayer-Rokitansky-Küster-Hauser syndrome (MRKH syndrome) is characterized by Müllerian duct structures agenesis: the vagina atresia is the commonest variant. There can be some anomalies associated, such as renal, skeletal, spine malformations and others. Patients with MRKH can show different presentation from newborn period to adolescence.

We report our experience in treatment of the vaginal atresia presenting in two young girls as a sign of MRKH syndrome.

RIASSUNTO: **Atresia vaginale in sindrome di Mayer-Rokitansky-Küster-Hauser: esperienza di due casi.**

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La sindrome di Mayer-Rokitansky-Küster-Hauser (sindrome MRKH) è caratterizzata da agenesia delle strutture del dotto di Muller: l'atresia vaginale rappresenta la variante più comune. Vi possono essere associate anomalie renali, scheletriche, spinali o altre. I pazienti con sindrome MRKH possono presentarsi con caratteristiche diverse dalla nascita all'adolescenza.

Riportiano la nostra esperienza nel trattamento dell'atresia vaginale come manifestazione della sindrome di MRKH in due giovani donne.

KEY WORDS: Vaginal atresia - Embryogenic abnormalities - Vaginoplasty.
Atresia vaginale - Anormalità embriogeniche - Vaginoplastica.

Introduction

Mayer-Rokitansky-Küster-Hauser syndrome is a disease diagnosed every year in 1/4500 female live births, whose etiology, despite the existence of many theories, remains unknown. It consists of congenital absence of upper vagina and uterus but the syndrome may be associated with other embryogenic abnormalities as well as unilateral renal agenesis or dysplasia and skeletal malformations. External genitalia and secondary sex characteristics are normal in most cases. The MRKH syndrome, when described in familial aggregates, seems to be transmitted as an autosomal dominant trait with an incomplete degree of penetrance and variable expressivity.

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At the time of expected menarche, most patients with vaginal agenesis refer to the physician for absence of menstrual bleeding and are found to have vaginal agenesis or hypoplasia during the physical examination. Treatment of vaginal agenesis is the vaginoplasty, i.e. is the creation of a neo-vagina that could be satisfying in shape, function and sensitivity.

Case reports

Case n. 1

In January 2004 a twenty years old woman presented in our department with the diagnosis of suspected MRKH syndrome. Family history was negative. Her medical history was unremarkable too. She reported us that, when she was fifteen years old, she asked for medical investigation because she hadn't no period at all. She was advised to undergo to an ultrasound abdomen examination and a pelvic magnetic resonance: both the clinical tests showed the presence of an infant uterus. Later she began an hormonal therapy, but she interrupted it after few months because she didn't tolerate it.

During the hospital stay in our department she underwent again to a pelvic magnetic resonance that confirmed the vagina

and uterus hypoplasia. Moreover she underwent also to an X-ray urography that was negative for any kind of anomalies. During the gynaecologic evaluation, a rudimental uterus i.e. two small areas within the uterus region, was appreciated. We decided to perform also the analysis of her karyotype that resulted normal.

In March 2005 she underwent the surgical operation of vaginoplasty for the confection of a neo-vagina using a skin layer from the big right gluteus (Mc Indoe's method). In the neo-vagina a tutor made of foam rubber was inserted.

The post-operative period was good; there weren't any surgical complication; in XVI day she was discharge. She was advised to practice daily vaginal gym using a lubrificated tutor.

Patient underwent periodically outpatient examination. At the moment she is in good general health conditions and she assures us that her sexual activity is satisfactory.

Case n. 2

In December 2005 an eighteen years old woman presented in our department with the diagnosis of suspected MRKH syndrome. Family history was negative. Her medical history was negative too. When she was sixteen years old, she decided to undergo a gynaecologic evaluation because of primary amenhorrea. After a period of hormone-therapy without any result, the specialist requested an isteroscopy examination. It showed the absence of the vagina. Then she underwent a diagnostic laparoscopy, that evidenced a rudimentary uterus with the partial presence of the histmus region, tortuous uterine tubes and regular annexa. It seemed a situation compatible with MRKH syndrome.

During the hospitalizayion she underwent a magnetic resonance that confirmed the laparoscopic data. An urography was negative for any kind of anomalies. We decided to perform also the analysis of her karyotype that resulted normal.

In January 2006 she underwent the surgical operation of vaginoplasty using the Mc Indoe's method. The postoperative period was uneventfull, and in IV postoperative day she was discharged. She came back ten days later for a control and to take out stitches.

This girl too was adviced to practice daily vaginal gym with a lubrificated tutor and underwent periodically outpatient examination.

Actually she is in good health and her sexual activity is satisfactory.

Discussion

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH syndrome) is characterized by Müllerian duct structures agenesis, vaginal atresia, rudimentary or absent uterus with normal ovaries and Falloppian tubes in females normal from a genetic (46XX), phenotypic and developmental features (1). It can be associated with renal, skeletal, spine anomalies or other malformations.

It is a congenital defect that occurs once every 5000 female live births (2). Patient with MRKH syndrome has a varied presentation from newborn period to adolescence. Falloppian tubes, uterus, cervix and upper vagina develop from Müllerian duct between 8th to 12th gestational week. The defects in development in this stage leads to agenesis of Müllerian structures. The development of kidneys, ureters, and bladder occurs at the same time (6th-12th week of gestation). Hence renal anomalies, such as renal agenesis, ectopic kidney, fused kidney, renal hypoplasia, and horseshoe kidney are seen in 30-40% of MRKH patients (3). Vertebral abnormalities are also found in 10% of patients (4). Other rare complications are cardiac anomalies and ano-rectal malformations; in particular, regarding the ano-rectal malformations, the Müllerian duct agenesis is frequently found with common cloacal anomaly.

Primary amenhorrea in an adolescent girl is the most usual presentation. Firstly, it is very important to perform the perineal examination which could be seen as a diagnostic test. In fact it shows either a vagina or a shallow and blind vaginal pouch, the part of vagina which develops from ectoderm. Secondly it is necessary that the patient undergoes instrumental examinations (ultrasonography, magnetic resonance, urography) to classify the syndrome and to find out the possible anomalies associated. The laparoscopy is the last chance and usually gives the confirmation and classification of MRKH, moreover it gives help in planning the reconstructive surgery.

Various surgical (5) and non surgical methods (6) have been proposed for the creation of a neo-vagina. The outcome of these procedures is often evaluated considering the anatomical and functional results. The anatomical success concerns an adequately sized vagina and the functional one refers to satisfactory sexual activity. The construction of a neo-vagina either with free skin grafts with mould or vascularised flaps has been a common method (7, 8). The most common complication that occurs with a skin neo-vagina is stenosis, that requires frequent dilatation. Moreover it can cause dispareunia because the neo-vagina has no secretion and lubrification.

Regarding the surgery, it is necessary to treat also the anomalies possibly associated to MRKH. For example, the vaginoplasty is done at the time of final anorectal reconstruction. In cases of cloacal anomalies, it is abdomino-perineal ano-rectal pull through with sigmoid vaginoplasty. If there is associated a rectovestibular fistula, a vaginoplasty is done at the time of anorectoplasty. The complex reconstructive procedures are usually performed at around 1 year of age.

Eventually another point to consider is counselling parents about the anatomical problems, associated anomalies, the method of reconstruction, and the risks (9, 10). The patient and her parents must know that will not be any menstruation and conception and that the problems regards mainly the sexual activity. It is not easy to accept this situation so very often a psychosocial rehabilitation is required.

In conclusion it is clear that women affected by MRKH syndrome could need to undergo to a complex vaginal reconstructive surgery and a detailed counselling about the function of menstruation and the fertility.

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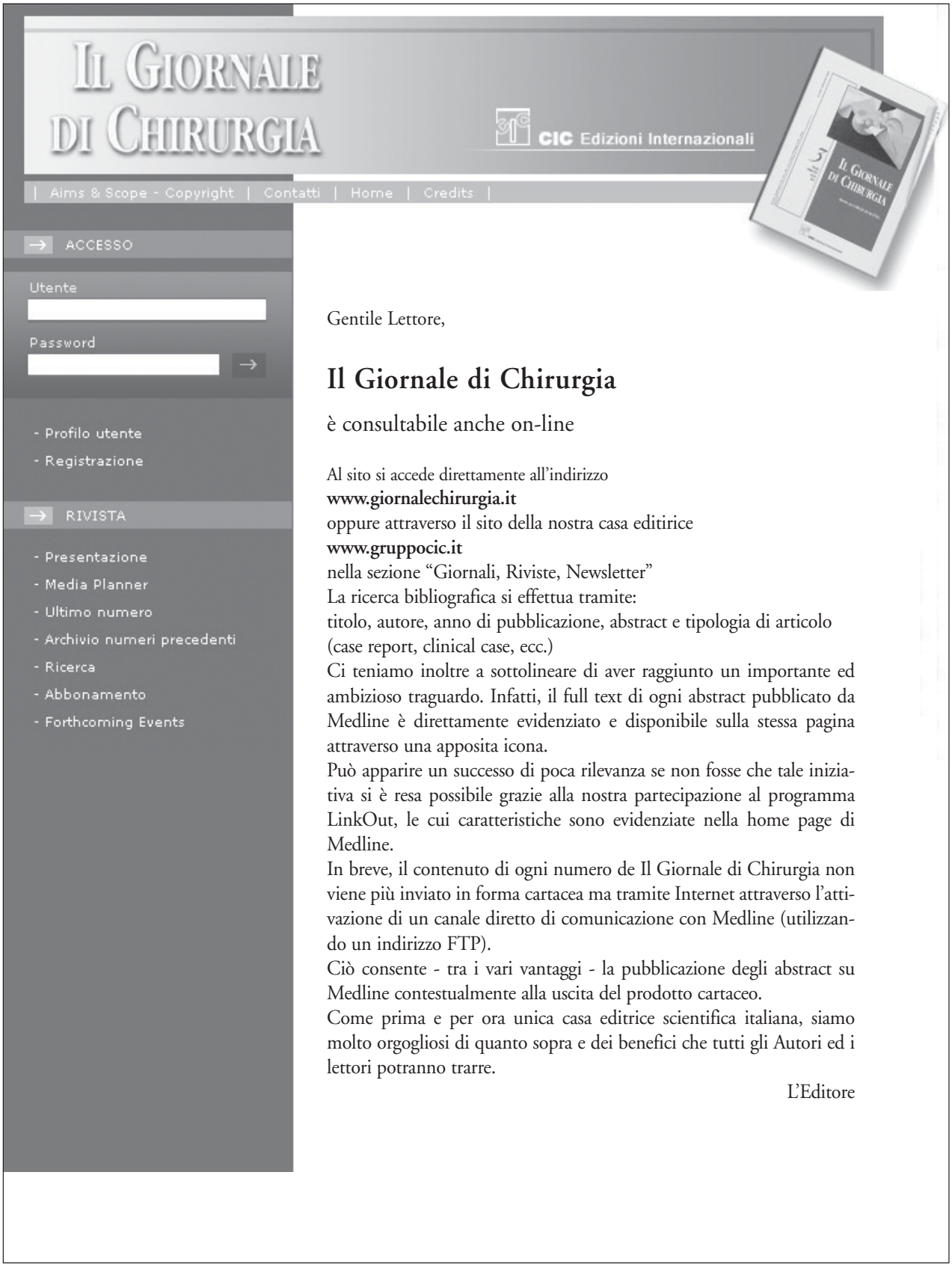

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