Clitoromegaly with Associated Anomalies in the Female of a Fraternal Twin: A Case Report

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ABSTRACT— ***We report this uncommon case of clitoromegaly with associated anomalies in a 2 months old female, the first of fraternal twins. The initial diagnosis of ambiguous genitalia on karyotyping was changed to clitoromegaly most probably due to congenital adrenal hyperplasia (CAH).***
Karyotyping, biochemical analysis, ultrasonography, contrast studies and magnetic resonance imaging were done as the preoperative workup of this patient.

Keywords— Ambiguous genitalia, clitoromegaly, female fraternal twin, karyotyping

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1. INTRODUCTION

The clitoris is a female sexual organ that is similar in nature to the male penis [1].

Clitoromegaly is a congenital or acquired anomaly of the female genitalia and represents an abnormal enlargement of the clitoris [1]. The typical clitoris as documented by Dickinson[1] in the Atlas of Human Anatomy, has a width of 3mm-4mm and a length of 4mm- 5mm[1].

The commonest cause of clitoromegaly is congenital adrenal hyperplasia (CAH) [1, 2]. The use of progestogens by pregnant women in early pregnancy is known to cause virilisation in the female infant [1]. Prematurity is a rare associated factor with clitoromegaly as the androgen levels are high in such infants [2, 3]. The rare Fraser's syndrome and the rare maternal virilising ovarian tumours are all known causes of clitoromegaly [1].

We report this uncommon case of a 2months old female, the first of a fraternal twin with congenital clitoromegaly with co-existing urogenital and musculoskeletal anomalies.

2. CASE REPORT

A 2 months old female, confirmed by karyotyping, the first of a fraternal twin born prematurely at 33weeks of gestation presents at birth with ambiguous genitalia. Pregnancy till 33weeks and delivery were uneventful .Maternal history ruled out the use of progestogens during pregnancy and antenatal imaging was negative for adrenal or pelvic mass in the mother. She however, had been on injectable contraception (depo –provera) for a year to effect birth control and conception occurred 8 months on cessation of the contraception.

Clinically, a live infant, not pale and anicteric was seen.

The respiratory and cardiovascular systems were essentially normal.

She had an umbilical hernia.

Examination of the genitalia and perineum revealed what looked like a micropenis measuring 20mm by 13mm in length and width, as well as a prominent labioscrotal fold. A mass was palpated in the left labioscrotal fold. An anterior midline perineal opening distal to the micropenis and a posterior anal orifice were visualized. Mild bilateral talipoequinovarus deformity was noted. A clinical diagnosis of ambiguous genitalia secondary to CAH was made.

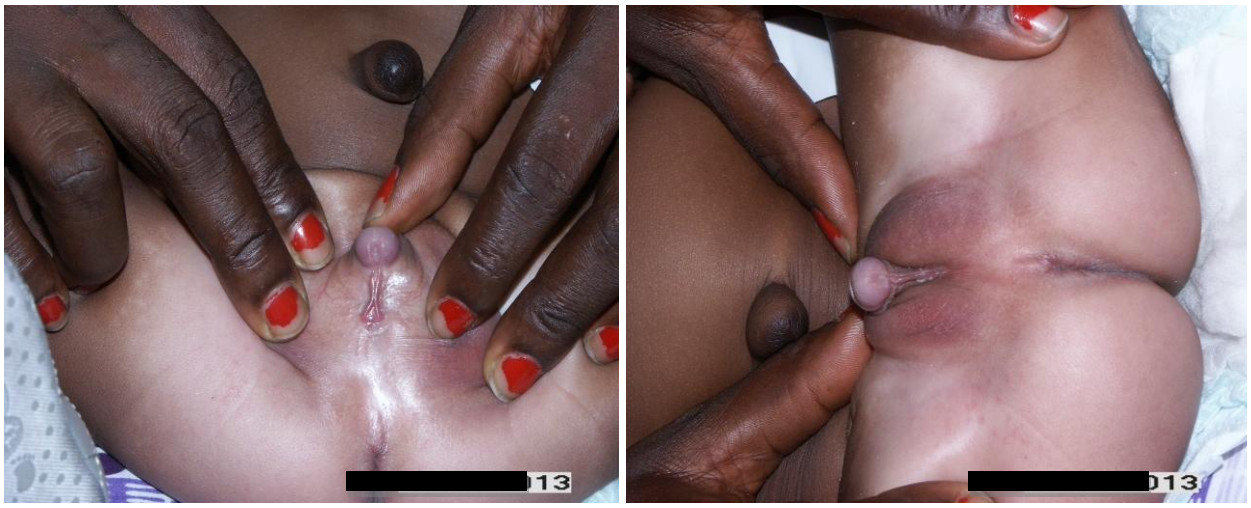

Figure1 Shows a micropenis like structure and hypertrophy of the labia majora

Karyotyping (genotype) showed a modal number of 46XX karyotype in 14 cells analyzed characteristic of a normal female chromosomal pattern.

The neonate's 17-hydroxyprogesterone (17-OHpg) level of 1.7nmol/L was within the normal reference for the patient's age.

Ultrasound scan of the abdomen was essentially normal, focusing on the adrenal glands and kidneys.

The pelvic scan using a 7.5 MHz probe revealed an anteverted infantile uterus with an endometrial cavity line, **Figure 2.**

Continuous with the uterus in its postero- inferior aspect is a dilated anechoic collection, representing fluid in a dilated blind ending vagina (hydrocolpos). The ovaries were not visualized.

An ovoid soft tissue mass was seen in the left half of the labioscrotal fold by ultrasound which is most probably an ovarian or testicular tissue.

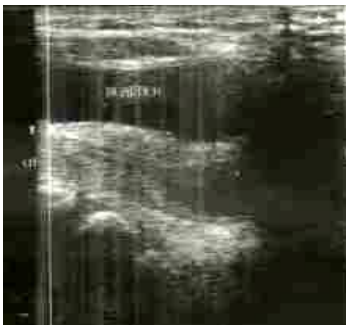

Figure2 Showing the anteverted uterus and hydrocolpos

Genitogram, **Figure 3**, done via the anterior perineal opening showed a dilated vagina (hydrocolpos), with contrast filling the anteverted uterus retrogradely. A modified enema study revealed a normal rectum.

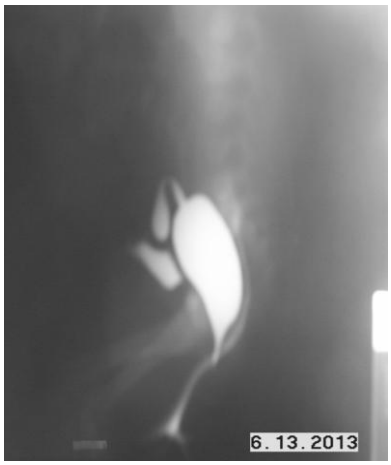

Figure3 Showing the contrast filled vagina, cervix and uterine cavity

MRI scan, Fig 4, demonstrates the urinary bladder with a urachal diverticulum, the uterus with the hydrocolpos, a normal rectum and lumbosacral spine. An ovoid hypo intense mass, **Figure 5**, with a central intensity in the left labiosrotal fold is a probable ovary or testis.

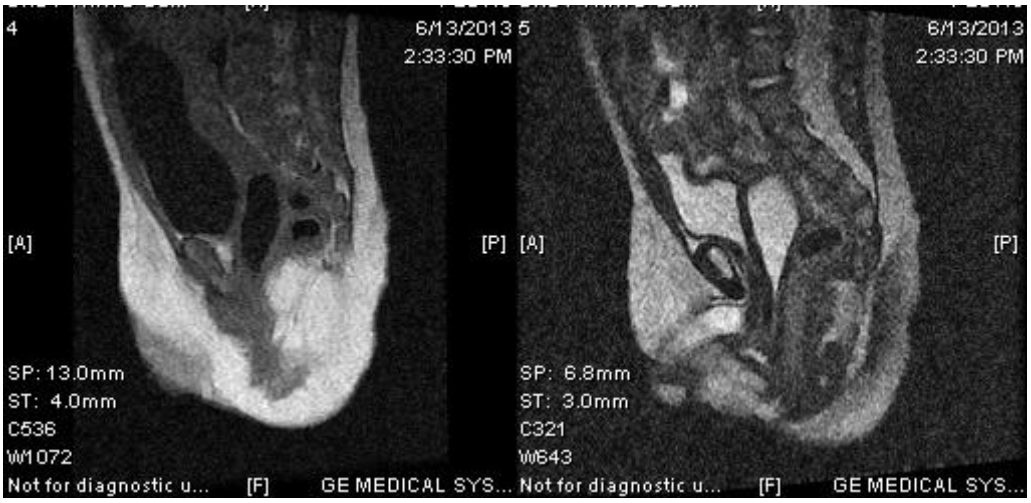

Figure4 T1W anad T2W MRI scans demonstrating the urachal diverticulum and hydrocolpos

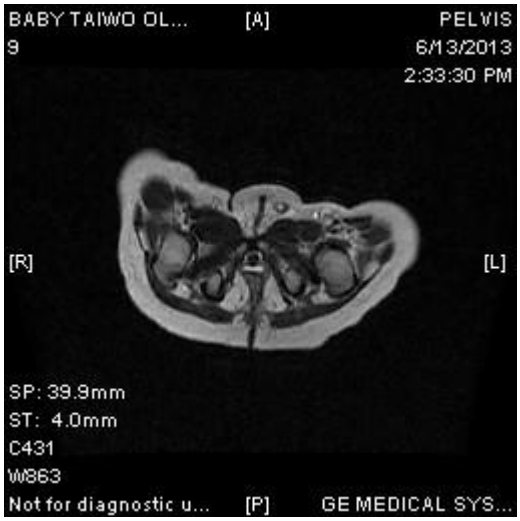

Figure5 Showing mass in the left labioscrotal fold

3. DISCUSSION

Ambiguous genitalia can be defined as genitals that are neither distinctly male nor female. Clitoromegaly is rare and can be congenital or acquired with the congenital variety being commoner [1].The most common cause is hormonal due to excessive androgen [1, 2, 3, 4]. Rare non-hormonal causes have also been documented [4].

Neonatal clitoromegaly like the case presented is usually attributed to androgen stimulation secondary to CAH or in- utero androgen exposure [1, 2, 3]. CAH is the commonest cause of ambiguous genitalia in the newborn [1, 2, 3, 4,5].The excess production of androgens results in a female with a virilised phenotype.CAH is an autosomal recessive genetic condition and biochemical confirmation of the diagnosis is with elevated 17 hydroxyprogesterone(17OHP) and androgens. However, in the indexed patient, the 17 ketosteroid level was normal.

We believe that persistent high levels of progestogens in the mother from thedepo- provera contraception used for birth control combined with prematurity are the most probable causes of the virilisation in our patient .Williams et al [3] talked about persistent unexplained clitoromegaly in females born extremely premature. The infant presented was born premature at 33weeks with a grade 3 clitoromegaly [5, 6] according to the Prader's classification.

Few cases have been documented in literature of one of a fraternal twin having clitoromegaly. Gunther et al [7].in their study, showed a female fraternal twin who had CAH diagnosed antenatally. The female twin was normal, but an older male sibling earlier diagnosed with CAH died at 18 months of age thus confirming heredity [7]. The male fraternal twin in this case with the three older siblings appears normal. There was no family history of CAH.

The association between ambiguous genitalia and urogenital sinus anomalies is well documented [5, 8, 9, 10]. In this patient the urethra and vagina both open into a common channel on the perineum [5, 8, 9, 10]. In the evaluation of ambiguous genitalia, karyotyping, biochemical analysis, genitogram, voiding cystography, ultrasonography and magnetic resonance imaging may be used. Magnetic resonance is the imaging modality of choice because of its excellent soft tissue detail. The non- availability of this equipment and the cost of examination limits its use in most developing economies.

In conclusion, the persistent high levels of circulating maternal progestogens in a woman on injectable contraception should be borne in mind as a possible cause of ambiguous genitalia in the female infant.

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