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Infertility Work-up Reveals Adult Female Hypospadias Diagnosed via Pelvic MRI: A Rare Case Report

M. Boussif[1*], A. Outrah[1], S. Ouassil[1], H.C. Ahmanna[1], B. Zouita[1], D. Basraoui[1], H. Jalal[1]

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

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***Corresponding author:** M. Boussif
Department of Radiology, Mother and Child Hospital, University Hospital of Mohamed VI, Marrakech, Morocco

Abstract **Case Report**

Female hypospadias (FH) is an extremely rare congenital anomaly characterized by an abnormally located urethral meatus on the anterior vaginal wall. Unlike male hypospadias, FH is seldom observed and is often overlooked, typically coexisting with other urogenital anomalies. The primary treatment is surgical urethral reconstruction. We present the case of a 26-year-old woman with a history of congenital adrenal hyperplasia and female pseudohermaphroditism. During an infertility work-up, a pelvic MRI revealed a urethro-vaginal communication concurrent with FH.

Keywords: Adult Female Hypospadias, Pelvic MRI, Infertility, Congenital Anomaly, Urethro-Vaginal Communication.

INTRODUCTION

Female Hypospadias (FH) is a congenital anomaly characterized by partial or complete agenesis of the urethra-vaginal septum, which results from incomplete development of the urogenital sinus. This defect causes the urethral meatus to open on the anterior vaginal wall, anywhere from the introitus to the vaginal fornix [1]. There is considerable confusion regarding this condition, specifically when trying to distinguish it from urogenital sinus abnormalities, as some authors consider it a form of urogenital sinus while others see it as a different anomaly altogether [2]. It is usually associated with other conditions related to urogenital septal defects, such as vaginal atresia or other anomalies like 46XX disorders of sexual development [3]. Clinically, the presentation of FH varies widely; it may be asymptomatic and discovered incidentally during procedures like catheterization, or it can manifest through symptoms such as vaginal voiding, recurrent urinary tract infections (UTI), urethral syndrome and dyspareunia, etc. [4]. Diagnosis is mainly radiological, establishing a communication between the urethra and the vaginal wall. This can be done by antegrade cystography via a suprapubic catheter or MRI with endovaginal opacification [1]. Treatment is exclusively surgical and is based on transposition of the external opening of the urethra from the vagina to the perineum in its subclitoral location [5].

CASE PRESENTATION

A 26-year-old nulligravid female (46XX karyotype) with a history of congenital adrenal hyperplasia (CAH) and female pseudohermaphroditism presented for evaluation of female infertility. Physical examination revealed an enlarged clitoris and an imperforate hymen. As part of an infertility evaluation, hormone levels were assessed. Follicle-stimulating hormone (FSH) was 6.6 IU/L, luteinizing hormone (LH) was 9.2 IU/L, and estradiol was 59.3 pg/ml, all within normal ranges. Testosterone was elevated at 1.6 ng/ml. A 40-day progesterone withdrawal test yielded a negative result. Pelvic MRI was ordered which revealed a uterus reduced in size measuring 26 mm with a ratio cervix/uterus at 0.8, and an enlarged clitoris, with no other notable anomalies specifically in the ovaries [Figure 1], however after vaginal opacification, we noted intravesical passage of vaginal gel with subsequent opacification of the bladder [Figure 2] confirming the existence of a urethro-vaginal communication in favor of FH.

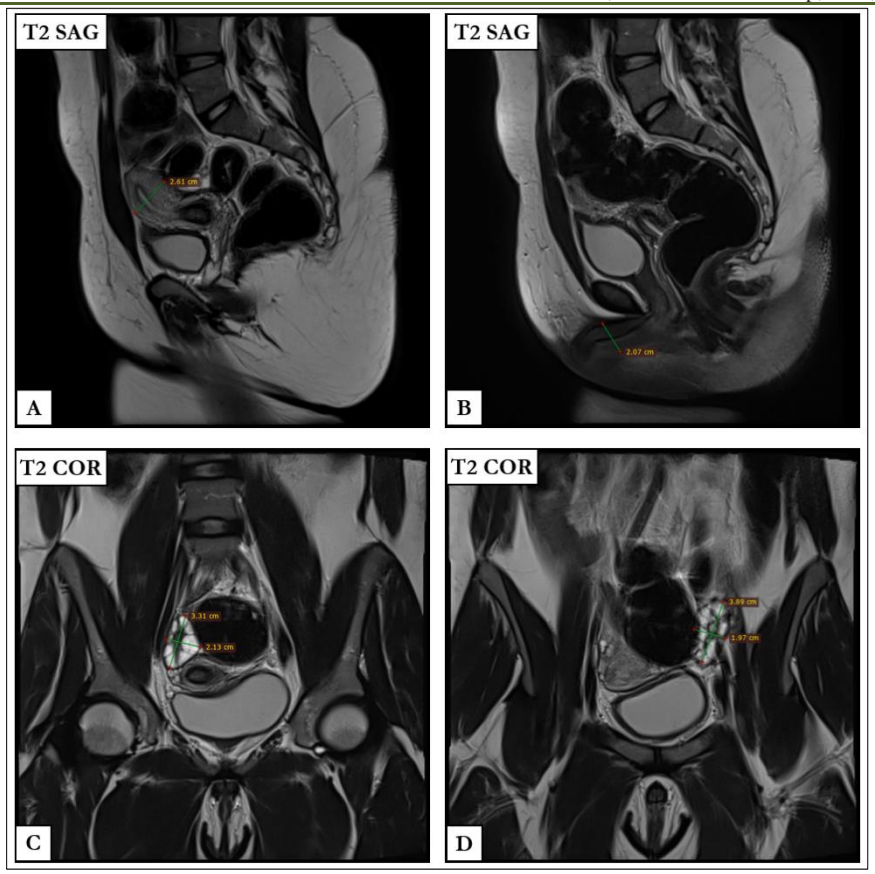

Figure 1: Pelvic MRI on T2-weighted sequences on sagittal (A,B) and coronal views (C,D) showing a uterus reduced in size measuring 26 mm (A) with an enlarged clitoris (B) and normal ovaries (C: right ovary, D: left ovary)

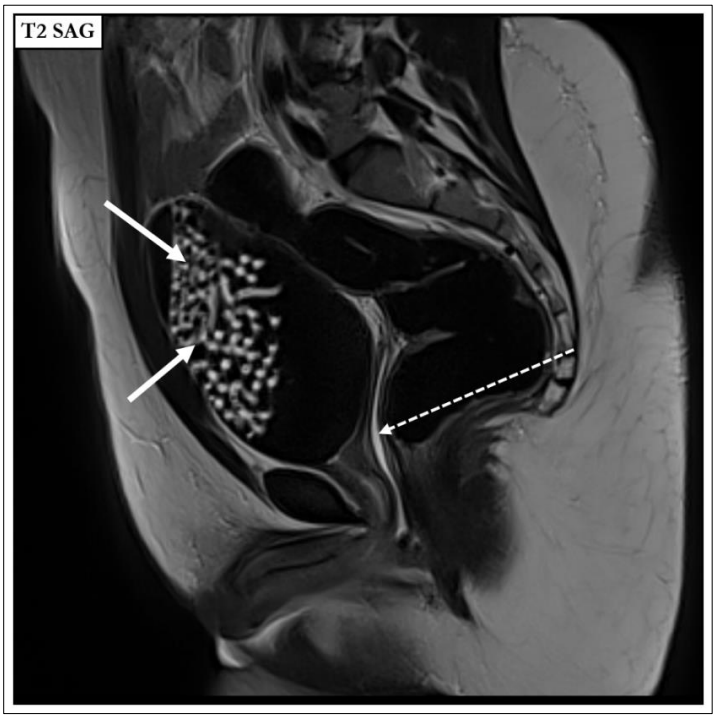

Figure 2: Pelvic MRI on T2-weighted sequences on sagittal view showing opacified vaginal lumen (dotted arrow) with subsequent passage of vaginal gel in intravesical (solid arrows) demonstrating a urethra-vaginal communication

DISCUSSION

Female Hypospadias (FH) is an extremely rare congenital condition defined as female urethral opening on the anterior wall (proximal to the hymenal ring) anywhere between the introitus and the vaginal fornix [1]. The nomenclature is often described as a variant of urogenital sinus (Merguerian and McLorie, 1992), whilst others see it as a different condition altogether (Knight *et al*., 1995), although they share the same pathological process which is a lack of development of urogenital sinus [2]. From an anatomical standpoint, there are multiple classifications, however, the most cited one is Solov'ev classification which separates this entity into : (a) vestibular (partial), (b) vesibulovaginal (subtotal) and (c) vaginal (total) types [3, 4]. Clinically, the condition can be asymptomatic or cause symptoms such as postmicturition incontinence and imperfect control, recurrent urinary tract infections, and urethral syndrome (referring to isolated urethritis with symptoms of increased frequency, pain during micturition, urgency, and dyspareunia) once sexual intercourse has commenced [2]. FH is a rarely isolated condition and is seen in association with 46XX disorders of sexual development (DSD), and is commonly associated with other genitourinary such as non-neurogenic neurogenic bladder, ectopic ureter, urethral duplication and renal anomalies, whilst genital anomalies like septate vagina and bifid uterus can also occur [2][1]. Diagnosis is often suspected on the occasion of a failed attempt of catheterization usually in the case of a radiological evaluation of the urinary tract when the examiner fails to locate the urethral meatus [2], and is confirmed by demonstrating a communication between the vaginal introitus and the bladder often by either anterograde cystography via a suprapubic catheter or a pelvic MRI [1-3]. Other radiological means, such as ultrasound and nuclear scintigraphy serve to evaluate the severity and impact this condition on renal function [3]. In our case, female infertility caused by FH is a possibility, however it remains a remote one, as this particular presentation has only been reported once in literature and is founded [6], according to its author, on the occurrence of pregnancy after surgical treatment of FH, but fails to provide a clear cause-and-effect relationship between the two entities. Treatment is surgical and is done by transposition of the external opening of the urethra from the vagina to the perineum in sub clitoris location [7, 8]. Various methods have been described such urethroplasty using either a vaginal or bladder flap [4, 5].

CONCLUSION

Female hypospadias (FH) is a rare condition that is often diagnosed late, typically after complications arise. It should be considered in the differential diagnosis of recurrent urinary tract infections, particularly when the urethral meatus cannot be located. Additional research is required to determine whether FH contributes to female infertility. Treatment is surgical, and the outcomes are generally favorable.

Conflict of Interest: The authors declare no conflicts of interest.

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