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Case Study

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MAYER ROKITANSKY-KUSTER-HAUSER SYNDROME: 3 CASES STUDY

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ABSTRACT

Mayer Rokitansky-Kuster-Hauser syndrome or utero-vaginal aplasia syndrome is a very rare birth defect. It is characterized by an absence of the uterus or a rudimentary uterus when the ovaries are normal. This anomaly is mainly represented by primary amenorrhea as well as unsatisfactory or even impossible sexual intercourse. The external genitalia and secondary sexual characteristics are normally developed, in particular breast growth and pubic hair growth. The karyotype is 46 XX, with no clearly visible chromosomal anomaly, and the hormonal balance is considered normal. The diagnosis is confirmed by imaging. This syndrome can be isolated (Type I) or associated with other renal, skeletal, auditory or cardiac malformations (Type II). The therapeutic attitude is very complicated, and should always start with non-surgical methods; it is only after failure that the surgical methods are indicated and requires an experienced medical team without forgetting the psychological support of the patient. Through our 3 cases, we will study Mayer Rokitansky-Kuster-Hauser syndrome with a review of the literature.

KEYWORDS: Mayer Rokitansky-Kuster-Hauser syndrome (MRKH syndrome), isolated form, imaging.

INTRODUCTION

Mayer Rokitansky-Kuster-Hauser syndrome or utero-vaginal aplasia syndrome is a very rare congenital malformation with an estimated incidence of one out of 4,500 women.[2] It is characterized by an absence of the uterus and of the upper two thirds of the vagina while the ovaries are normal. This anomaly manifests itself mainly by primary amenorrhea as well as unsatisfactory or even impossible sexual intercourse. The external genitalia and secondary sexual characteristics are normally developed, in particular breast growth and pubic hair growth. The karyotype is 46 XX.

This syndrome, of unknown etiology, can be isolated (Type I) or associated with other renal, skeletal, auditory or cardiac malfunctions (Type II).

In this study, we report 3 cases of young girls all with Type I Mayer-Rokitansky-Kuster-Hauser syndrome.

MATERIALS AND METHODS

Clinical Case 1: This is a 16-year-old patient, with no particular medical history, who consults for primary amenorrhea and in whom clinical examination finds very developed sexual characteristics. Pelvic ultrasound hasn't visualized neither the uterine cavity nor the ovaries. The MRI (figs. 1 and 2) was then performed, showing a very diminished uterus measuring 17 x 10 mm with ovaries with normal size, normal morphology and normal follicles. The kidneys appeared normal. The karyotype and the hormonal balance were not made.

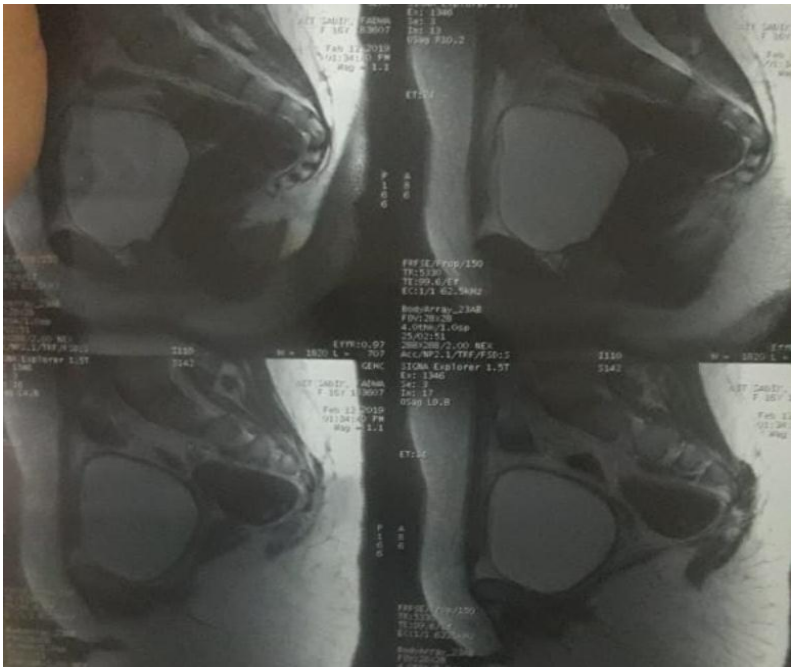

Fig. 1: Sagittal cutting of a magnetic imaging wich shows the absence of vagina and hypoplasic womb.

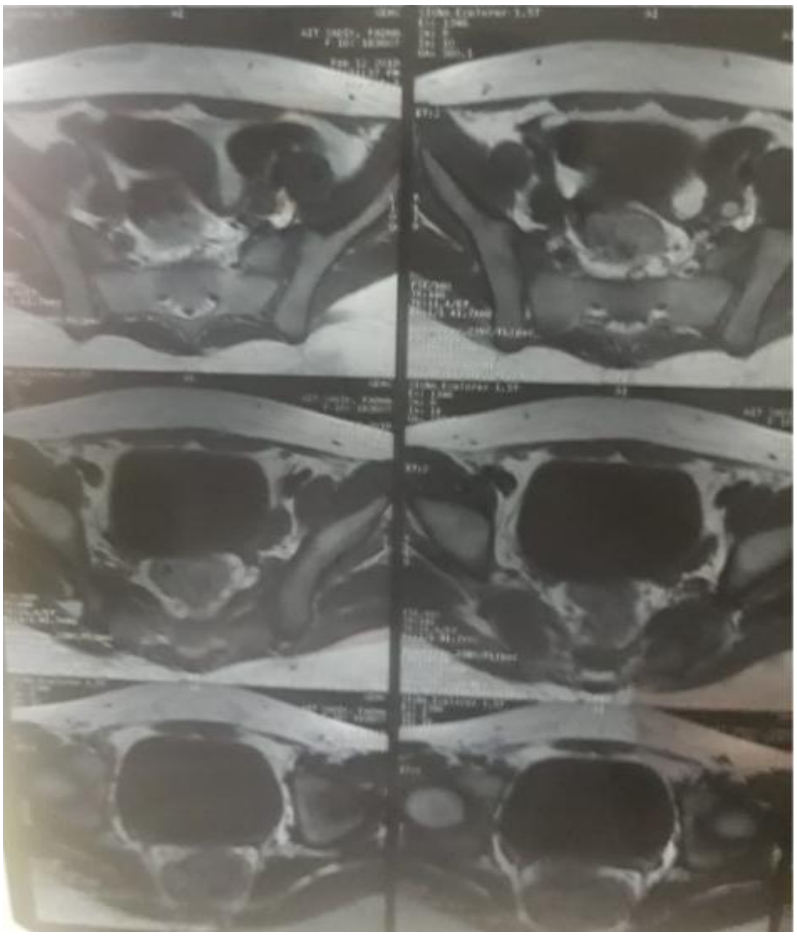

Fig. 2: Cross sections of magnetic imaging shows severe hypoplasia of womb.

Clinical Case 2: This is a 24-year-old patient, with no particular medical history, admitted to the emergency room on her wedding day for hymenal imperforation and who presented primary amenorrhea but was never consulted. The clinical examination found secondary sexual characteristics developed with a gynecological examination showing the absence of the hymen and the vagina. The MRI (figs. 3 and 4) was performed

objectifying a hypoplastic uterus measuring 18 mm with no visualization of the vagina. Both ovaries and both kidneys are visualized normal. The karyotype produced is normal. The hormonal assessment was not done.

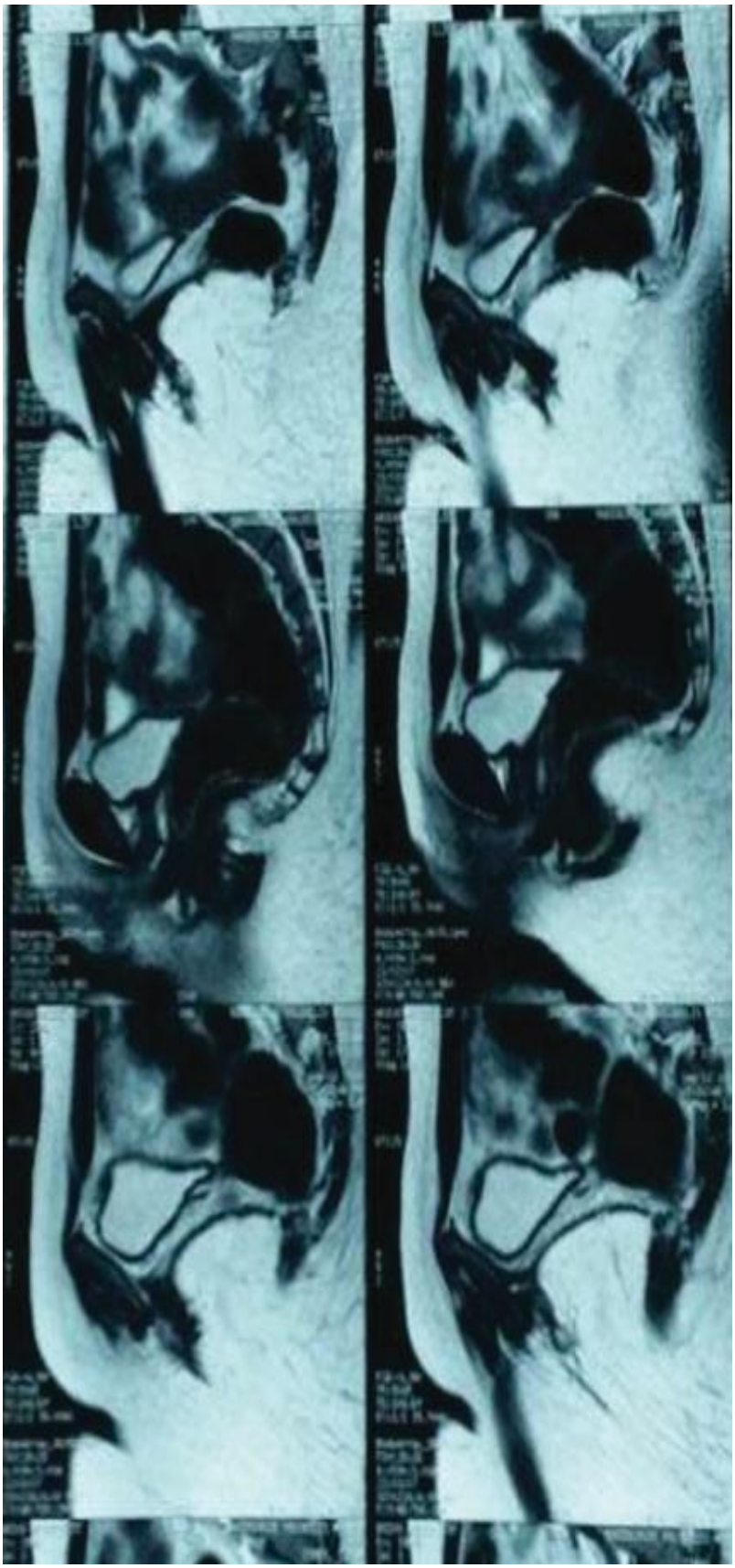

Fig. 3: Sagittal cutting of a magnetic imaging showing hypoplasic womb and vaginal aplasia.

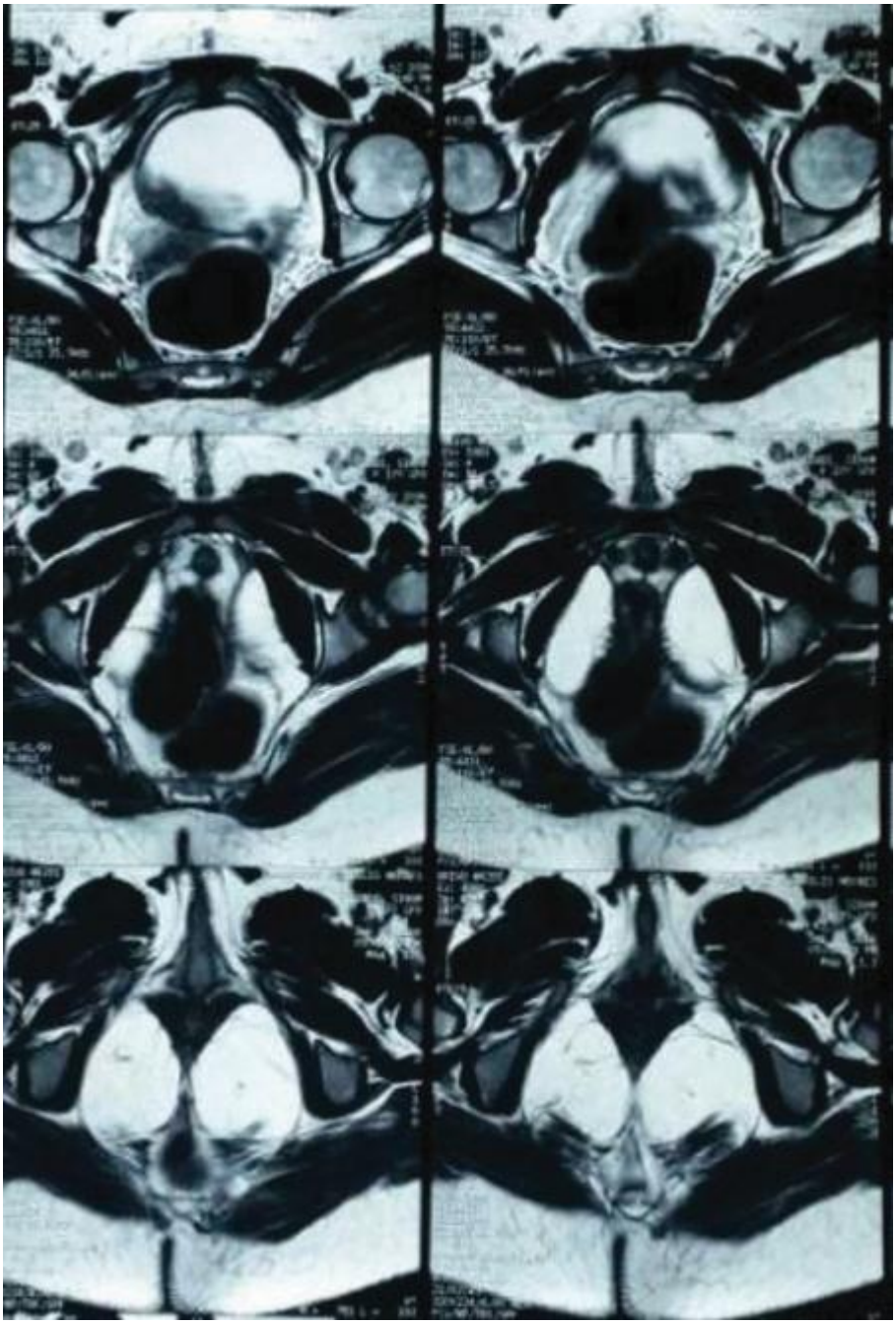

Fig. 4: Cross sections of magnetic imaging shows hypoplasic womb.

Clinical Case 3: This is a 19-year-old patient, with no remarkable pathological history, who has shown for a prenuptial consultation and who presented a primary amenorrhea never explored. The clinical examination found the sexual characteristics poorly developed and a malformed hymen found during gynecological examination.

Pelvic ultrasound (fig. 5) showed a reduced uterus to a fibrous band of 7 mm in anteroposterior diameter and 25 mm in length. The ovaries are in place and in normal size and present the site of some follicular images. Kidney ultrasound found no associated kidney defects.

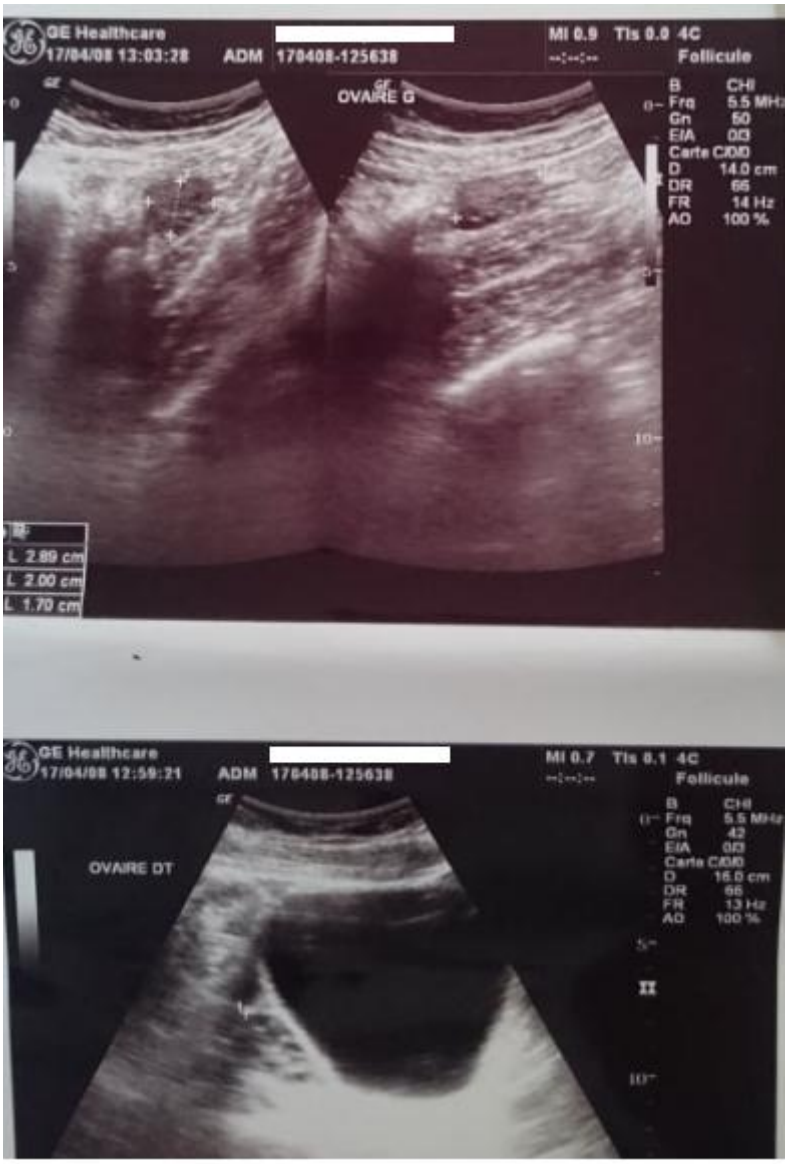

Fig. 5: Pelvic ultrasound shows normal ovaries and hypoplastic womb.

RESULTS AND DISCUSSION

Mayer Rokitansky Kuster Hauser's syndrome is a rare congenital utero-vaginal aplasia that manifests clinically essentially as primary amenorrhea in girls with normal secondary sexual characteristics; because the ovaries are normal, and karyotype is normal.

From an embryological point of view, utero-vaginal aplasia is caused by early discontinued development of the Mullerian canals at the 7th week of conception.[1]

Suprapubic ultrasound is requested as the first procedure since it is simple and non-invasive. It can show the absence of the uterus or a hypoplastic uterus. In this case, the search for associated renal malformations must be systematic.[2,3]

Magnetic resonance imaging is the complementary reference examination for the diagnosis of uterine malformations.[4] It is more sensitive and more specific than the suprapubic ultrasound.[2] It makes it possible to visualize uterine aplasia and vaginal aplasia as well as the presence of normal ovaries and helps in the search for other associated malformations, especially renal and osseous ones.[3]

Laparoscopy should only be indicated if there is any diagnostic doubt after performing the MRI.[3] Currently, it is mainly indicated in case of surgical treatment for the creation of a new vagina.[4]

Biologically, the blood karyotype is normal with no visible chromosomal abnormality. The endocrine check-up (FSH, LH, 17 B plasma estradiol) is normal, proving the integrity of the ovarian function.[2]

The announcement of the diagnosis of utero-vaginal aplasia is a psychological trauma because of the sterility due to the impossibility of sexual intercourse. A

psychological follow-up must therefore be offered to all cases.

Many surgical and non-surgical techniques are described in the literature. The treatment of vaginal aplasia must first use non-surgical techniques mainly Franck's method, if the vaginal cup allows it, which consists of the use of vaginal dilators produced by the patient herself allowing passive dilation of the vaginal cup.[4] Surgical methods should not be proposed until after Franck's method fail.[5] There are several techniques for creating a new vagina with the primary objective of providing the patient with satisfactory sexual function. Unfortunately, for the 3 cases that we reported, none of them have received therapeutic monitoring and no record was kept afterwords.

CONCLUSION

The Mayer Rokitansky Kuster Hauser in its typical form is very rare. Imaging is an essential step in establishing the diagnosis. Whatever the therapeutic technique used; special monitoring should be considered as well as psychological support.

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