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Utero-Ovarian Agenesis: One Case Report

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Abstract: The congenital malformation results from an abnormal development of the Müllerian ducts during embryogenesis. Primary amenorrhea and primary infertility are the main symptoms leading patients to seek medical consultation. Pelvic ultrasound remains the gold standard for diagnosis. The therapeutic approach is multidisciplinary, and treatment should only be considered once the patient has reached adequate awareness and emotional maturity.

Keywords: *Mayer-Rokitansky-Kusterhauser Syndrom, Infertility, Primary, Amenorrhea.*

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I. INTRODUCTION

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital malformation with an estimated prevalence of 1 in 5,000 female births [1]. It is characterized by uterine aplasia and absence of the upper two-thirds of the vagina, with a normal karyotype (46,XX) and normal secondary sexual characteristics reflecting normal hormonal function [2][3].

➢ *Two Main Clinical Forms of this Condition have been Described [4]:*

- The Typical form: characterized by vaginal agenesis and the presence of two rudimentary uterine horns connected by peritoneal folds, with normal ovaries and fallopian tubes. No extragenital anomalies are observed [5].

- The a Typical form: characterized by asymmetric uterine hypoplasia with or without fallopian tube dysplasia. This variant is often associated with other malformations, including renal, cardiac, or skeletal anomalies [6].

Another congenital cause of uterovaginal agenesis that represents the main differential diagnosis of MRKH syndrome is complete Androgen Insensitivity syndrome (CAIS), which presents with a 46,XY karyotype [7].

Before any therapeutic intervention, whether surgical or non-surgical, the patient must be fully informed and provide written consent, ensuring her understanding and agreement with the proposed treatment approach.

➢ *Observation*

17-year-old female patient was referred for pelvic MRI in the context of primary amenorrhea. Clinical examination was unremarkable, with normal development of secondary sexual characteristics.

A transabdominal pelvic ultrasound revealed no visible uterus or ovaries.

➢ *Pelvic MRI Findings:*

- No visualization of the uterus or ovaries
- No palpable mass
- No lymphadenopathy
- No pelvic effusion

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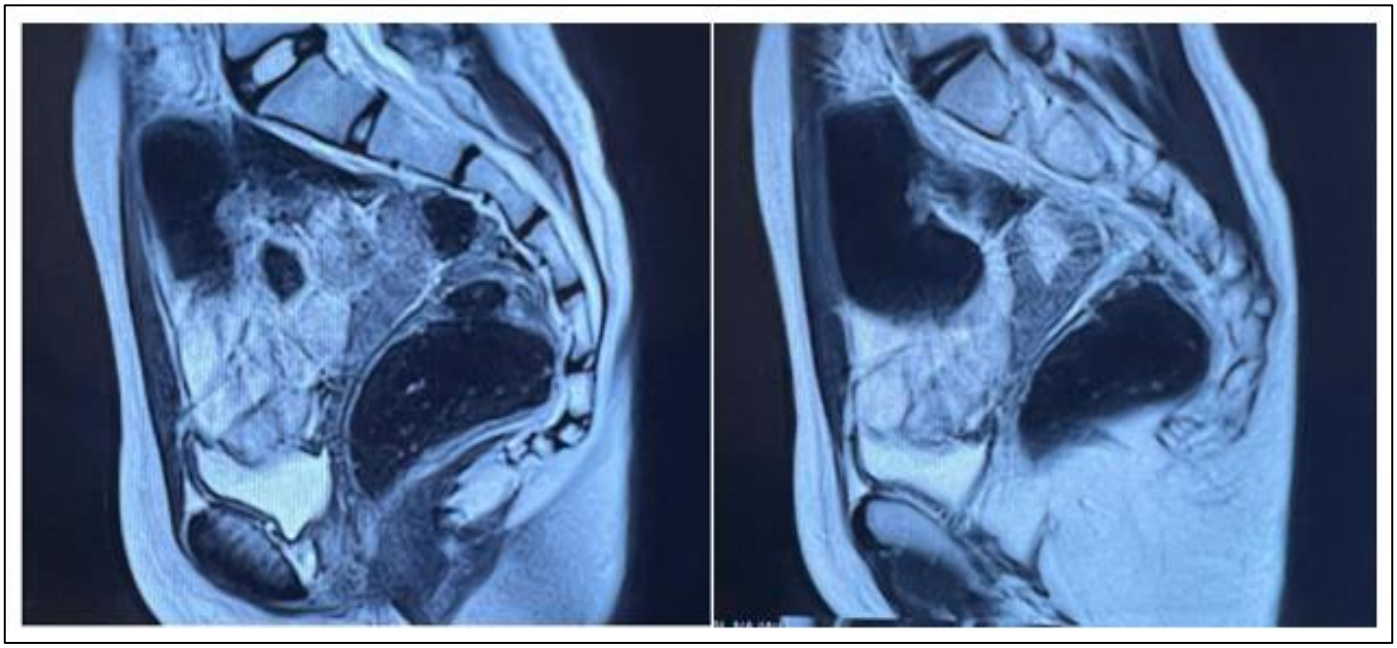

Fig 1 Sagittal MRI View Demonstrating Utero-Ovarian Agenesis

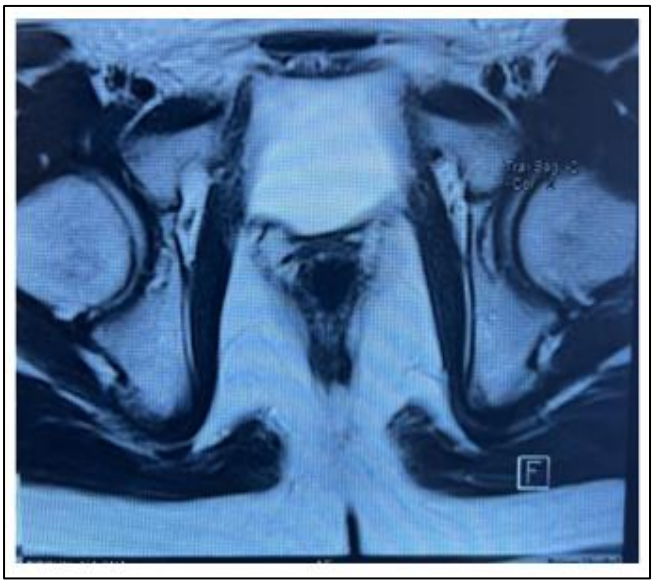

Fig 2 Axial View Showing no Visualization of the Uterus and Ovaries

II. DISCUSSION

Primary amenorrhea is often the initial clinical sign suggestive of a uterovaginal malformation. Gonadal dysgenesis is the most frequent etiology of these anomalies, typically leading to ovarian insufficiency despite normal pubertal development [8][9]. Congenital uterovaginal aplasia, commonly known as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a form of Müllerian agenesis [10], observed in individuals with gonadal agenesis and an XY or X0 karyotype [11].

Müllerian duct aplasia suggestive of MRKH syndrome has also been described in association with gonadal dysgenesis involving abnormal sex chromosome karyotypes affecting the X chromosome [12][13][14].

Ultrasound—whether transabdominal [15], transvaginal, transperineal, or transrectal—is currently the first-line imaging modality. Magnetic Resonance Imaging (MRI), however, offers higher sensitivity and specificity [16].

Laparoscopy is no longer considered a diagnostic tool, but rather a therapeutic option, mainly indicated in the context of vaginal reconstruction [17].

- *Three Main Therapeutic Approaches are Proposed:*

Psychological support, which is a crucial component of the therapeutic strategy, especially for adolescents seeking an active reproductive life and dealing with issues of self-esteem [18].

- *Non-Surgical Vaginal Reconstruction Techniques:*

FRANK's method, based on progressive dilation using vaginal dilators of increasing diameter [19].

- *Surgical Techniques, which may Involve:*

The use of prostheses [20],

- Rectovaginal space dissection followed by coverage using the pelvic peritoneum [21],
- Labial flap techniques, particularly when the neovaginal diameter exceeds 3 cm.

III. CONCLUSION

The therapeutic approach to uterovaginal malformations is complex, primarily due to the psychological impact on young adolescent patients, whose perception of femininity is often profoundly affected. This emotional dimension strongly influences the decision to pursue surgical or non-surgical interventions, as well as the management of associated infertility. Recent therapeutic advances have contributed to facilitating more personalized and holistic care.

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➢ *Ethics Commitee Authorization:*

Our institution does not find any conflict of ethics commitee.

➢ *Author Contribution:*

- Maha Lhaloui; Hassnaa Sarhane; Kaoutar Bahida; Nouhaila Yartaoui; Fatimazahra Belouazza; . performed surgery, paper writing and picture editing.
- Amina ETBR; NISRINE BENOUICHA,Aziz Baydada: Bibliography, written direction.

➢ *Guarantor*

- Maha Lhaloui.

➢ *Research Registration Number*

- No applicable data.

➢ *Conflicts of Interest Statement:*

- No conflict of interest have been announced by the authors for to this research.

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