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The Imaging of True Hermaphrodisim at the Medical Clinic "Marie Curie" in Bamako: About a Case

Traore Ousmane[1,2*], Kouma Alassane[2], Diarra Ouncoumba[2], Traore Mamadou Sylla[1], Diarra Phylip[1], Bagayoko Ousmane Lansenou[1], Diallo Mahamadou[2], Keita Adama Diaman[2]

[1]Department of Radiology and Medical Imaging, The Medical Clinic Marie Curie, Bamako, Mali
[2]Faculty of Medicine and Odontostomatology (FMOS), University of Sciences, Techniques and Technology of Bamako (USTTB), Bamako, Mali
Email: *ghousno1@yahoo.fr

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Abstract

Introduction: True hermaphroditism, also known as mixed gonadism, is a rare genetic anomaly manifested by the presence of both male and female gonads, giving the individual a mixed phenotype and thus sexual ambiguity. The objective was to demonstrate the value of imaging in this pathology through an observation in our department. **Observation:** This was an 18-year-old subject with no known history, claiming to be male. He was admitted to the Marie Curie medical clinic for sexual ambiguity. Clinically, there was gynecomastia, a female-type pelvis with a rudimentary vulva, and female-type suprapubic hair. Primary amenorrhea, recurrent pelvic pain, ejaculation, and morning erections were noted. A micropenis without a urethral meatus in place of the clitoris was present. There was no vulvar vestibule or labia minora. Biologically, serum testosterone was normal (3.6 ng/ml), serum FSH was normal (11UI/L), and LH was abnormal (11.4 IU/l). Pelvic ultrasound and magnetic resonance imaging revealed a right ovary, a uterus, a left testicle, and structures similar to the corpus spongiosum and cavernosum of the penis. Molecular testing revealed the XX genotype. **Conclusion:** Cross-sectional imaging remains essential in the management of true hermaphroditism.

Keywords

Hermaphroditism, Ultrasound, MRI, Marie Curie Clinic

1. Introduction

True hermaphroditism, also called mixed gonadism, is a genetic anomaly during

embryogenesis manifested by the presence of male and female gonads, which are biologically functional, giving the individual a mixed phenotype and therefore sexual ambiguity. The normal sexual development of the fetus depends on several chromosomal, gonadal and hormonal factors [1] [2]. True hermaphroditism is a rare anomaly (1/100,000) that can severely affect the social life of affected individuals. Individuals may present a variety of physical and hormonal characteristics. Diagnosis and management are based on a multidisciplinary approach, while taking into account ethical and psychosocial aspects. Medical imaging remains at the center of this management [3]. The aim of our work was to bring the interest of imaging in true hermaphroditism through a clinical case observed in our department.

2. Observation

This was an 18-year-old subject with no known history, claiming to be male. He was received at the Marie Curie medical clinic in the radiology and medical imaging department in Bamako, Mali, for sexual ambiguity. Clinically, the subject was seen in a multidisciplinary consultation consisting of a general practitioner first and secondarily specialists such as the obstetrician-gynecologist, the urologist, the plastic surgeon and the pediatric surgeon. There was gynecomastia, a female-type pelvis with a rudimentary vulva, and female-type suprapubic hair. Primary amenorrhea was reported, repeated pelvic pain, ejaculation and morning erections were present. A micropenis without a urethral meatus in place of the clitoris was seen by the urologist and the obstetrician-gynecologist (**Figure 1**). There was no vulvar vestibule or labia minora. Biologically, testosterone levels were normal (3.6 ng/ml), serum FSH was normal (11 UI/L), and LH was pathological (11.4 IU/l). The reference values are for FSH (Man: 1.22 - 19.25 IU/L, Woman in ovulation: 6.3 - 24 IU/L, in follicular phase 2.9 - 12 IU/L) and for LH (Man: 1.7 - 7 IU/L, Woman in follicular phase: 1.5 - 8 IU/L and in ovulation: 9.6 - 80 IU/L). Molecular testing

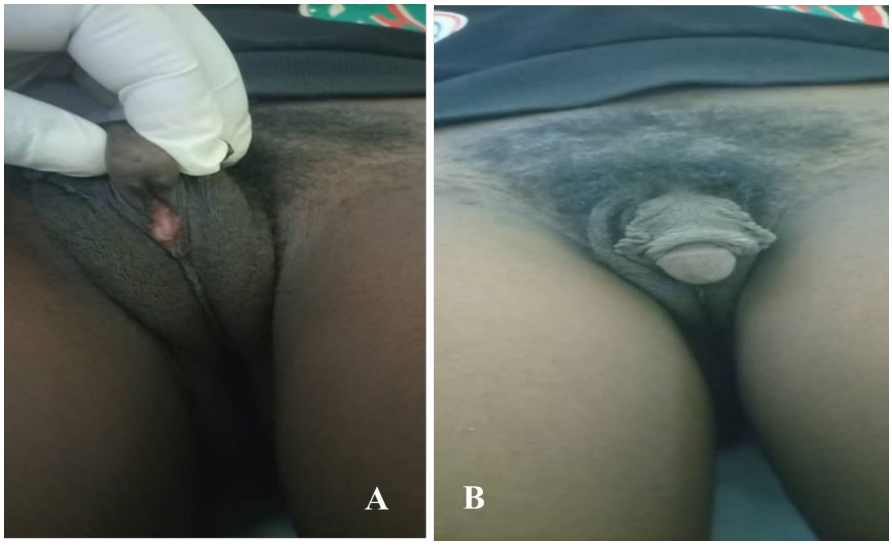

Figure 1. Photo with visualization of micro penis and suprapubic hair (A) and the rudimentary vulva (B).

revealed the XX genotype. Radiologically: abdominopelvic ultrasound and Magnetic Resonance Imaging (MRI) revealed both kidneys in place without abnormalities, a right ovary and uterus present, a left testicle, and structures similar to the corpus spongiosum and cavernosum of the penis were also present (Figure 2). The prostate was visible on MRI (Figure 3).

The treatment option in our case was surgery proposed by the pediatric surgeon

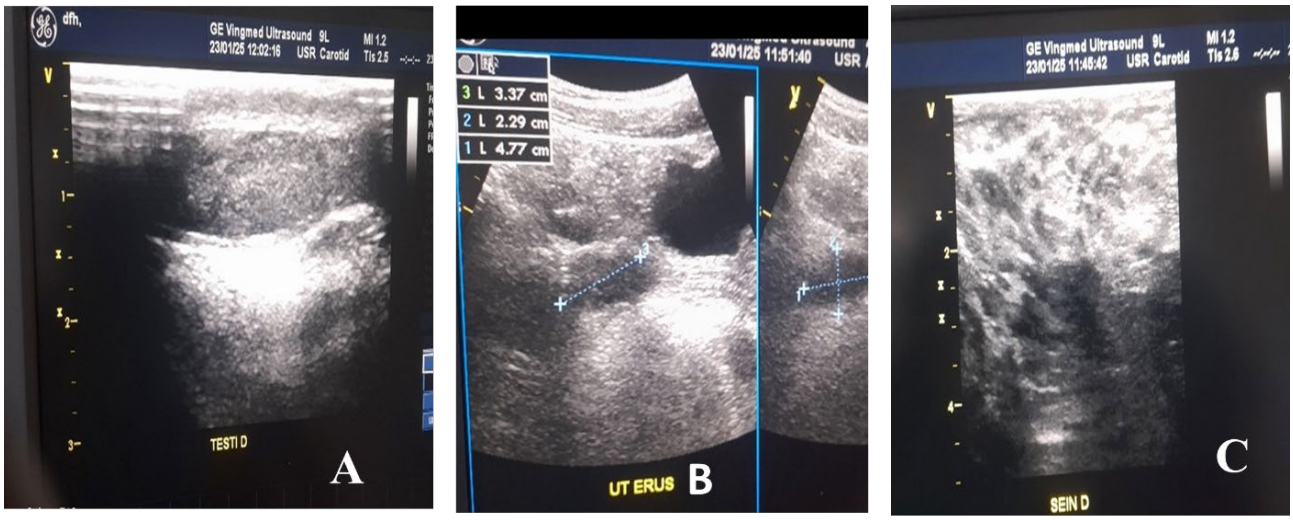

Figure 2. Abdominopelvic ultrasound showing the single testicle (A) and the small uterus (B) and a grammatical ultrasound (C) showing mammary glands without nodules.

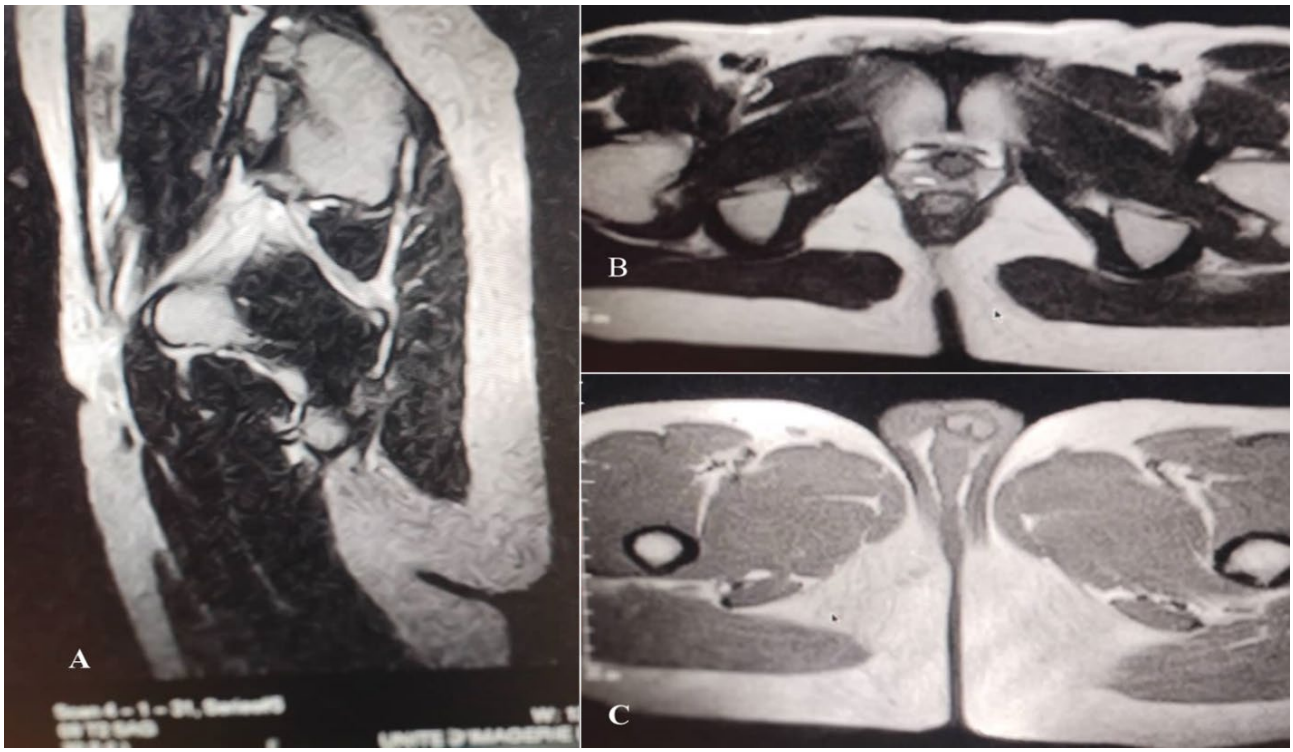

Figure 3. MRI in sagittal T2 sequence; axial and axial T1 showing the presence of the uterus in (A), the prostate in (B) and the presence of a right testicle (C).

and the gynecologist. Our subject had accepted the surgery but preferred to be male and the subject's father had refused for socio-cultural reasons according to the psychologist's report. Our case was lost to follow-up one month later.

3. Discussion

The simultaneous presence of functional male and female genital organs (ovaries and testes) as well as mixed secondary sexual characteristics confirms true hermaphroditism, the management of which remains complex and multidisciplinary [1] [3] [4]. In our African context, access to psychologists, geneticists, plastic surgeons, urologists, gynecologists, endocrinologists and biologists remains a challenge given the geographical, economic and religious context which can raise complex questions. However, strict compliance with ethics and active listening for better social and medical-surgical care are fundamental measures observed in this case [1] [4]. True hermaphroditism constitutes a major diagnostic challenge because it affects genetics, developmental biology and surgery, of which imaging remains at the heart of this management. Ethical considerations play a crucial role in this management [1] [5]. Allen's classification is described in the literature [1] [6]. It is based on the histological findings of the gonads and includes six groups: female pseudohermaphroditism 46XX, male pseudohermaphroditism 46XY, true hermaphroditism 46XX, 46XY or mosaic, mixed gonad dysgenesis 46XO/46XX, pure gonad dysgenesis 45X0 (Turner syndrome), 46XX, 46XY and other forms with dysgenetic testes, teratogenic factors [6]. Our observation was classified as a true hermaphroditism type. This Allen classification is old and has been replaced by the Chicago Consensus (2006-2016). The term true hermaphroditism was replaced by the concept of disorders of sex development (DSD) or intersexuality in the Chicago Consensus of 2006 and 2016, which used modern terminology to describe variations in sexual characteristics. The current recommendations (2016) emphasize the awkwardness of using the term "true hermaphroditism" due to its outdated and inaccurate connotation, preferring the term intersex or Variation of Sex Development (VSD). The use of the term "hermaphroditism" is considered derogatory and historically misleading. Modern terminology for SDT is more accurate and respectful, as it recognizes the diversity of conditions and emphasizes variations in sexual development rather than the idea of an individual having both male and female reproductive organs, which is a misleading simplification [7]. Advances in imaging methods, hormonal dosages, molecular biology and urosurgery techniques have contributed considerably to the improvement of diagnosis which has become more precise and faster, as well as to the management of the child with sexual ambiguity [1]. Imaging methods are based on examinations such as abdominopelvic ultrasound and MRI. This MRI is rarely cited in the literature, only once to our knowledge which should give more details of the reproductive system and evaluate the anatomy of the external and internal genital organs [1]. The prostate, the right testicle and the uterus were found in MRI in our subject. The abdominopelvic ultrasound allowed the identification of the location and

morphology of the gonads by showing ambiguous genital organs or mixed structures, both male and female which is fundamental for the diagnosis of true hermaphroditism. The ultrasound had found a single testicle and a uterus in our observation. The diagnosis of true hermaphroditism or disorder of sexual development was made based on the clinical-biological aspect and imaging. The therapeutic decision was surgical and hormonal but the subject was lost to follow-up. Patients with disorders of sexual development are due to congenital adrenal hyperplasia, caused by enzymatic deficiencies involved in the hormone synthesis pathway. The other cases are explained by genetic anomalies involved in the development of the genital system [8]. Our observation did not have adrenal dysplasia but abnormal development of the genital system.

4. Conclusion

True hermaphroditism is a very rare and complex pathology requiring precise assessment and careful management to improve the quality of life of these individuals in society. Its discovery will be responsible for a huge and serious health problem that could hinder the social development of an average individual. Cross-sectional imaging remains essential in the management of this pathology.

Ethical Consideration

Informed consent was obtained and anonymity was maintained.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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