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A RARE CASE OF AMBIGOUS GENITALIA

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Introduction

Disorders of sex development (DSD) are rare disorders occurring when there is a discordance between chromosomal, gonadal, or phenotypic sex. These occur in the presence of genetic mutations that affect one of the two major processes in sex determination or sex differentiation, sex development. In sex determination, the biopotential gonad is genetically programmed based on the sex chromosome complement to become either a testis or ovary. Sex differentiation occurs in the presence of a formed testis or ovary, and is dependent upon the ability of the gonad to produce hormonal factors and/or the presence of the appropriate receptors in extragonadal tissues.

Aim

Aim of the study is to bring awareness about disorders of sex development (DSD) among general public to assign gender at the earliest to prevent psychological stress for both parents and the child

Case Report

A 23 yr old primigravida at gestational age 34 weeks came for regular antenatal checkup .USG obstetrics done at 34 weeks which was suggestive of ambiguous genitalia. At 35 weeks emergency lscs done in view of preterm premature rupture of membranes with fetal distress. She delivered a baby with ambiguous genitalia with birth weight 2.3kgs with apgar 8/10,9/10. USG done for the baby in which both testis were seen in scrotal sac with maintained vascularity. There is no evidence of uterus and ovaries and per vaginal canal.

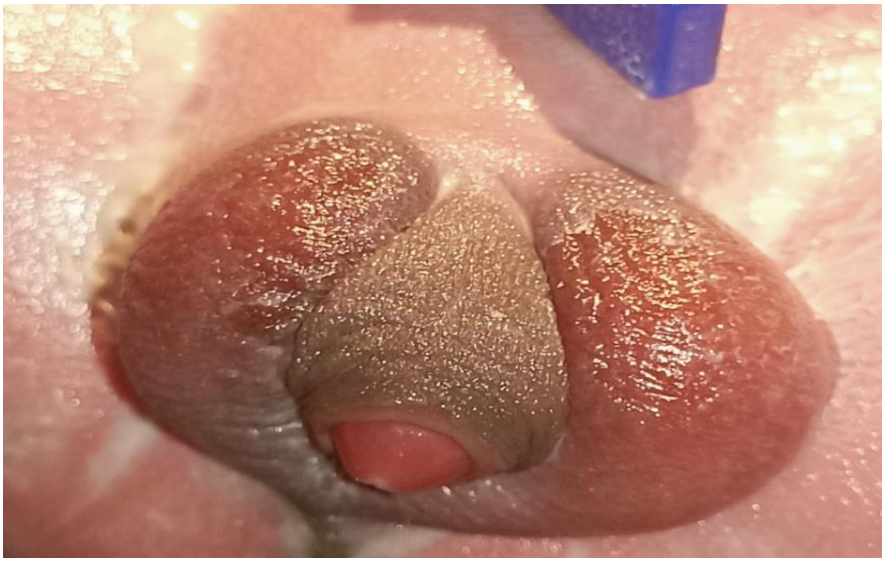
There is no obvious renal anomolies. Karyotyping was done which showed 46XY. 17 hydroxy progesterone levels were found to be normal. Testosterone levels were found to be normal. Dihydrotestosterone levels were found to be reduced. Based on the investigations and karyotyping baby was diagnosed clinically to have 5 alpha reductase enzyme deficiency

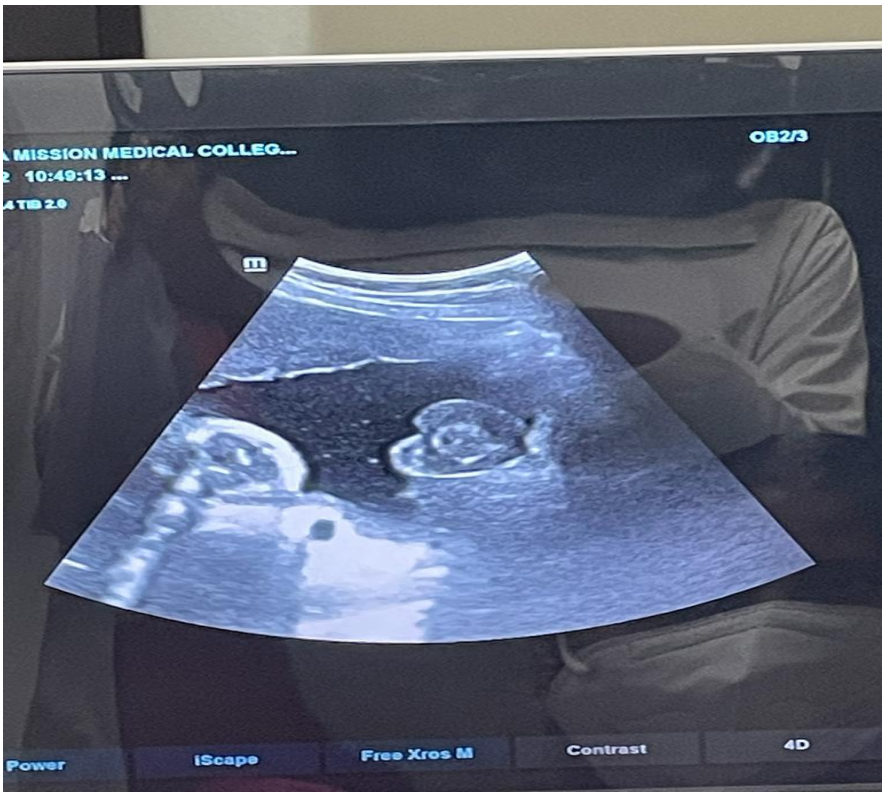
Discussion

5-AR enzyme catalyzes testosterone to Di hydro testosterone. DHT is broadly responsible for male phenotypic development and pubertal androgen-mediated tissue growth. 5-AR deficiency involves mutation in the 5-AR enzyme It is a very rare disorder seen most commonly due to a mutation in the gene SRD5A2 located on the short arm of chromosome 2 [1]. Children with 5-AR deficiency are 46XY karyotype and have varying presentations of their external genitalia ranging from a microphallus with degrees of hypospadias to a phenotypic female with clitoromegaly. Often these patients have a bifid scrotum and a diminutive phallus can be mistaken for female genitalia with clitoromegaly [2,3]. Management of ambiguous genitalia requires a multidisciplinary approach. Patients with 5-alpha reductase deficiency who are raised as female requires orchidectomy and hormone replacement therapy. Who are raised as male requires phalloplasty, scrotoplasty, urethroplasty, orchidopexy [4,5].

Conclusion

Children with 5-alpha reductase deficiency are often raised as girls till puberty. Some of these individuals exhibit virilization at puberty, which is often accompanied by gender identity disorder from female to male. Management of ambiguous genitalia in the newborn requires an entire multidisciplinary team in every step of the diagnostic procedure, the choice of sex assignment and the treatment strategy. It is important that the diagnosis be made in infancy by biochemical and molecular studies before gender assignment to prevent psychological stress for parents and child.

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