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Morris Syndrome at the Marie Curie Medical Clinic in Bamako: A Case Report

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Abstract **Case Report**

***Introduction*:** MORRIS syndrome is a rare hereditary disease causing complete peripheral androgen insensitivity. The aim of this study was to demonstrate the value of imaging in this syndrome through a case in our department. ***Case Report*:** This was a 22-year-old female patient with no known medical or surgical history, apart from primary amenorrhea in her 18-year-old younger sister. She was admitted to the Marie Curie Medical Clinic for primary amenorrhea and a desire to conceive. Clinically, the patient had an unambiguous female morphotype of the external genitalia, with well-developed Tanner stage 4 breasts. There was an absence of axillary and pubic hair. There was evidence of uneventful sexual intercourse, confirming the existence of a normal vagina. Suprapubic and endovaginal ultrasound, as well as magnetic resonance imaging, revealed a vaginal canal without visualization of the uterus or ovaries. Molecular genetic testing revealed an XX phenotype. ***Conclusion*:** Insensitivity syndrome is a very rare condition, and cross-sectional imaging is central to diagnostic management.

Keywords: MORRIS Syndrome, Ultrasound, MRI, Marie Curie Clinic.

INTRODUCTION

MORRIS syndrome is a rare hereditary disease resulting in complete peripheral androgen insensitivity. It represents 1 case in 20,000 to 1 case in 100,000 births, responsible for the total absence of male sexual differentiation during embryogenesis [1]. This pathology is one of the disorders of sexual development with a potential risk of degeneration linked to the Y chromosome, hence the interest in early diagnosis and management [2]. The objective of this work was to bring the interest of imaging in the diagnosis of Morris syndrome through a case in our department.

OBSERVATION

This was a 22-year-old patient with no known medical or surgical history, apart from primary amenorrhea in her 18-year-old younger sister. She is single, the 5th child in the family (8 children including 2 boys and 6 girls). She was admitted to the Marie Curie medical clinic in the radiology and medical imaging department in Bamako, Mali, for primary amenorrhea and desire for a child. Clinically, the patient had an unambiguous female morphotype of the external genitalia, well-developed breasts, Tanner stage 4 **(Figure 1).** There was an absence of axillary and pubic hair. There was a notion of sexual intercourse without problems, which confirms the existence of a normal vagina. Biologically, the testosterone level was normal (3.9 ng/ml), the serum FHS normal and the LH was pathologically abnormally high (11.9 IU/l). The molecular test had found the XX genotype. Radiologically, the suprapubic and endovaginal ultrasound as well as the Magnetic Resonance Imaging had highlighted: a vaginal canal without visualization of the uterus or the ovaries **(Figure 2).** The presence of oval formations in contact with the external iliac axes in T2 hyper signal whose signal resembled the testicles and the absence of uterine structure in MRI **(Figure 3).**

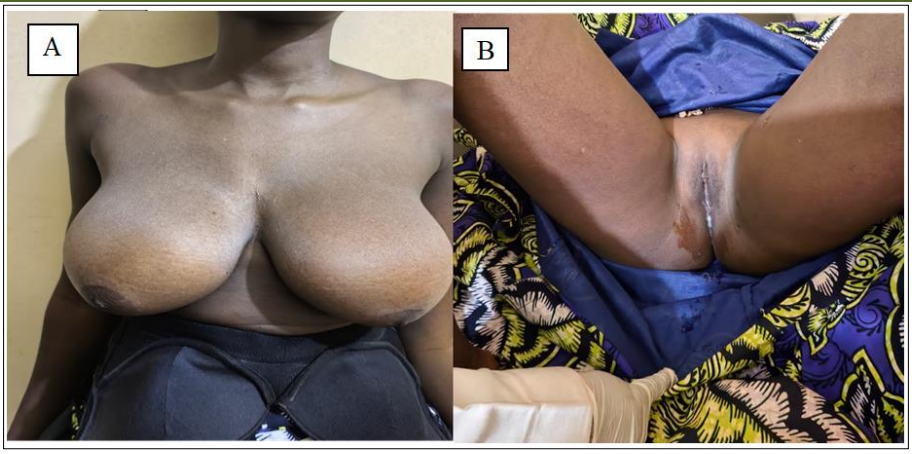

Figure 1 (A and B): Photo with visualization of well-developed breasts stage 4 of tanner (A) and an absence of pubic hair (B)

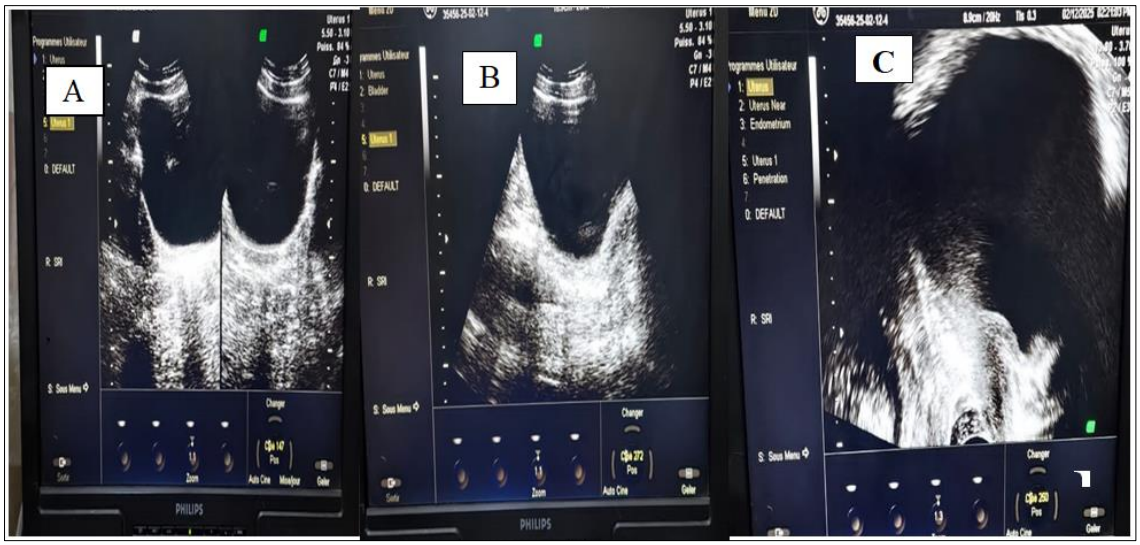

Figure 2: Endo-vaginal and supra-pubic pelvic ultrasound (A, B and C) showing a well-filled bladder without visualization of the uterus or ovaries

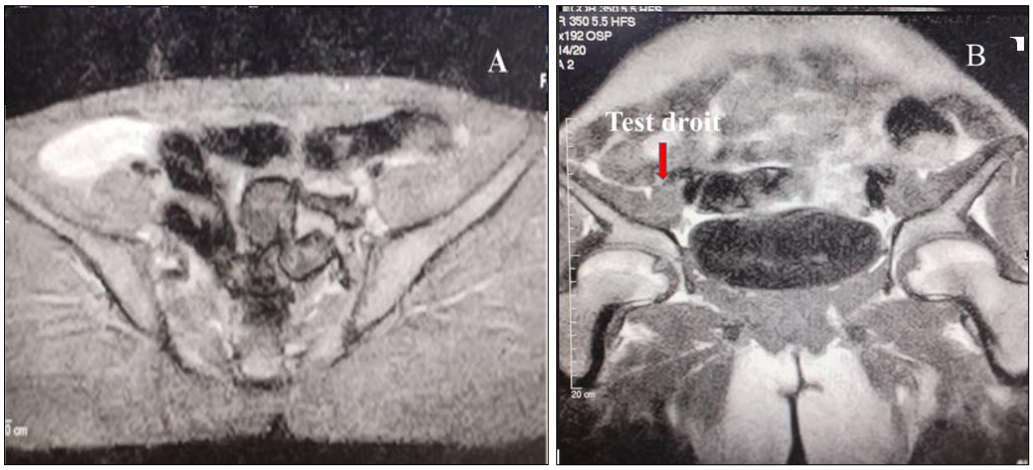

Figure 3 (A and B): MRI in axial T1 and coronal T1 sequence; showing the absence of the uterus or ovaries in a 22-year-old girl with the presence of a right testicle

DISCUSSION

The diagnosis of total androgen insensitivity is based on clinical and biological findings. Other types of androgen insensitivity are based on clinical findings and the notion of family history [3]. Our case was noted clinically as part of the infertility assessment (primary amenorrhea). The female morphotype, associated with primary amenorrhea and the familial character in our case allow us to retain MORRIS syndrome which is

confirmed by pelvic MRI and subsequent genetic and hormonal assessments (karyotype and testosterone, FSH and LH). In partial or mild forms of androgen insensitivity, carrier women often have delayed puberty and/or less dense pubic or axillary hair. There is also a certain degree of bone demineralization [2, 3]. Our case was the partial form. Biologically: Testosterone levels are normal or increased. LH is high. FSH is at a normal or slightly elevated level and the estradiol concentration is also increased. During pharmacological stimulation by HCG, the testosterone response is normal [1-4]. Late diagnosis would be linked to negligence or ignorance. Ethical considerations were crucial in this diagnostic approach and will remain as such in the management. Radiologically: pelvic ultrasound and MRI (magnetic resonance imaging) are the imaging size to confirm the absence of the uterus and ovaries but will visualize the ectopic testes in the iliac or lumbar fossae [1-5]. In our case the testicle was found in the right iliac fossa. An improvement in the quality of life is obtained, however infertility remains the major challenge faced by the affected individuals.

CONCLUSIONS

Insensitivity syndrome is a very rare condition that can be diagnosed pre- or post-pubertally, with variable management depending on the early diagnosis. Ultrasound and MRI were invaluable in our diagnostic approach and confirmed the absence of internal genitalia. Cross-sectional imaging therefore remains central to the diagnostic management of this condition.

Ethical Consideration: Informed consent was obtained and anonymity was maintained.

Conflict of Interest: None.

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