

The Role of Ultrasound in the Diagnosis of Sexual Ambiguity: A Case Report from the Gabriel Touré University Hospital (CHU)

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ABSTRACT

Sexual ambiguities or anomalies of sexual differentiation or Disorders of sex development sexual (DSD) are defined as congenital conditions in which the development of chromosomal, gonadal or anatomical sex is atypical [1,2].

There are several types of ambiguity, listed in order of frequency: female pseudohermaphroditism, male pseudohermaphroditism, and finally, true pseudohermaphroditism, which is the least frequent of the three, involving the coexistence of ovarian and testicular tissue [4]. Diagnosis is generally made at birth, based on abnormal external genitalia, but can be much later, at puberty. This is where imaging studies become important [4]. Abdominopelvic ultrasound is an essential diagnostic tool.

Hence the interest in reporting this clinical case of sexual ambiguity in the imaging department of the CHU GT.

Keywords

Disorders of sex development (DSD), Sexual ambiguity, Ambiguous genitalia, True hermaphroditism / Ovotesticular DSD, Gonadal dysgenesis, Abdominopelvic ultrasound.

Introduction

Sexual ambiguities or anomalies of sexual differentiation or Disorders of Sex development sexual (DSD) are defined as congenital conditions in which the development of chromosomal, gonadal or anatomical sex is atypical [1,2].

There are several types of ambiguity which are in order of frequency: female pseudohermaphroditism, male pseudohermaphroditism and finally true pseudohermaphroditism which is the least frequent of the three, involving the coexistence of ovarian and testicular tissues [4].

In Mali, the frequency of sexual ambiguities is estimated at 0.54% of reasons for consultation in internal medicine [3].

The diagnosis is usually made at birth, based on abnormal external genitalia, but can be much later, at puberty. At this time, ambiguity is suspected in cases of delayed puberty or the appearance of secondary sexual characteristics that are inconsistent with the assigned sex. Therefore, it is necessary to diagnose ambiguous genitalia in the neonatal period. It is also an emergency for two reasons: it may be a case of adrenal hyperplasia causing salt-wasting syndrome, which is a metabolic emergency requiring immediate management; once this emergency has been ruled out, the diagnostic workup must be rapid in order to assign a sex to the child. The objective of our study is to clarify the contribution of ultrasound in the diagnosis of ambiguous genitalia.

Methodology

This was a descriptive study of a clinical case of sexual ambiguity at the medical imaging department of Gabriel Touré Hospital in Bamako from October 2024 to October 2025. The study focused on a woman referred to the imaging department for a confrontational abdominal-pelvic ultrasound.

Sociodemographic variables, clinical and paraclinical data were used. A SIEMENS HEALTHINERS ultrasound machine equipped with four multifrequency probes with pulsed-wave color Doppler option was used: a 10MC3 endovaginal probe; a 16-5 MHz linear probe; an 11L4 superficial probe; and a 5-2 MHz deep convex probe.

Observation

This was Miss AK, aged 20, of Bozo ethnicity, declared female at birth with no family history, as no member of the family has this condition.

She is the third child born into a monogamous family of five, as she has two older brothers and two younger sisters. Due to the uncertainty of obtaining a reliable diagnosis after numerous consultations at healthcare facilities, the patient was referred to us for a confirmation ultrasound.

The onset of the condition dates back 5 years, marked by the absence of development of secondary sexual characteristics and of the menstrual cycle, motivating the parents to make several consultations without favorable outcome until the date of 13/05/2025 when we received the patient for an ultrasound examination at the university hospital center (CHU) Gabriel Touré.

The subject's general condition was good, with feminine clothing, lipstick, pearls on her wrist, and neatly braided hair. She presented with ambiguous genitalia. Her body type was feminine, and her breasts were less developed elsewhere. Sexually, she had labia majora with a small vagina, no scrotum, but visible traces of pubic shaving. The rest of the examination was unremarkable.



Figure 1: Image of the patient

IMAGING

The various transverse and sagittal sections of the abdomen and pelvis obtained with the 5.2 MHz MG deep convex probe revealed: The pelvic ultrasound: found in retrovesical, an echogenic, homogeneous structure with clear contours measuring

30x30x21mm, i.e. an estimated volume of 10 grams in relation to the prostate without seminal vesicles associated with two normal-sized testicles located at the bilateral inguinal level all vascularized on color doppler whose ultrasound appearance suggested an anomaly of sexual differentiation (ASD): a male pseudohermaphroditism.

The absence of the uterus and its adnexa was noted.

Biology

A hormonal assessment consisting of:

- Free Thyroxine (FT4): 11.70 pg/ml N (8.9 – 17.20)
- Ultrasensitive TSH: 2.49 μ UI/ml N (0.30 to 4.50)
- Prolactin: 172.00 μ UI/ml N (66.0 – 490.0)
- FSH: 14.30 mIU/ml N (3.2-15.0) Follicular phase
- LH: 25.30 mIU/ml N (12.0 – 82.0) Ovulatory peak
- Androstenedione – Delta 4 (RIA): 5.50 ng/m N 0.40 – 2.80)
- Estradiol: 105.11 pg/ml N (15-112) Follicular phase
- Total testosterone: 2.730 ng/ml N (less than 1.00)

and molecular sex determination tests were requested, which proved favorable with an XY sex. This result supports a male sex. This test is highly specific in the genetic profiling of the ATL-SRY genes. Given this diagnostic confirmation of sexual ambiguity and in a symptomatic context, surgery was proposed followed by hormone therapy.



Figure2: Ultrasound image showing the prostate in the anterior-post-medial position.

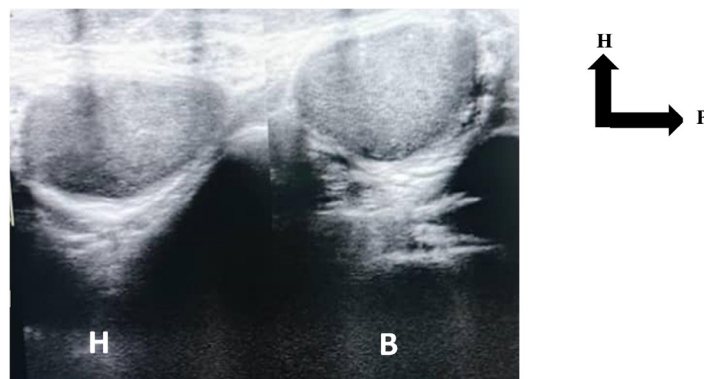


Figure 3: Ultrasound image of the right (A) and left (B) testicles in the inguinal region.

The postoperative course was straightforward and the patient was discharged on postoperative day 3.



Figure 4: Image of the extracted testicular tissue (Bilateral Orchiectomy).

Discussion

The international consensus on DSDs proposes classifying the different anomalies according to the karyotype. We received the patient on May 13, 2025, for a case of a 46XY DSD chromosomal abnormality. Cases of DSDs have been reported elsewhere in Africa. In Mali, a study collected data on 12 patients over a seven-year period [3], in Senegal over a ten-year period, 24 DSDs were identified [5] and similarly in Congo over five years, four cases were identified [6]. These isolated cases of DSDs in Africa did not reflect the reality, as Diakité pointed out in his work [7]. He reported that patients faced a problem of referral, which made it difficult to obtain a national registry on DSDs in African countries. The discovery was late, which is comparable to the study conducted by Cissé D et al. [8], in which the patient was 21 years old. According to several authors, ultrasound examinations were requested in response to pubertal abnormalities such as virilization in a girl, absence of breast development, or primary amenorrhea, with the aim of establishing a diagnosis [9-11]. These signs of abnormality were found in our case. In this case study, imaging is the first-line paraclinical examination, comparable to that of Cissé D et al. [8], demonstrating the presence of the prostate and testes in the inguinal region. In our study, the patient was able to undergo karyotyping, which was 46,XY. In the study by Mamadou Diallo [4], one out of five patients [12] underwent karyotyping (46, XX). In the study by Hué et al. [13], seven out of 23 patients registered during the study period underwent karyotyping. In 2 cases, ADS 46XY was noted, in 3 cases, ADS 46 XX, and in 2 cases, ADS due to chromosomal abnormality.

The treatment was multidisciplinary, as is the case in many instances of sexual ambiguity. Our patient did not undergo vaginoplasty because the depth of the vagina was acceptable and was expected to improve under hormone therapy and intercourse.

Conclusion

Intersex conditions, although relatively rare, exist in Mali and throughout the world. In Mali, due to limited access to diagnostic equipment, diagnosis relies on patient history, physical examination, abdominal and pelvic ultrasound, and blood tests for sex hormones. Early diagnosis is crucial to ensure normal growth, puberty, and satisfactory fertility.

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