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Rare cause of woman amenorrhoea: Mayer-Rokitansky-Küster-Hauser syndrome

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ABSTRACT

Uterine malformations are developmental abnormalities of the Müllerian ducts. It is a heterogeneous group of pathology and anatomy whose diagnosis must be accurate. Imaging plays an essential role in the diagnosis with 2D/3D ultrasound and magnetic resonance imaging (MRI) is the gold standard in accurate diagnosis. We reported two cases of uterine malformations about Mayer-Rokitansky-Küster-Hauser syndrome in a 20-year-old patient and 22-year-old patient. These anomalies are associated with renal malformations. The diagnosis was made by chance, by MRI which was performed in these patients. The management of these uterine malformations includes a psychological support component. There is a non-surgical treatment, but also, depending on the malformation, a surgical treatment. The complications of these malformations are dominated by recurrent miscarriages and infertility.

Keywords: Malformation, Mayer-Rokitansky-Küster-Hauser syndrome, MRI, Uterus

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INTRODUCTION

Uterine malformations are mostly the result of an abnormality in the development of the ducts of Müller during the process of genital cavity formation. They can occur at any of the three stages of organogenesis giving three types of anomalies. They are benign anomalies with a prevalence between 4% and 7% [1]. These malformations have an important impact on reproduction and are the cause of certain obstetrical complications and infertility. Imaging examinations remain essential for the accurate diagnosis of these malformations. 2D/3D ultrasound (US) remains the first-line examination, but MRI remains the gold standard for accurate diagnosis, with a view to optimal management. Several classifications exist but the most used are those of the American Fertility Society (AFS) and the European Society of Human Reproduction and Embryology (ESHRE) and the European Society for Gynaecological Endoscopy (ESGE): CORNUTA (congenital uterine anomalies) [1].

The definitive treatment is surgical in most cases. Psychological support is essential in the management of some of these malformations.

CASE SERIES

Case 1

A 20-year-old female patient with a history of primary amenorrhea and development of secondary sexual characters in whom a pelvic computed tomography (CT) scan revealed a pelvic kidney and suspected uterine agenesis. Pelvic MRI revealed: total uterine agenesis, inferior vaginal remnant, normal ovaries, left pelvic kidney (Figures 1 and 2). The diagnosis of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome was retained, classified as AFS 1/U5C4V4.

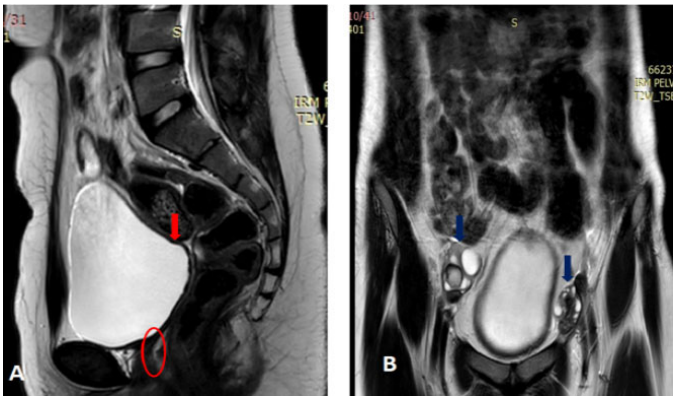

Figure 1: Pelvic MRI T2-weighted sagittal plane (A) showing uterine agenesis (red arrow), inferior vaginal remnant (red circle) and in coronal plane (B) normal ovaries (blue arrows).

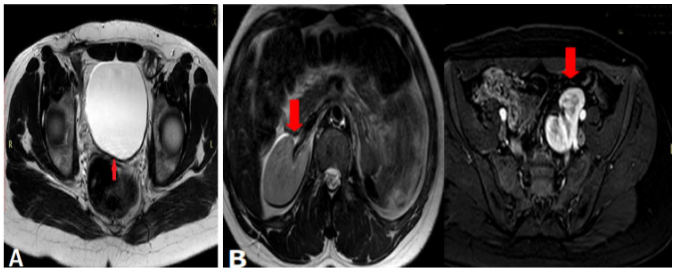

Figure 2: Pelvic MRI T2-weighted (A) axial plane showing uterine agenesis, without the image of the uterus between bladder and rectum (arrow), in the abdomen in (B) presence only of the right kidney (arrow), and T1 fat sat gadolinium the left pelvic kidney (arrow).

Case 2

A 22-year-old female patient with a history of primary amenorrhea and development of secondary sexual characters. A US scan was performed which suspected a uterine malformation. A pelvic MRI was ordered. Pelvic MRI revealed (Figures 3 and 4):

The presence of two distinct rudimentary uterine horns with functional endometrium: the right horn sits at the level of the right iliac fossa communicating with the homolateral ovary, the left horn sits next to the iliac vessels communicating with the left ovary. It is associated with uterine cervix and vaginal agenesis. Ovaries were normal.

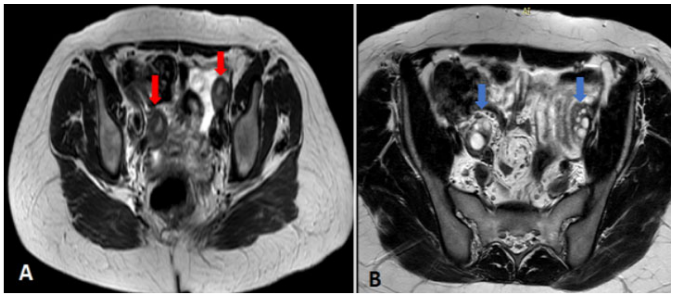

Figure 3: Pelvic MRI in T2-weighted axial plane showing in (A) two distinct rudimentary uterine horns with functional endometrium (red arrows) and in (B) normal ovaries (blue arrows).

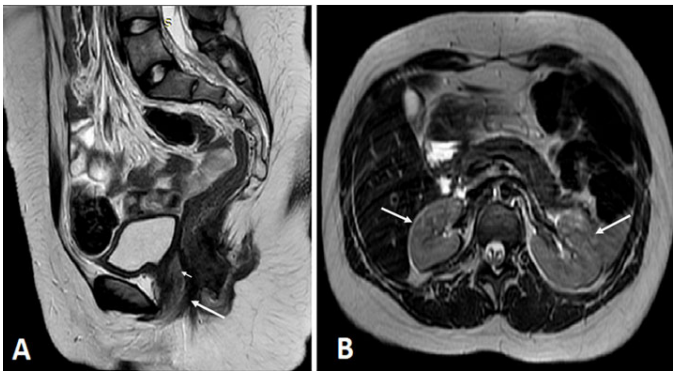

Figure 4: Pelvic MRI T2-weighted sagittal plane (A) revealed uterine cervix (small arrow) and vagina (arrow), and in the abdomen in axial plane (B) showing kidneys in normal topography.

The diagnosis of MRKH syndrome was retained, classified as AFS 1/U5C4V4.

DISCUSSION

The frequency of uterine malformations has a significant impact on reproduction, ranging from 0.5% to 47%. Mayer-Rokitansky-Küster-Hauser syndrome is rare with a frequency of 1/5000 women. In the general population, it varies between 0.5% and 4% [2]. The frequency rate of malformations in the fertile population is 3.8%, whereas it is 6.3% in the infertile population. These malformations are secondary to an anomaly in the development of the Müllerian ducts. They are therefore divided into three types according to organogenesis.

Depending on the stage of organogenesis [3]:

- Müllerian aplasia: MRKH syndrome
- Disorders of the fusion of the Müllerian ducts: bicornuate uterus
- Disorders of resorption of the intermüllerian septum: septate uteri

These anomalies are most often associated with anomalies of the urinary tree (renal agenesis, ectopic kidney) in 10–20% of cases [2]. In the infertile patient,

population malformations are described with a prevalence of 7% with uterus arcuatus, 34% with septated uterus, 39% with bicornuate uterus, 11% with uterus didelphis, 5% with uterus unicornis, and 4% with aplasia of the uterus [4].

These different malformations have led to classifications in order to harmonize understanding. The best known are those of the American Society of Reproductive Medicine (ASRM) [5] and The European Society of Human Reproduction and Embryology (ESHRE), and the European Society of Gynaecological Endoscopy (ESGE) [1] (Tables 1 and 2).

The circumstances of discovery vary: primary amenorrhea, dyspareunia, infertility, repeated miscarriages, systematic ultrasound findings (of an obstetric ultrasound), imaging, complications (uterine ruptures, placental anomalies) [6].

The gynecological examination has an important place in the diagnosis. It allows the detection of vaginal and cervical abnormalities in cases of amenorrhea. It must be meticulous in looking for malformations of the external genitalia, the vagina, and the cervix. It is not sufficient, however, and must be supplemented by imaging examine [7].

Table 1: Classification of Müllerian duct anomalies according to the AFS

Uterus anomaly		Cervical anomaly	Vaginal anomaly
Main class	**Sub-class**		
U0 – Normal uterus	**a** T-shaped **b** Infantilis **c** Others	**C0** – Normal cervix	**V0 –** Normal vagina
U1 – Dysmorphic uterus	**a** Partial **b** Complete	**C1** – Septate cervix	**V1** – Longitudinal non-obstructing vaginal septum
U2 – Septate uterus	**a** Partial **b** Complete **c** Bicorporeal septate	**C2** – Double normal cervix	**V2** – Longitudinal obstructing vaginal septum
U3 – Bicorporeal uterus	**a** With rudimentary cavity (communicating or not horn) **b** Without rudimentary cavity (horn without cavity/no horn)	**C3** – Unilateral cervical aplasia	**V3** – Transverse vaginal septum and/or imperforate hymen
U4 – Hemi-uterus	**a** With rudimentary cavity (bi- or unilateral horn) **b** Without rudimentary cavity (bi- or unilateral uterine remnants/aplasia)	**C4 –** Cervical aplasia	**V4** – Vaginal aplasia
U5 – Aplastic			
U6 – Unclassified malformations			

Table 2: Classification of female genital tract anomalies according to the ESHRE/ESGE

Class 1	Segmental hypoplasia/agenesis	Vaginal Cervical Fundal Tubal Combinated
Class 2	Unicornuate	Rudimentary horn with endometrial tissue – Communicating with the main uterine cavity – Not communicating with the main uterine cavity – Rudimentary horn without endometrial tissue – No rudimentary horn
Class 3	Didelphys	
Class 4	Bicornuate	Complete Partial
Class 5	Septate	Complete Partial
Class 6	Arcuate	
Class 7	Diethylstilbestrol drug related	T-shaped

The initial diagnostic method is the two-dimensional ultrasound (2D US), but also used are three-dimensional ultrasound (3D US), MRI, hysterosalpingo-contrast-sonography, X-ray hysterosalpingography, video hysteroscopy, and video laparoscopy [7].

For diagnostic purposes, 3D US remains the examination of the first choice. It has good reproducibility, a high level of agreement among different observers provides additional and more reliable images, and allows for the evaluation of the cervix and the vagina.

The second reference examination is pelvic MRI which is the gold standard in the detection of uterovaginal malformations. Magnetic resonance imaging offers objective and reliable tridimensional information about all the genital and peritoneal anatomy, except for the tubes; it can be used in all cases, including obstructive malformations. Magnetic resonance imaging can differentiate between a hypoplastic, non-functional uterine horn, and a non-communicating horn with functional potential, and it provides excellent noninvasive evaluation of complex anomalies such as MRKH syndrome or obstructed hemivagina ipsilateral renal agenesis syndrome. Magnetic resonance imaging is also ideal for accurate visualization of concurrent ovarian, renal, and lower urinary tract, and musculoskeletal abnormalities included in the imaged region.

The respective sensitivity and specificity are: Sensitivity: MRI 28.6–100%/3D US: 98–100%; Specificity: MRI 66–100%/3D US: 100% [8].

Mayer-Rokitansky-Küster-Hauser is a uterine aplasia and accounts for 3.4–10% of Müllerian anomalies. It is the most common form of Müllerian hypoplasia. The ovaries are normal and functional. In about 28% of these women, there is an ovarian malposition. Most girls have two rudimentary horns.

These are:

- symmetrical forms:
  - Major aplasia: absence of uterine horns
  - Limited aplasia: presence of canaliculated uterine horns and functional endometrium
- asymmetrical “hemi-rokitansky” forms
  - 1 side = typical or major MRKH
  - 1 side with limited or total/subtotal aplasia

They can be seen on MRI as bilateral small soft-tissue masses of intermediate to slightly hyperintense signal similar to the myometrium, located adjacent to the ovaries along the pelvic sidewalls and sometimes connected by a triangular band of intermediate-signal tissue.

Treatment varies according to the stage of diagnosis. It is essentially two-fold [4, 7]:

- Non-surgical methods: Franck: consists of posterior dilatation and requires regular sexual activity and is therefore limited to women with a sexual partner. It requires the cooperation of the patient.
- Surgical methods: consists of digestive transplants (hydrorrhea), cleavage by the lower route: vesicorectal cleavage, skin graft, and vaginal dilator, risk of urethral injury (McIndoe) or by the abdominal route (Vecchietti): transabdominal traction of an olive in the cup.

Surgery and psychological support play an essential role in the management.

CONCLUSION

Uterine malformations are a heterogeneous group of disorders secondary to abnormal development of the Müllerian ducts. They are classified into three groups. Imaging plays an essential role in the diagnosis by 3D US, followed by MRI, which remains the gold standard for diagnosis. They are most often associated with urinary and vaginal malformations. Mayer-Rokitansky-Küster-Hauser syndrome is the rarest. In order of frequency, septate uteri are more common, followed by bicornuate uteri. Management varies according to the type of malformation and surgery plays an essential role; psychological support is sometimes necessary for follow-up.

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Koudouhonon Rita Oze – Acquisition of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

ABOUT THE AUTHORS

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