Original Research Paper

Gynaecology

MAYER-ROKITANSKY-KUSTER-HAUSER (MRKH) SYNDROME: A CASE
REPORT

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KEYWORDS:

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser syndrome is an uncommon condition, with an incidence of 1 in 4000- 5000 female births and is the second most frequent cause of primary amenorrhea after gonadal dysgenesis .The reproductive abnormalities of MRKH syndrome are due to incomplete development of the Müllerian duct which develops into the uterus, fallopian tubes, cervix, and the upper part of the vagina. The cause of the abnormal development of the Müllerian duct in affected individuals is unknown. This abnormality is characterized by congenital aplasia/hypoplasia of the uterus and the upper part (2/3) of vagina, in young women presenting otherwise with normal endocrine status. It may include absence or hypoplasia of the uterus and fallopian tubes. The patients present with normal secondary sexual characteristics, as the functional ovaries are present, but menstruation is absent.

Syndrome is classified into three types according to the involvement of other systems than reproductive system. The typical syndrome (type I) is represented by abnormalities restricted to the reproductive system. The second one (type II) is an atypical syndrome, with the presence of asymmetric uterine remnants and abnormal fallopian tubes. Such syndrome type may be associated with ovarian disease, congenital renal, bone abnormalities and hearing defects. A third one, the so called MURCS type, involves utero-vaginal hypoplasia or aplasia, renal, bone and cardiac malformations. Renal malformations include: unilateral agenesis, horse-shoe kidney, renal hypoplasia, ectopic kidneys and hydroureteronephrosis. Bone malformations occur particularly in the vertebrae, most commonly with vertebral fusion (particularly cervical vertebrae), Klippel-Feil syndrome and scoliosis. Cardiac alterations and digital alterations such as syndactyly and polydactyly.

CASE REPORT

A 19-year-old girl who presented with no onset of menstrual cycle (primary amenorhea). Her height was 148cm, weight 54Kg, and blood pressure 110/70mm Hg. No other affected members in the family were detected. At clinical examination, the patient has a development of secondary sexual characteristics compatible with her chronological age. Hormonal studies were in normal limit. At gynaecological examination a grooved urethra with elevated edges was observed.



Trans-abdominal ultrasonography did not demonstrate the presence of uterus. Therefore, a pelvic MRI examination was conducted. At magnetic resonance imaging, the uterus could not be visualized and the ovaries presented with good volume and without any abnormality. Also, the vaginal canal could not be visualized.

: MRI shows absent uterus and presence of ovaries



Genetic evaluation revealed karyotype 46 XX, thus determining the diagnosis of MRKH syndrome. A surgeon specialized in reconstructive surgery and a consultant psychologist was involved and no surgery was planned for the patient at this stage. The findings and implications regarding potential fertility and child bearing were explained to the patient. An interdisciplinary management in a specialized centre was planned. After initial evaluation, the patient can undergo vaginoplasty.

DISCUSSION

The typical clinical presentation of this syndrome is primary amenorrhea, in association with or without cyclic colicky pain, in an adolescent with secondary sexual characteristics compatible with age, with no sign of virilisation. Gynaecologic examination may detect either absence of the vaginal canal or vaginal shortening. Imaging studies such as ultrasonography and magnetic resonance imaging, in association with or without laparoscopy, are necessary to allow the determination of the anatomic characteristics of the syndrome. Ultrasonography is the initial method of choice. This method can demonstrate the absence of the uterus between the bladder and the rectum . Magnetic resonance imaging is the most sensitive and specific imaging method in the evaluation of this syndrome, not only for allowing the acquisition of multiplanar images, but also for allowing the acquisition of sequences with fat saturation. It allows a good definition of anatomical alterations such as uterine agenesis, as well as evaluating ovariesvagina and associated anomalies.

Laparoscopy is indicated only in cases where the evaluation by the two previous imaging methods is inconclusive . The final diagnosis is achieved by the association of the imaging findings with the presence of the karyotype 46, XX. The differential diagnosis should be made with other situations where the patient presents primary amenorrhea and normal secondary sexual characteristics, such as congenital absence of uterus and vagina, isolated vaginal atresia with androgeninsensitivity syndrome and transverse vaginal septum with imperforate hymen.

Because of the typical anatomic alterations, MRKS syndrome generates anxiety and psychological distress with consequences on the patient's quality of life, thus requiring a multidisciplinary approach The indicated anatomic treatment is the surgical or non-surgical creation of a neovagina, which may allow these patients to have a normal sex life . As the surgical approach is chosen, uterine remnants can be removed to avoid future endometriosis .Patients who want to have children should be encouraged to adopt, or the possibility of having biological children by means of surrogacy should be suggested, considering that the presence of functional ovaries in these women allow the production of normal ovules . Even with advanced management of this syndrome, its diagnosis causes significant psychological distress, affecting the patients' quality of life because of the absence of menstruation and impossibility of pregnancy. The distress caused by the diagnosis may be alleviated by surgical or non-surgical treatments, by the passage of time, by counselling, by family's support and by support groups.

CONCLUSION

MRKH syndrome is one of the most common causes of primary amenorrhea and Ultrasonography is useful for diagnosing any associated renal anomalies. MRI is more precise than USG and less invasive and expensive than laparoscopy, contributing significantly to treatment planning and patient management. Although this condition has psychologically devastating consequences today anatomical defects can be surgically treated allowing a normal sexual function and reproduction thanks to the assisted techniques. so correct evaluation of these patients and proper management is mandatory.

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