

# RADIOLOGICAL EVALUATION OF AMBIGUOUS GENITALIA: A CASE REPORT AND REVIEW OF LITERATURE

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**Abstract-** Ambiguous genitalia represent a group of congenital disorders where the external genitalia of a child appear atypical. In our environment, the huge joy and celebrations that normally accompany the arrival of such a baby are replaced with great apprehension and confusion in the family. Radiological evaluation of the gonads and internal genital organs constitutes an important component of multidisciplinary team approach required in the work-up and treatment of this condition. We report a case of ambiguous genitalia in a female with clitorimegaly, unicornuate uterus and single patent fallopian tube. It demonstrates the central role of imaging in the diagnosis and care of such patient.

**Keywords:** Ambiguous genitalia, Clitorimegaly, Unicornuate uterus, Flush genitogram.

## I. INTRODUCTION

When the external genitalia of a child does not have the typical anatomic appearance of a normal male or female, he or she is said to have ambiguous genitalia (AG). In our region, the joy and celebrations that normally accompany the arrival of such a baby are replaced with great apprehension and confusion not only by the family but also the care-givers.

AG is a result of disorder of sexual developments (DSDs) in which the development of chromosomal, gonadal, or anatomical sex is atypical [1]. Fausto-Sterling's estimate of 1.7% was initially documented as prevalence of ambiguous genitalia when other conditions such as Klinefelter's syndrome, Turner's syndrome, and late onset adrenal hyperplasia were also considered [2]. If a disorder of sex developments (DSDs) is defined exclusively as a condition in which chromosomal sex is inconsistent with phenotypic sex, or in which the phenotype is not classifiable as either male or female; The estimated prevalence is 0.018% (i.e. 1 in 5555 patients) [3]. In Enugu state, Nigeria, a prevalence rate of 6.8% of congenital aberrations of external genitalia was noted among secondary school students not diagnosed at birth [4].

The ability to diagnose this condition has improved rapidly in recent years due to the advances in molecular genetics. Also, the hitherto paradigm has shifted away from early reconstructive procedures in some cases.

Radiological evaluation of the gonads and internal genital organs constitutes an important component of multidisciplinary team approach required in the work-up and treatment of patients with ambiguous genitalia.

In this report, we describe a case of AG in a female with clitorimegaly, unicornuate uterus and single patent fallopian tube. The findings at radiological evaluation are discussed

thus, demonstrating the central role of imaging in the diagnosis and care of patients with AG.

## II. CASE PRESENTATION

An 8year old referred from General hospital, Kafanchan to Ahmadu Bello University Teaching Hospital (ABUTH) Zaria with history of ambiguous genitalia since birth.

There was no enuresis or passage of urine per rectum. The mother did not ingest unprescribed drugs during the child's pregnancy and both parents were less than 30 years of age when the child was delivered. There was no history suggestive of other congenital anomaly. The child had earlier presented in the neonatal period at the referring facility where female gender was assigned.

Genital examination revealed clitoral hypertrophy resembling phallus (figure 1). The labia majora and minora were well developed and not fused. The urethra opening was noted at the normal anatomical position just below the hypertrophied clitoris. The perineum was dry; no jet of urine was noted from the urethral meatus. The vaginal opening was also noted postero-inferior to the urethra, and found normal for the patient's age. There was no palpable mass suggesting testicle in the inguinal regions or in the labia. No pubic, axillary or facial hair noted. The remaining systems were unremarkable.

A clinical diagnosis of ambiguous genitalia to rule-out congenital adrenal hyperplasia was made. The abdominopelvic ultrasound scan showed uterus with normal myometrial and endometrial echocomplex (figure 2). The adnexae were free and there was no definite mass lesion seen. The urinary bladder was uniformly filled with sonolucent urine. Its wall was not thickened and no wall defect or fistulous track to the uterus or rectum noted. There was no

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extra-luminal free fluid collection seen. The kidneys were normal in their anatomical positions, sizes and outline. The renal parenchymal echotexture, sino-parenchymal distinction and pelvi-calyceal systems were preserved bilaterally. There was no supra-renal (Adrenal) mass lesion seen. The remaining abdominal organs were normal. Flush genitogram showed normal cervix, unicornuate uterus and patent right fallopian tube (figs. 3a & b). The left fallopian tube was not visualized (congenital absence).



Figure 1. A clinical / pictorial image of the patient's perineum showing enlarged clitoris, mimicking phallus (arrow). The labia majora appears normal for patient's age. The tube artifact noted is a nasogastric tube passed for flush genitogram.

The Karyotype of the patient was not done as the facilities for this were not available at ABUTH and neighboring Teaching Hospitals. Hormonal investigations were within normal limits.

A radiological diagnosis of female gender with congenital absence of the left fallopian tube was made. The child was to have clitoroplasty following appropriate conference and consent from the parents. She however defaulted.

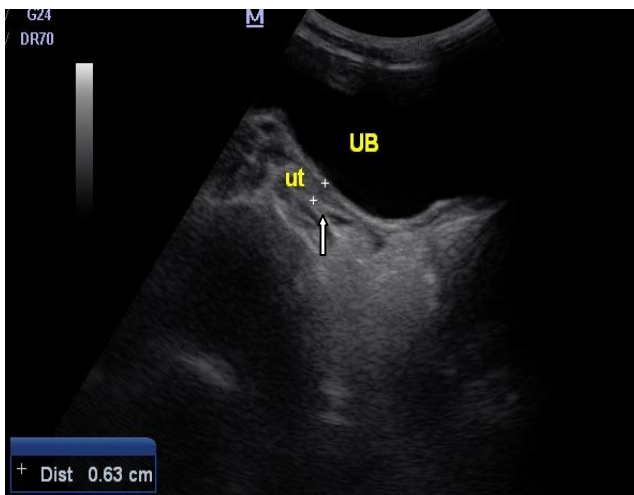


Fig.2. A longitudinal pelvic ultrasonogram of the same patient as in figure 1 showing an infantile uterus, measuring 0.63cm in maximum AP diameter (arrow).



Figure 3a is an AP view of a flush genitogram showing a contrast-filled uterus (point star) with a patent right fallopian tube (arrow). The cervix is preserved and the left fallopian tube is not visualized.



Figure 3b. A lateral view of a flush genitogram of the same patient as in fig 3a still demonstrating a single fallopian tube (arrow) and a contrast filled uterus (point star).

### III. DISCUSSION

The chromosomal basis for sex is determined at conception. The internal and external genital structures remain undifferentiated up to 6 weeks gestation. Three important precursor components of the genital system are the germ cells, the genital ridge, and the two sets of internal sex ducts,

namely, the Mullerian-Paramesonephric ducts and the Wolffian – Mesonephric ducts [5].

Disorders of sexual differentiation (DSD), formerly known as ambiguous genitalia are classified into 4 different types: Female Pseudohermaphrodites (46, XX DSD), Male Pseudohermaphrodites (46, XY DSD), True hermaphrodites (ovotesticular DSD) and Pure gonadal dysgenesis [6]. Congenital Adrenal hyperplasia is the commonest cause of genital ambiguity in 46 XX DSD [7]. It accounts for more than 70% of cases of AG in genetic females [8].

Overt genital ambiguity, poor breast development, inability to empty the urinary bladder, suprapubic mass due to haematometria and haematocolpus, monthly cramping or pain without menstruation, amenorrhoea in a supposed female gender, dyspareunia and repeated miscarriages presumably due to abnormal uterus are the usual presenting symptoms. Our patient passes urine freely and the remaining symptoms are yet to be appreciated as she was just 8 years and therefore, yet to attain menarche.

Clinical signs in some of the patients could include abnormal vagina, abnormal or missing cervix, bladder extrophy, ambiguous genitalia, labia that are stuck together or unusual in size, and swollen clitoris. Our patient has ambiguous genitalia with clitorimegaly. The remaining signs were not found in the child.

The Mullerian ducts (paramesonephric ducts) which are paired develop into the female genital tract in a cephalocaudal fashion. The more cephalad ends of the paired paramesonephric ducts are opened to the peritoneal cavity and develop into the fallopian tubes, while the more caudal portion fuses in the lower midline to form the uterovaginal primordium, which later develops into the epithelium and glands of the uterus and cervix [9]. If one Mullerian duct fails to develop, a unicornuate uterus results which consists of one uterine horn with one fallopian tube. Complete failure of the Mullerian system results in the absence of the fallopian tubes, uterus, cervix, and most of the vagina (Rokitansky – Kuster – Hauser Syndrome) [10].

In this patient, there was failure of development of the left Mullerian duct which resulted in a single uterine horn and absence of the left fallopian tube which was noted on radiological investigations.

A Chromosomal Karyotype should be done in all patients. This was not feasible in the index patient, as there were no facilities for Giemsa banding Karyotyping and spectral karyotyping in our centre and neighbouring hospitals. Imaging plays an important role in depicting the internal organs and urogenital anatomy in children with AG. Ultrasound is the primary modality for the evaluation of the internal reproductive organs, whereas genitography and micturating cytourethrography are used for evaluation of urethral and vaginal tracts for fistulas. Magnetic Resonance Imaging and Computed Tomography are problem solving modalities which clarify anatomy and helps in search of internal gonads.

In this patient, ultrasound which is non-invasive and non-ionizing was useful in appreciating the infantile uterus and the urinary bladder. Patient also had a benefit of flush

genitogram which clearly showed the uterine cavity with a delineation of patent right fallopian tube in its entire length. The kidneys and the remaining abdominal organs were evaluated radiologically and found normal. Evaluation of other organs was necessary as possible association of other congenital anomalies with ambiguous genitalia has been documented [11-12]. The adrenals were not visualized on ultrasonography as they are not enlarged. Enlargement of the adrenal glands is a common finding in patients with congenital adrenal hyperplasia, which is the commonest cause of AG [7].

The key issues in the care of patients with AG include; accurate diagnosis, gender assignment, indications and timing of major surgery and sharing of medical information with patients and parents. Emergency treatment may be necessary in some cases. For instance, adrenocortical crisis, hyponatremia or hypoglycemia in congenital adrenal hyperplasia should be looked for and managed accordingly. However, long-term treatment involving reconstructive procedures may be required. Our patient was scheduled to have feminizing genitoplasty involving clitoroplasty but she defaulted.

#### IV. CONCLUSION

Radiological evaluation of the gonads and internal genital organs constitutes an important component of multidisciplinary team approach required in the work-up and treatment of patients with ambiguous genitalia. In clinical practice, abdomino-pelvic ultrasonography is the preferred first-line imaging modality. Aside from defining the uterus, it also gives information on suprarenal gland and ectopic gonad. However, some cases may necessitate genitography, CT or MRI.

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