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Case Report

Congenital adrenal hyperplasia with clitoromegaly: a case report

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ABSTRACT

Congenital adrenal hyperplasia (CAH) is a rare but significant medical disorder that affects the adrenal glands, causing excessive production of androgens, a group of hormones responsible for the characteristics associated with male sexual development. CAH can present in a variety of forms, with one typical clinical presentation being clitoromegaly, an enlarged clitoris in females. Clitoromegaly, or clitoral enlargement, is a visual manifestation of excessive androgen exposure during fetal development. Clitoromegaly is a sensitive and potentially anxiety-inducing condition, especially for girls and their families. In this report, we present a case of a young girl with clitoromegaly who underwent clitoroplasty.

Keywords: Congenital adrenal hyperplasia, Clitoromegaly, Clitoroplasty

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a rare but significant medical disorder that affects the adrenal glands, causing excessive production of androgens, a group of hormones responsible for the development of male sexual characteristics.

CAH can present in a variety of forms, with one of the typical clinical presentations being clitoromegaly, which refers to an enlarged clitoris. CAH is caused by a genetic mutation in an enzyme that synthesizes cortisol, a hormone important for regulating stress and metabolism.[1] Clitoromegaly, or enlargement of the clitoris, is a visual manifestation of excessive androgen exposure during fetal development. Clitoromegaly is a sensitive and potentially anxiety-provoking condition, especially for girls and their families.

The degree of clitoromegaly can vary, and it may be associated with other symptoms, such as virilization (development of male secondary sexual characteristics) and genital ambiguity.[2]

CASE REPORT

A 4-year-old girl presented with complaints of genital enlargement. The patient had resumed treatment one month prior after a two-year hiatus due to the COVID-19 pandemic. Upon parental anamnesis, it was revealed that the primary concern was genital enlargement. The patient was referred from Karanganyar Regional Hospital and was born via cesarean section at 40 weeks gestation due to premature rupture of membranes. The patient was declared female at birth but her mother noted discrepancies in gender characteristics since then. There were no abnormalities in bowel or urinary function, and the child did not exhibit fever or respiratory symptoms. Laboratory tests and ultrasound examinations conducted were at Dr. Moewardi Hospital.

Genitourinary examination revealed virilization and a phallus measuring 4 cm in length. The external urethral orifice was located below the clitoris and the labia matched the surrounding skin color (Figure 1). The prader staging and virilization of external genitalia in this patient was grade III.

Ultrasound examination of the patient showed that the patient had a normal uterine structure with vaginal introitus structure and no visible testicular image (Figure 2). Chromosome analysis of the patient revealed that 50 cells were counted, 8 cells were analyzed, and the karyotype was determined to be 46, XX. In the metaphase that was counted and analyzed, there were no abnormalities in the structure and number of chromosomes. The karyotype was in accordance with the female sex (Figure 3). A progesterone hormone examination on the patient indicated elevated progesterone levels at 77.07 ng/ml (normal range is ≤3.6 ng/ml).

The patient was resuscitated with D5 ¼ NS 1000 cc/24 hours and scheduled for clitoroplasty. Her routine labs and thorax X-ray were normal. During surgery, both the clitoris and hood were visible with normal urethral opening, and a catheter was installed. A circular incision was made at the proximal part of the hood. The pudendal artery and nerve were separated bluntly from the corporea. Clitoroplasty involved cutting the corporea proximally and resizing the clitoris accordingly before suturing it back to the corporea and labium majora mucosa. (Figure 4). The post operative wound showed no signs of necrosis or bleeding, and urination was normal (Figure 5). A follow-up examination at the pediatric surgery polyclinic confirmed that the clitoroplasty wound appeared healthy without complications. The wound is treated with gentamicin ointment (Figure 6).

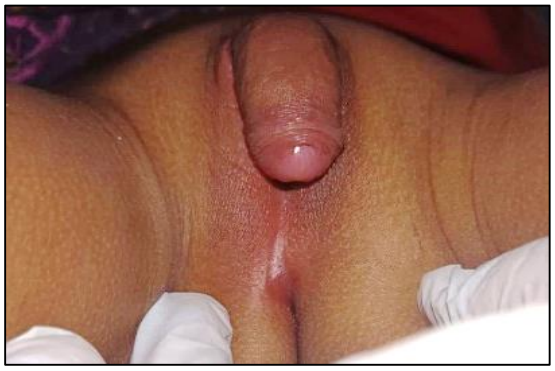

Figure 1: Genitourinary examination.

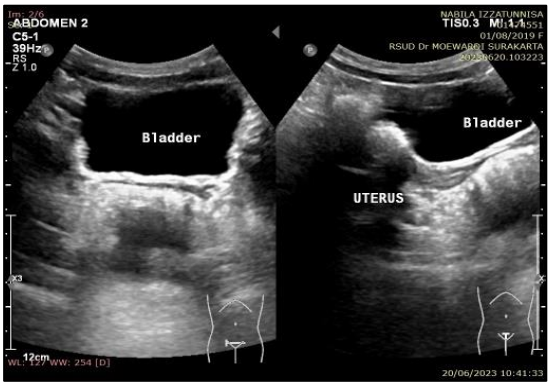

Figure 2: Genitalia ultrasonography.

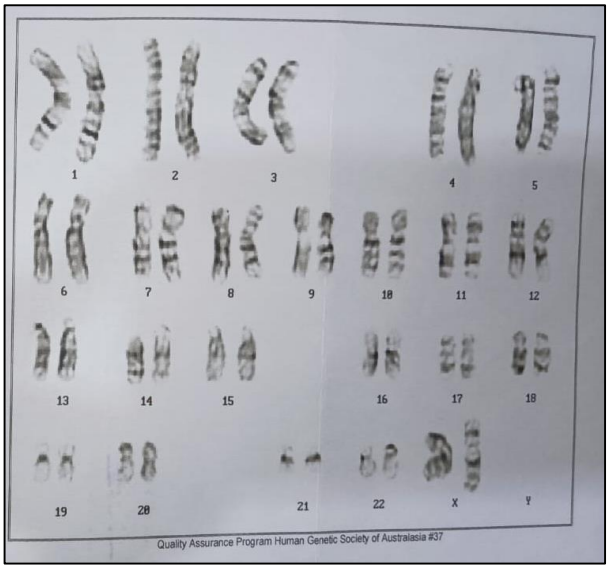

Figure 3: Chromosome examination.

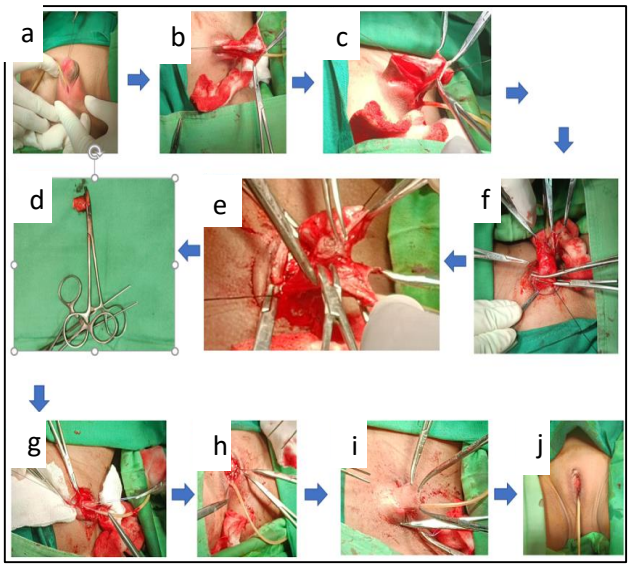

Figure 4: Intraoperative images (a-j).

Figure 5: Post operative wound.

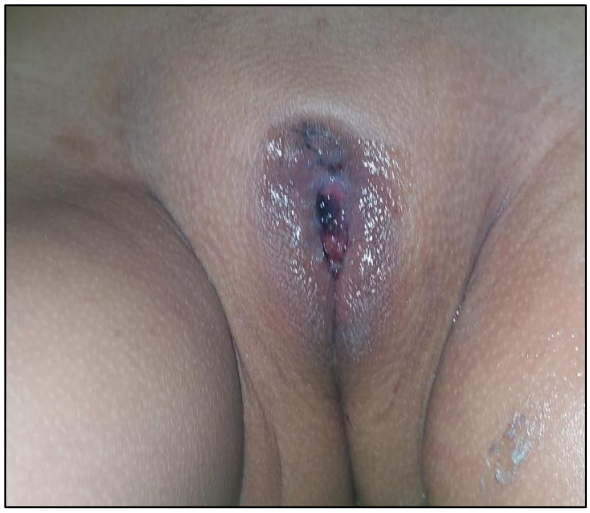

Figure 6: Follow up examination.

DISCUSSION

Clitoromegaly is an abnormal enlargement of the clitoris with various etiologies. This condition is more common in congenital cases while acquired conditions are rare. Several case reports describe clitoromegaly with or without clinical or biochemical hyperandrogenism.[1-2] Disorders of sex development (DSD) are congenital conditions involving abnormalities in the development of chromosomes, gonads, or sex anatomy, which can result in ambiguous genitalia at birth. Clitoromegaly, an enlargement of the clitoris, usually occurs as part of a more complex disorder and is rarely seen as a single condition in intersex patients.[3-5]

The 46, XX DSDs, specifically disorders of hormone synthesis or action, encompass several conditions, with congenital adrenal hyperplasia (CAH) being the most common hormonal cause of virilization of the external genitalia. CAH is caused by enzyme abnormalities in the steroid synthesis pathway, primarily due to mutations in the CYP21A2 gene on chromosome 6p21.3. This gene encodes an enzyme that regulates the production of cortisol and aldosterone. CAH is divided into classic and late-onset or nonclassical CAH (NCCAH), which is often associated with isolated clitoromegaly. Fetal exposure to abnormal levels of androgens during development can cause masculinization of the female genitalia, which may be characterized by clitoromegaly.

Rare enzyme abnormalities such as 17α-hydroxylase deficiency can also cause clitoromegaly at puberty. In addition, other 46, XX DSDs involve disorders of gonadal development, including 46, XX testicular DSD and 46, XX ovotesticular DSD.[4,6] Patients with clitoromegaly should undergo a thorough evaluation, including a detailed medical history, careful physical examination, thorough endocrinological assessment, karyotype, and genetic analysis. In some cases, histologic examination may also be necessary. Appropriate management requires an accurate diagnosis.[3,7] Clitoral pain or enlargement may occur following incomplete retraction or reduction of the clitoris, especially in patients with CAH. In a study by Newman et al., two out of four patients with CAH experienced pain with sexual stimulation after surgery. In-depth counseling is needed before reoperation to explain the risks.[8]

Genital surgery in children with clitoromegaly is controversial in disorders of sex development (DSD). There are three rationales for performing this surgery: to create an anatomy appropriate for sexual intercourse, to allow urination consistent with gender identity, and to provide a physical appearance consistent with assigned gender. However, this surgery also carries the risk of reducing clitoral innervation, which can lead to decreased sensation and difficulty achieving orgasm later in life for women who undergo surgery as children. Medical guidelines recommend delaying surgery for mild clitoromegaly (<2 cm) and considering more experienced surgeons for severe cases. Clitoral surgery is often performed in conjunction with vaginoplasty to utilize the common urogenital sinus for reconstruction. If vaginoplasty is not performed in conjunction, the use of sinus tissue is still important to create a vulva that is more consistent with a female appearance.[7,8]

There are three types of surgical techniques to manage clitoromegaly: clitorectomy, reduction clitoroplasty, and clitoral corpus preservation techniques.[9] Clitoral pain or enlargement may occur after incomplete retraction or reduction of the clitoris, especially in patients with CAH. In a study by Newman et al, two out of four patients with CAH experienced pain with sexual stimulation after surgery. In-depth counseling is needed before reoperation to explain the risks.[10] Clitoral atrophy may occur in patients undergoing clitoroplasty, especially after subtotal or total resection of the clitoral shaft. Several studies have reported atrophy of the glans clitoris, including children with a mean age of 13 years. It is important to preserve the vascularity of the glans and minimize traction on the nerve sheath and blood vessels to reduce the risk of vasospasm.[11,12] Clitoral surgery carries the potential for neurologic injury.

Several studies using intraoperative nerve imaging and pudendal evoked potential measurements have shown insignificant electromyographic responses. Clitoroplasty with preservation of the dorsal nerve can result in normal sensation and orgasmic potential, but results are variable.[13,14]

The optimal timing for clitoroplasty is still controversial. Some suggest that older patients tend to prefer earlier surgery, while others argue that delaying surgery empowers the individual to participate in the decision. The psychological impact of later surgery may be more significant, but there is no definitive evidence to determine whether earlier or later surgery is better.[15,16]

CONCLUSION

This case highlights the importance of multidisciplinary management in addressing congenital adrenal hyperplasia associated with clitoromegaly. Surgical intervention not only addresses physical concerns but also plays a crucial role in supporting the psychological well-being of patients and their families.

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