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Case Report

Ambiguous Genitalia in a 22-year-old Nigerian: A Case Report of Successful Perineoplasty and Clitoroplasty

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Abstract

Background: External genitalia are considered to be ambiguous whenever there is difficulty assigning gender to a child based on the appearance of the external genitalia. It is a common presentation of Disorders of Sexual Development (DSD). This is a case of a 22-year-old single undergraduate who had a disorder of sexual development with ambiguous genitalia resulting from congenital adrenal hyperplasia. Although the appearance of her genitalia in childhood could not be ascertained, she presented with an inability to achieve penetrative vagina intercourse, enlarged clitoris, poor urinary stream, and terminal dribbling. **Case summary:** She had clitoromegaly with fused labia minora at the midline which formed anterior to the urethral meatus with a normal capacious vagina behind the fused labia. She also had acne and hirsutism. Findings of investigations were suggestive of congenital adrenal hyperplasia with no salt wasting. She had successful perineoplasty and clitoroplasty and reported satisfactory appearance of her genitalia, she has been able to achieve penetrative vagina intercourse with satisfactory sexual function and urinary streams have improved with terminal dribbling resolved. She was counseled about the need for fertility support when ready. **Conclusion:** Disorders of sexual development though rare may have significant psychosocial effects on its victims and understanding the pathology with surgical intervention as indicated in this case may help improve the quality of life of the affected persons.

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INTRODUCTION

Disorders of sexual dysfunction (DSD) may be defined as the presence of both male and female external and/or internal genital organs in the same individual, confusing the diagnosis of true sex.[1] Simply put, it is described as ambiguity in genitalia. External genitalia are considered to be ambiguous whenever there is difficulty assigning gender to a child based on the appearance of the external genitalia.[2]

Ambiguous genitalia is a spectrum of disorders of sexual function with varying patterns of appearance of the external genitalia, which may range from being a feminized male to a masculinized female to different extents.[1] Better understanding has also significantly improved management. Ambiguous genitalia may be a part of a syndrome involving other organ systems.[1] Congenital Adrenal Hyperplasia (CAH) is the most common cause of intersex disorder.[3] It is otherwise known as androgenital syndrome. It is an autosomal recessive disorder due to an inborn error in adrenal steroid metabolism. There is a lack of cortisol production, resulting in excess production of Adrenocorticotropic Hormone (ACTH) from the

pituitary, which stimulates excessive adrenal production of androgens with virilization of female patients. Associated aldosterone deficiency may lead to excess salt depletion in some cases.[3,4]

Affected girls are potentially fertile, and they have normal internal genitalia [4] CAH has been classified into the classic(severe) and the non-classic (mild) types based on the age of onset and the clinical presentation.[5] Cases of ambiguity of sex detected at birth are due to adrenogenital syndrome unless proved otherwise. It is important to manage features of aldosterone deficiency, which may be associated as this may present as an emergency.[4] Surgery remains a valid treatment option for cases with markedly ambiguous genitalia.[6]

CASE PRESENTATION

The patient is a 22-year-old single undergraduate who resides with her parents. She presented with an inability to achieve penetrative vagina intercourse due to a lack of appropriately sized vaginal introitus which was first noticed in childhood but dismissed. This abnormality became pronounced after puberty with associated clitoromegaly, especially at sexual arousal. This gradually became a source of embarrassment and distress for her, and she always avoided being naked in the presence of anyone, including her female friends. She also reported feeling ashamed when she was sexually aroused in the presence of her sexual partner due to clitoromegaly. There was associated poor urinary stream, terminal dribbling, and post-void wetting of her underwear.

Her onset of secondary sexual characteristics was at 17 years, and menarche was at 18 years. She had normal, well-developed female breasts, but she had excessive hair growth involving her face, chest, and abdomen, requiring regular shaving. There has been associated oligomenorrhea with cycle length varying between 60 and 120 days. History of minimal expressible galactorrhea in both breasts. No history was suggestive of salt wasting, hypoglycemic crises, or stunted growth. She has no heat or cold intolerance, no neck swelling, no eye pain or recurrent frontal headaches, no visual impairment, nor absence of perception of smell. There was no history of insertion of corrosives or other foreign bodies into the vagina or of abnormal vagina discharge in childhood. There was no history of similar complaints in any of her two older sisters or other known family members. Mother did not use non-prescription medication during pregnancy, and there was no exposure to radiation.

She had hirsutism, a male pattern of abdominal hair distribution, clitoromegaly with fused labia marked only by hyperpigmentation, and a narrow introitus of <2cm between the clitoris and the fused labia. The urethral meatus was only visible on parting the fused labia, with no palpable gonad in the inguinal or perineal regions. A capacious vagina was felt behind the fused labia. Digital rectal examination revealed an anterior rectal wall that is depressible to a length of about 8-10cm with a firm bulge into its proximal end which is suggestive of a vaginal canal with proximal ectocervix.

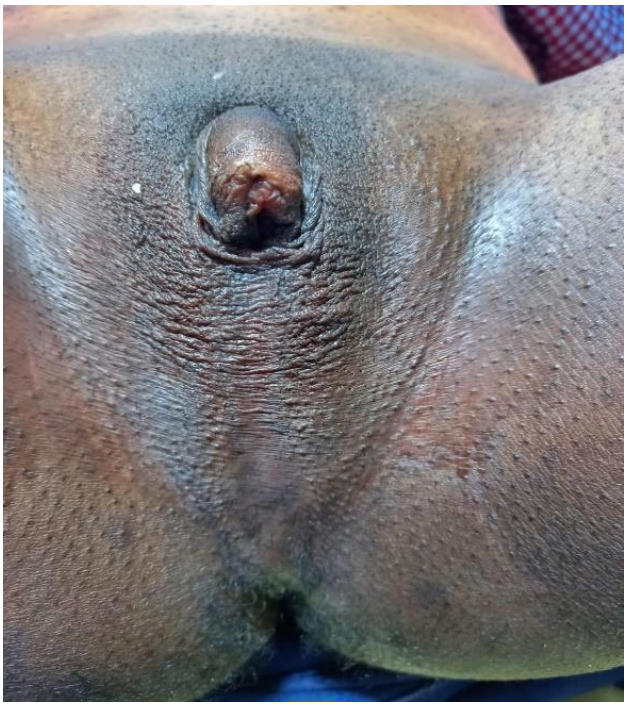

Fig 1, Perineum showing the clitoromegaly with fused labia

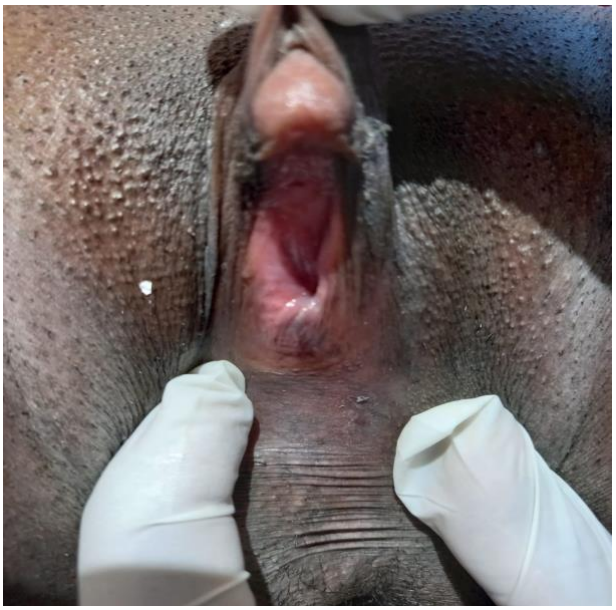

Fig 2. The perineum showing the clitoromegaly with the narrow vaginal introitus in-between parted fused labia

A clinical diagnosis of a possible disorder of sex development (likely XX variant) was made. Ultrasound scan showed a small-sized uterus measuring 23 X 33 X 52mm with an endometrial thickness of 5mm. The ovaries were normal looking with bilateral functional cysts (Right-20mm; Left-28mm). The hormonal profile showed slightly elevated luteinizing hormone of 15.8 IU/mL (1.6 -15.0 IU/mL), normal FSH level – 6.8IU/mL with LH/FSH ratio of 2.3, Serum testosterone level was normal (1.2 nmol/L). The fasting plasma glucose, serum electrolytes, urea, and creatinine were normal as well. Karyotyping was not available and enzyme assays could not be done due to financial constraints as they were not readily available.

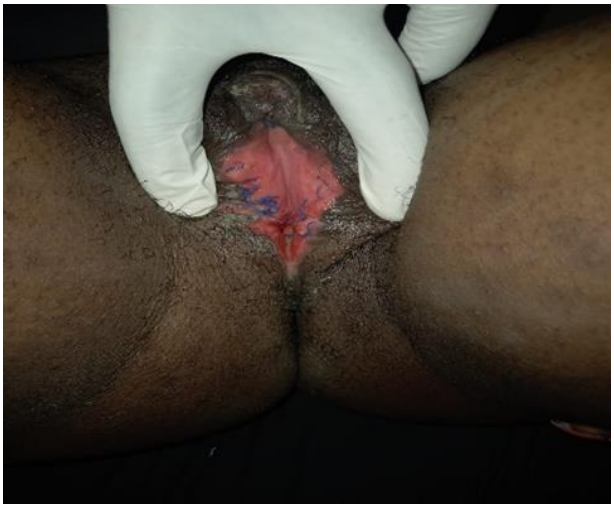

Fig 3. Perineum post-clitoroplasty and perineoplasty with separation of the fused labia.

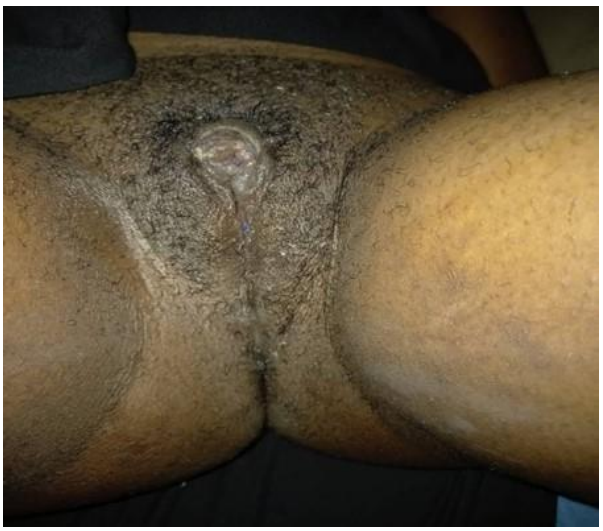

Fig 4. Perineum three months post-clitoroplasty and perineoplasty

She had regional anesthesia; saddle block technique. Examination under anesthesia is the same as described above with findings of a urethral meatus obscured but the fused labia, 2cm (stretchable) opening into a capacious vaginal of about 8cm in length, and smooth cervix felt. She had perineoplasty with separation of the fused labia and suturing apart of each leaf to secure hemostasis. She also had clitoroplasty with excision of the bulk of the erectile tissue. preservation of the neurovascular bundle and prepuce.

Speculum examination after perineoplasty showed normal normal-looking cervix. An improvised vagina mold was inserted into the vagina to keep the separated labia leaves apart for 24 hours. She had a satisfactory post-operation period and was discharged 5th day after surgery. Follow-up reviews at 2 weeks as well as 3 months post-operation showed normal findings. (Figs. 3 and 4 respectively). She has since resumed sexual intercourse which she describes as satisfactory. She was counseled and referred to the reproductive endocrinologist and clinical psychology teams.

DISCUSSION

Ambiguous genitalia represent a rare but important group of disorders. The exact prevalence of Disorders of Sexual Development is not known, especially in developing countries.[7] In most developed countries, children with Disorders of Sexual Development are often diagnosed in infancy, with interventions planned that may be implemented in adolescence. It is, however, not unusual for children with ambiguous genitalia in developing countries to present in adolescence or even much later. This may, however, be associated with the onset of conflict of sexual identity. Some individuals may never present but rather develop means to cope with the disorder.[1,8] Although this patient had ambiguous genitalia, which was noticed since childhood, she did not present until 3 years after her first failed attempt at sexual intercourse at the age of 22 years.

Ambiguous genitalia represent a spectrum of disorders of sexual function which may range from being a feminized male to a masculinized female.[2] Ambiguous genitalia may be a part of a syndrome involving other organ systems.[1,2] The decision of the sex of rearing can be critical and must be based on several factors, including the appearance of the genitalia, the possible outcome of gender assignment surgeries, and the genetic sex of the patient. It should be noted that as much as about one-quarter of such children may grow up and be dissatisfied with their sex of rearing.[9] This patient was raised as female, which was the sole decision of the parents, and at puberty, she developed

female secondary sexual characteristics, and the internal genitals were well-developed Mullerian duct, as is often the case with Congenital Adrenal Hyperplasia.

Congenital Adrenal hyperplasia (CAH) is one of the most common causes of ambiguous genitalia in females. It is an autosomal recessive disorder resulting from a deficiency of one of the five enzymes required for the synthesis of cortisol. The most common of which is 21α-hydroxylase deficiency. CAH may be associated with salt-wasting from mineralocorticoid deficiency and failure of glucocorticoid functions, with life-threatening effects.[5] CAH may rarely be associated with microcephaly.[10] Congenital Adrenal Hyperplasia often has fewer clinical manifestations in males, except in cases with 5α reductase deficiency, which may appear as feminization of the male. It is common for such males with ambiguous genitalia to be initially raised as females until puberty.[11]

Parents of neonates with ambiguous genitalia often experience some degree of anxiety with the decision on gender role assignment. The dilemmas that parents face in the complex and challenging situation following the birth of a child with ambiguous genitalia require early support from insightful professionals such as nurses. This dilemma is worsened by the background of socio-cultural expectations to determine the gender within a short period of at most a few days after birth. [6]

Psychosocial aspects of the treatment of disorders of sex development (DSDs) concern gender assignment, information management and communication, timing of medical interventions, consequences of surgery, and sexuality. Although the outcome is often satisfactory, a variety of medical and psychosocial factors may jeopardize the psychological development of children with DSDs. This sometimes results in the desire to change gender later in life.[12] In this case, she was raised as female and developed female secondary sexual characteristics albeit later than her peers.

A thorough evaluation of infants, adolescents, or adults with ambiguous genitalia should be done to understand the pathophysiology and determine the cause. This evaluation is often better with a multidisciplinary approach.[9] Karyotyping is an important investigation but it is not readily available in Nigeria, although Barr body examination may be helpful. It is also common that many of our patients cannot afford biochemical hormonal assays required to make specific diagnoses of CAH, which are also often not readily available, as it was the case. [13] Radiological, surgical, and histological examination of the internal genitals can contribute to diagnosis.[13] The Ultrasound scan done here demonstrated essentially normal ovaries and uterus, suggesting that internal genitalia were female and she had menstrual cycles, although irregular and characterized by oligomenorrhea. She could not afford enzyme assays, and karyotyping was not available.

Whereas surgery may play a significant role in the treatment of the ambiguous genitalia associated with CAH, it is important to look out for and appropriately treat the life-threatening complications of salt wasting and glucose metabolism defects.[4] The timing of surgical intervention has been a subject of debate, whereas some clinicians believe gender assignment must be done early in childhood, some others argue the need to delay till adolescence or even till the individual is mature enough to determine their preferred sexuality.[6] The type and extent of surgery that can be done will be dependent on the appearance of the external genitalia.[5] The appearance of the clitoris can be a source of concern and psychosocial distress, and the outcome of clitoral surgeries may have a significant impact on sexual function. It is therefore of great importance to counsel the patients and/or their caregivers on the possible outcomes before taking such a step.[14] This patient. was appropriately counseled on this possibility, and she expressed her desire for clitoroplasty despite the possibility of reduced sexual function because the appearance of the clitoris was a source of psychosocial distress for her. This kind of distress is not uncommon in patients with DSD, and there may even be associated psychological dysfunctions.[14]

Clitoral surgeries remain the most common (over 90%) approach in the management of clitoromegaly, with good cosmetic outcomes.[6] It must, however, be noted that cosmetic appearance alone is not an adequate measure of success. Surgical interventions generally have excellent success, at least on a short-term basis. In the short term, patient. had a satisfactory outcome for cosmesis, body image, and sexual function. She is to continue follow up with the reproductive endocrinology and clinical psychology teams for further evaluation and intervention when necessary.

Conclusion

Disorders of sexual development, though rare, may have significant psychosocial effects on the patients. Different variants of these disorders may be seen, and CAH is the commonest cause. Understanding the pathology with appropriate surgical intervention may help improve the quality of life of the affected persons. A multidisciplinary approach to care is necessary to achieve optimal outcomes.

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