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Mayer-Rokitansky-Küster-Hauser Syndrome: A Case Report

S. Taoufiki[1*], J. Hamdane[1], C. Ahmmana[1], I. Zouita[1], D. Basraoui[1], H. Jalal[1]

[1]Radiology Department, Mother and Child Hospital, CHU Mohamed VI Marrakech, Morocco

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***Corresponding author:** S. Taoufiki
Radiology Department, Mother and Child Hospital, CHU Mohamed VI Marrakech, Morocco

Abstract **Case Report**

Mayer-Rokitansky-Küster-Hauser syndrome is a rare congenital anomaly affecting the development of the female reproductive organs. Its etiology remains undetermined, but genetic and environmental factors may play a role in its development. It comprises two types, type I isolated and type II associated with renal, vertebral or cardiac anomalies. MRI is the examination of choice to confirm the diagnosis. In the light of a Mayer-Rokitansky-Küster-Hauser observation in a 26-year-old woman who consulted us for primary amenorrhea with pelvic pain. The purpose of this case report is to raise the possibility of a diagnosis of primary amenorrhea in a young woman with normal sexual characteristics.

Keywords: Primary amenorrhea, pelvic MRI, vaginal agenesis.

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital developmental disorder affecting the female reproductive organs. It is characterized by congenital aplasia of the uterus and vagina. Two clinical forms type I which corresponds to isolated uterovaginal agenesis and type II which is associated with other malformations.

OBSERVATION

This is a 26-year-old girl, with no particular history, who consulted for primary amenorrhea, pelvic pain and a cypho-scoliosis-type spinal deformity.

Clinical examination revealed external genitalia of female phenotype, well-developed breasts with imperforate hymen, hence the failure to perform a vaginal examination. Hormonal balance was normal.

Imaging:

Pelvic and cervico-dorso-lumbosacral CT and MRI scans showed:

Complete agenesis of the uterus and upper two-thirds of the vagina (Figure 1, Figure 2), agenesis of the right kidney (Figure 4) and pelvic ectopy of the left kidney (Figure 2, Figure 3, Figure 4), associated with exaggerated dorsolumbar curvatures and a closure defect of the posterior arch of C1 with vertebral blocks at D6-D7 and D10-D11, leading to the diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome type II.

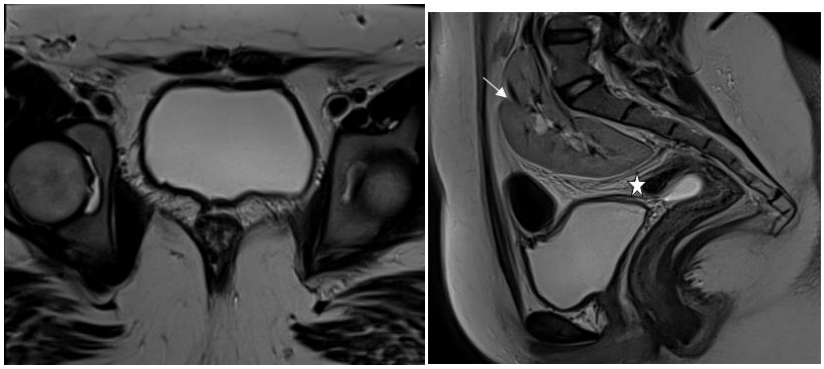

Figure 1 & 2: Pelvic MRI; axial and sagittal T2 sequences: uterine agenesis (star) with left kidney in pelvic position (arrow)

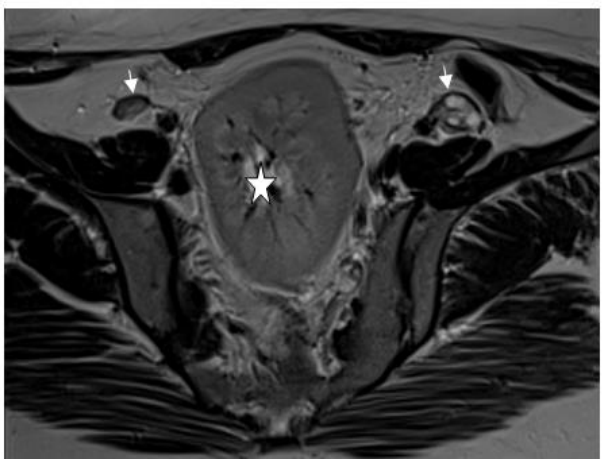
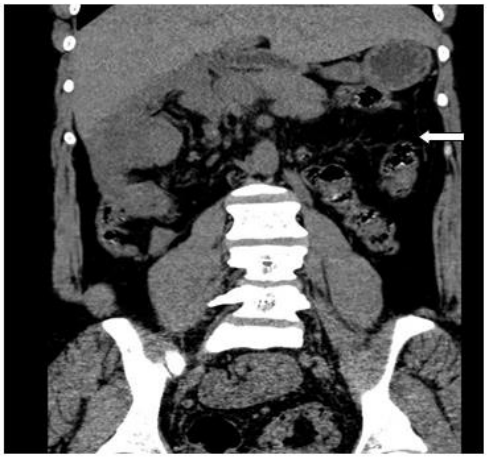

Figure 3 & 4: Left pelvic MRI: T2 Axial sequence: normal ovarian morphology and size (arrows), ectopic left pelvic kidney (star).right Abdominal CT coronal section shows empty left renal compartment (arrow)

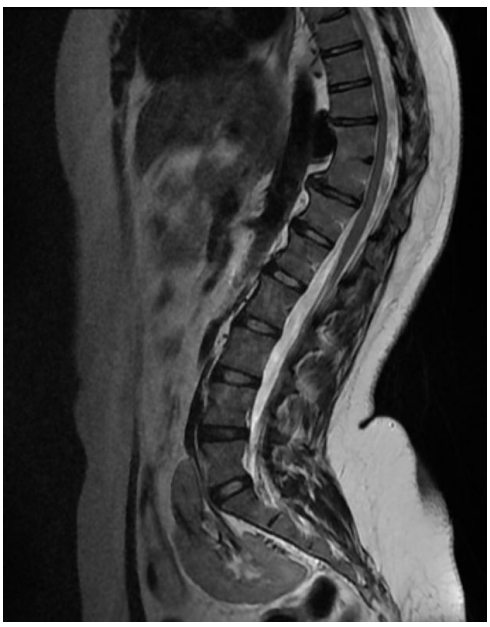
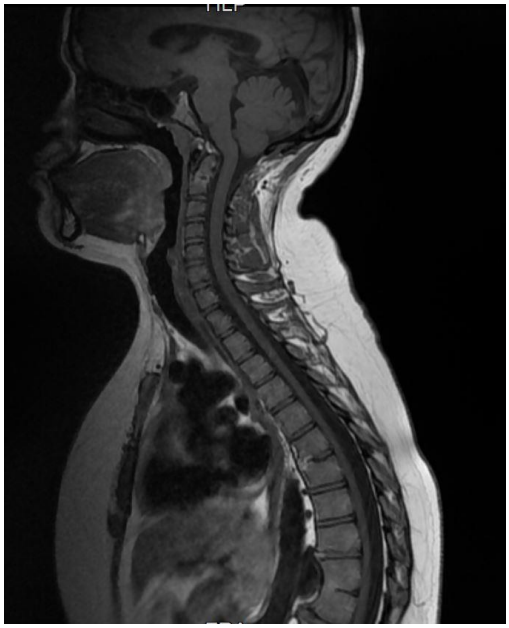

Figure 5 & 6: Spinal cord MRI: T2 sagittal slices: exaggeration of dorsolumbar curvatures with vertebral blocks D6-D7 and D10-D11

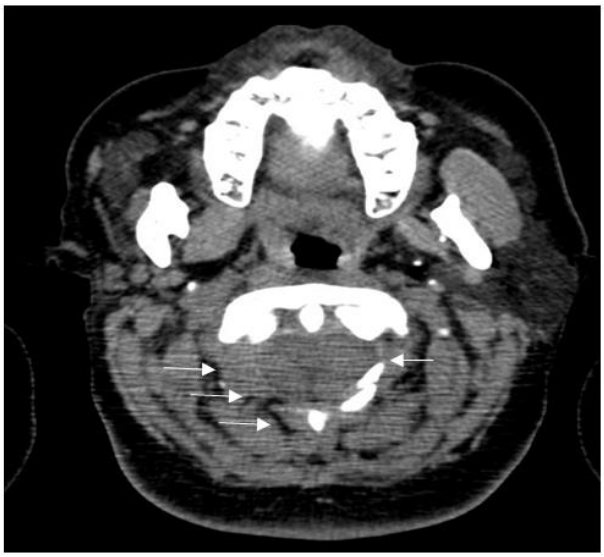

Figure 7: Cervical spine CT scan: axial section, C1 posterior arch closure defect (arrows)

DISCUSSION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is defined by congenital aplasia of the uterus and upper two-thirds of the vagina in women with normal development of secondary sex characteristics and a normal karyotype (46, XX) [1].

Two clinical forms have been described: MRKH type I, corresponding to isolated uterovaginal agenesis, and MRKH type II, characterized by incomplete agenesis and/or associated with other congenital malformations [2]. The main clinical sign is primary amenorrhea, and the karyotype is normal (46 XX). Ultrasound via the suprapubic route is a first-line method, and associated renal malformations should be systematically sought during this scan [6]. Nevertheless, MRI remains the most sensitive and specific examination compared with suprapubic ultrasound. Sagittal and axial T2 sequences allow accurate diagnosis, confirming uterine aplasia and the upper two-thirds of the vagina, and the normal appearance of both ovaries [3].

In addition, the search for other associated malformations (kidney and bone), our patient presented with associated vertebral anomalies. Psychological support is essential for patients with MRKH syndrome [4].

CONCLUSION

Mayer-Rokitansky-Küster-Hauser syndrome represents a clinical and emotional challenge for affected women. With its impact on fertility and sexual health, it requires multidisciplinary management, including specialists in gynecology, psychology and reconstructive surgery.

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