CONGENITAL ADRENAL HYPERPLASIA

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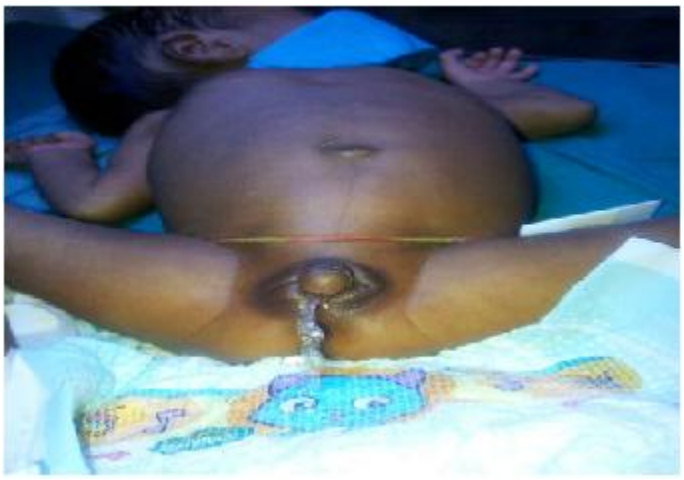
ABSTRACT: The birth of a child with ambiguous genitalia and distressing event for the family and physician. It is important to make a diagnosis, for psychological, social, and medical reasons for particularly for recognizing accompanying life threatening disorders such as the salt losing form of congenital adrenal hyperplasia.
KEYWORDS: Ambiguous genitalia, congenital adrenal hyperplasia, Disorders of sex development, Neonates

INTRODUCTION: The evaluation and management of a newborn with ambiguous genitalia must be undertaken with immediacy and great sensitivity. The pediatric urologist, endocrinologist, geneticist, and child psychiatrist or psychologist should work closely with the family in pursuing a dual goal: to establish the correct diagnosis of the abnormality and, with input from the parents, determine gender based on the karyotype, endocrine function, and anatomy of the child. In this section the authors outline a practical approach to the neonate born with a disorder of sex development (DSD).



CASE REPORT: A 25 days, old proband, born to consanguineous couple was admitted with the chief complaint of refusal to feed, lethargic and with ambiguous genitalia. The patient was subjected to the following scheme of examination: a detailed clinical history, family history, physical examination with photographs, chromosomal analysis and Karyotyping, reference for relevant special investigations and treatment.

A. HISTORY

1. Family history of
   CAH : Absent
   Hypospadias : Absent
   Infertility : No
   Pubertal delay : No
   Genital surgery: No

2. Death of previous sibling : No death
3. Maternal drug exposure : No Exposure
4. Maternal virilization : NO Maternal virilization
5. Placental insufficiency : NO

B. PHYSICAL EXAMINATION

1. External genitalia - Absence of vaginal opening, Pigmentation
   - Hypertrophied clitoris, increased anogenital ratio
2. Gonadal size, position and descent
   Clitoromegaly
   No inguinal hernia
3. Bimanual rectal examination cervix, uterus palpable in midline
4. Associated anomalies No Dysmorphic features and associated anomalies.

Diagnostic tests:

1. Laboratory tests

a. First- line testing: Karyotype 46XX

Serum Electrolyte

- Serum calcium :8 MG%
- Serum sodium :179.6 mmol/L
- Serum potassium :8.96 mmol/L
- BU 355.3mg/dl
- Creatinine ;2.9 MG%
- 17-hydroxyprogesterone: 27.00 ng/ml
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Testosterone: 1000ng/dl

b. Other tests: Pelvic ultra sonography:

- No sonologically detected abnormality in the abdomen and pelvis.
- Uterus is visualized and appears normal for the age.
- No E/O renal pathology.

DISCUSSION: 46 XX disorder of sex development Girls with 46 XX DSD, the most common DSD, are 46 XX with normal ovaries and Mullerian derivatives. The sexual ambiguity is limited to masculinization of the external genitalia that occurs as a result of exposure to androgens in utero. Congenital adrenal hyperplasia (CAH), which accounts for the majority of patients with 46 XX DSD, describes a group of autosomal recessive disorders that arises from a deficiency in one of five genes required for the synthesis of cortisol. The most common cause of CAH is inactivation of CYP21, which catalyzes the conversion 17-OH progesterone to 11-deoxycortisol, a precursor of cortisol, and the conversion of progesterone to deoxycorticosterone, a precursor of aldosterone. A spectrum of phenotypes from mild to severe clitoromegaly is possible. Classic 21a-hydroxylase deficiency is comprised of two forms of CAH: a severe, salt-wasting type with a defect in aldosterone biosynthesis and a simple, Virilizing type with normal aldosterone synthesis. A mild, non classic form also exists that can be asymptomatic or associated with signs of postnatal androgen excess. These newborns have severe CAH and exhibit signs of mineralocorticoid and glucocorticoid deficiency in the first week of life. Masculinization occurs as a result of DHEA conversion to testosterone in fetal placenta and peripheral tissues manifesting as mild to moderate clitoromegaly.

TREATMENT:

Symptomatic treatment.

- Broad spectrum antibiotics
- Correction of electrolytes
- Maintain good hydration
- Corticosteroids therapy
  Hydrocortisone 20mg/m2/24hr
  Fludrocortisone 0.3mg

Surgical treatment

REFERENCES:

1. 1 Thyen U, Lanz K, Holterus PM, et al. Epidemiology and initial management of ambiguous genitalia at birth in Germany. Horm Res 2006; 66:195.
2. Rogers BO. History of external genital surgery. In: Horton CE, editor. Plastic and reconstructive surgery of the external genitalia. Boston: Little Brown and Company; 1973. p. 3–50.
3. Speiser PW, White PC. Congenital adrenal hyperplasia. N Engl J Med 2003; 349:776.
4. Hughes IA, Houk C, Ahmed SF, et al. Consensus statement on management of intersex disorders. Arch Dis Child 2006; 91(7):554.
5. Morel Y, Rey R, Teinturier C, et al. Aetiological diagnosis of male sex ambiguity: a collaborative study. Eur J Pediatr 2002; 161:49.

CASE REPORT

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