Original Research Paper

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Paediatric Surgery

46 XX DSD : OUR INSTITUTIONAL EXPERIENCES

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ABSTRACT Disorder of sexual differentiation (DSD), previously known as intersex disorder is a spectrum of congenital disorders that are characterized by dissonance between anatomical, chromosomal, gonadal, hormonal and psychological sex of the individual. A neonate born with DSD is a social emergency and it is better handled by a well informed clinician.

KEYWORDS : DSD, Female DSD, 46 XX DSD, CAH

INTRODUCTION:

Disorder of sexual differentiation (DSD), previously known as intersex disorder is a spectrum of congenital disorders that are characterized by dissonance between anatomical, chromosomal, gonadal, hormonal and psychological sex of the individual. Diagnosing the type of disorder may now be easier than before, but assigning the sex of rearing is very difficult, especially in Indian scenario as it depends largely on the parental wish.

The most common subtype of 46 XX DSD is congenital adrenal hyperplasia (CAH). Incidence is around 1:15000 live births.[1] It is caused by excess of androgen in genetic females due to deficiency of enzyme responsible for synthesis of steroid hormones in adrenal gland resulting in virilization. Most common enzyme deficiency responsible for CAH is 21 alpha hydroxylase.[2]

Case Description :

- 3 Cases were referred to us from ENDOCRINOLOGY Department with presentation of ambiguous genitalia, all were under 5 years of age.
- 2 Cases of PRADER 3 AND 1 Case of PRADER 5
- One of PRADER 3 was due to CAH and other due to AROMATASE deficiency.
- PRADER 5 was due to CAH (21 ALPHA HYDROXYLASE DEFICIENCY)

- **Pre operative picture(Fig.1)**

Fig.1(prader 3)

Management:

After medical management, we planned for operation: feminising genitioplasty (clitoroplasty & vaginoplasty)

Intraoperative steps (Fig.2)

- Simultaneous clitoroplasty and u-shaped vaginoplasty
- Stay sutures placed in glans with skin marked for circumcision
- Markings of for perineal u- shaped flap and preservation of urethral groove
- Incision of urogenital sinus and approximation of u-shaped flap to vaginal vault and extension of ventral urethral incision to circumcision incision
- Circumscribed skin is used to make Labia minora.
- Fixation of glans to ventral pubic periosteum done with delayed absorbable suture.

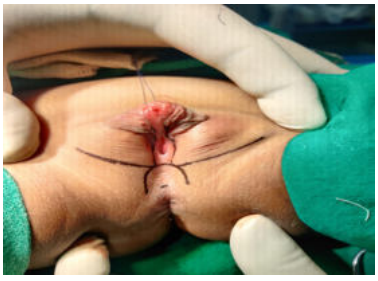

Fig.2

- **Post operative outlook (Fig.3)**

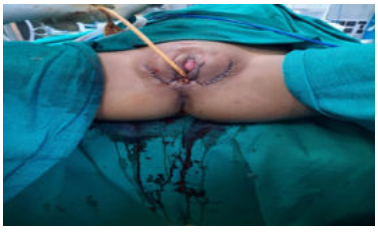

Fig.3

Case Discussion:

- Most common cause of 46 XX DSD is CAH. Enzyme deficiency causing CAH are 21 hydroxylase (most common), 17 alpha hydroxylase, 3 beta hydroxysteroid dehydrogenase, 11 beta hydroxylase and cholesterol desmolase. Clinically they are classified into 2 type. The salt losers variety present with electrolyte imbalance and virilised genitalia in the neonatal period [7].The non salt losers children are present late with varying degree of virilization [7].
- Physical Examination is very essential for diagnosis as well as future treatment plan. PRADER has classified presentation of virilisation in 5 types(Fig.4).

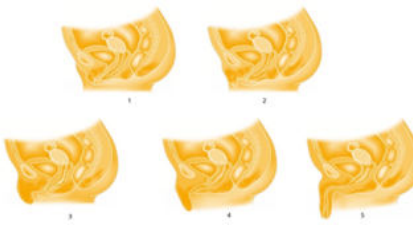

Fig.4 Prader Staging

- Medical management with glucocorticoids and mineralocorticoids must be done from neonatal stage.[3]
- Mild degree of virilisation (upto PRADER 2) does not warrant any surgery.

- After medical management,parents consult paediatric surgeons for clitoromegaly and urogenital sinus.Hence diagnostic cystoscopy is very important to measure the length of common channel,prior going for feminizing genitoplasty.
- Vaginoplasty may be done by either Furtunoff U flap[4] or Passerini-Glazel technique[5].
- Parental counseling regarding the nature of disease and the sex of rearing are very important.

- **Gender Assignment : Controversy?**

90 % ASSIGN THEM AS FEMALES
BUT 5 % ARE GENDER DYSPHORIC[6]
Hence few surgeons are of the opinion to wait till puberty when the child can give her opinion.

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