

A RARE CASE OF XY DISORDER OF A CHILD WITH AMBIGUOUS GENITALIA AND UNILATERAL ANORCHIA

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ABSTRACT

Introduction: In this disorder of sex development (DSD) the chromosome number is 46 XY. The pattern is normally found in males. In this condition an individual have one X chromosome and one Y chromosome in each cell. The genitalia of the individual cannot be distinguished clearly whether they belong to male or female. Infants having this disorder tend to have penoscrotal hypospadias, abnormal development of the testes, and reduced to no sperm production. **Methodology:** 8 yr old male child came with complaints of abnormal genitalia since birth. On examination, Visual anatomical analysis, Karyotyping and ultrasonographic evaluation like pelvic ultrasonography and transrectal ultrasonography done. **Result:** the child has a micropenis and on left sided unilateral anorchia. Urination through the female like orifice with labia major likes structure present anterior to micropenis. The pubic hairs are yet to develop. karyotyping the 47,XY karyotype is confirmed. After considering case history both parents were found to be first-degree cousins. **Conclusion:** The 46, XY DSD may be raised as males or females. The patients may have increased risk for gonadal tumors and may benefit from regular surveillance or surgery to remove abnormally developed gonads. The reconstructive surgery to external genitalia is advisable to be raised as males or females on maturity

KEYWORDS: disorder of sex development, unilateral anorchia, Karyotyping, Ultrasonography.**INTRODUCTION**

In this disorder of sex development (DSD) the generally chromosome number is 46 XY. The pattern is normally found in males. In this condition an individual have one X chromosome and one Y chromosome in each cell. The genitalia of the individual cannot be distinguished clearly whether they belong to male or female. Infants having this disorder tend to have penoscrotal hypospadias, abnormal development of the testes, and reduced to no sperm production.^[1] Anorchia, or vanishing testis syndrome, is defined as the lack of testes in a 46,XY individual with a male physical composition.^{[2]-[4]} It impacts one in 20,000 male births.^[5] Although some patients with anorchia present with ambiguous external genitalia.^[7] or micropenis^[8], most have a normal phenotype. The familial occurrence of anorchia.^{[2],[5],[7],[9]} and its association with other anomalies^[10] suggests a genetic origin, but the genetic cause remains unknown. The families of some patients with anorchia may include other individuals with pure or partial 46,XY gonadal dysgenesis. However, exploratory laparoscopy has suggested that at least some cases of anorchia are the result of prenatal testicular vascular accident associated with torsion during testicle descent.^[12]

A parent of 8year old child with ambiguous genitalia was referred for genetic laboratory, at the birth, child was considered as female. Ambiguous genitalia are a condition where there is an abnormal development of the genital organ, which creates a question about the child's gender. On examination, the child has a micropenis and on left sided unilateral anorchia. Urination through the female like orifice with labia major likes structure present anterior to micropenis. The pubic hair yet to develop. Radiological investigations revealed that the child had ambiguous genitalia. On karyotyping the 47, XY karyotype is confirmed. After considering case history both parents were found to be first-degree cousins.

METHODOLOGY**Data Collection**

The common parameters like age and sex were collected. The age at the time of diagnosis of. The presence of any consanguinity was investigated in the parents.

Medical Investigations

The structure of urogenital triangle of child was anatomically examined for confirmation of the equivocal genitalia and unilateral anorchia.

Radiological Investigation

Ultrasonography performed on lower region of abdomen and urogenital triangle of case.

Cytogenetic Investigation

Cytogenetic investigation was carried out on case under study. The case show clinical features similar to ambiguous genitalia. PHA-stimulated peripheral blood leucocytes were cultured for 72 hrs in RPMI-1640 medium supplemented with 20% qualified; heat inactivated fetal bovine serum, 100U/ml penicillin and streptomycine, without mitogene at 37°C.

The culture was exposed to colchicine (10µg/ml) for 30 min followed by hypotonic treatment (0.075M KCl) for 20 min at 37°C. Then fixed in Methanol: Glacial Acetic Acid (3:1) and dropped on wet ice cold grease free slides. The chromosomes were G-banded with trypsin-giemsma banding. Olympus BX51 Research microscope



Fig.1: Photograph showing ambiguous genitalia

DISCUSSION

In the present study we found the unilateral anorchia in the child is might be due to the presence of consanguinity in relation. Similar result obtained by the various researches.

The study of Raja Brauner *et. al.* (2011)^[13] documents several novel features associated with anorchia that have not been described previously. One is premature ovarian failure in two mothers and the absence of primordial follicles in the ovaries of one sister 46,XX after a medical pregnancy interruption for hygroma and anasarque. Two fathers had a tubular deficiency. As mutations involving the *NR5A1* gene have been associated disorders of sexual development (DSD) including anorchia^[14] and primary ovarian insufficiency.^[15] The first American case was described by Fisher (1839).^[16] A man, aged forty-five years, had died of pneumonia. Immediately after birth, the diagnosis of "natural castrate" had been made. He had never had sexual desire. Necropsy revealed a somewhat feminine habitus, with little facial or pubic hair. The penis was the size of that of a ten-year-old boy. The scrotum was small

was used to screen, capture and karyotype the metaphase chromosomes. The results interpreted according to International Standard Chromosome Nomenclature (ISCN).

RESULT

The visual examination of the patient shows that present of only one testis and micropenis but discharge urine through a superfluous opening (Fig. 1). Pelvic ultrasonography and transrectal ultrasonography shows presence of left testes of size 1.2×1.1×0.45 cms while right testes could not be seen in scrotal sac. On karyotyping the patient is confirmed as 46, XY though there is need to observe whether there is any microdeletion on Y chromosome using the PCR based molecular diagnosis. After considering case history both parents were found to be first-degree cousins.

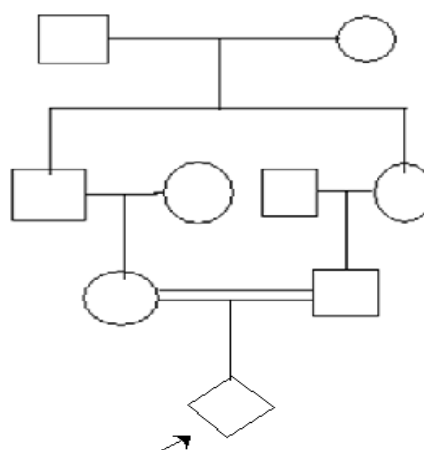


Fig. 2: Pedigree Analysis

and did not contain testes; the tunica dartos, tunica vaginalis communis, and cremaster muscles were normal on both sides. The vas deferens ended on the left side in a small nodule thought to be the epididymis; on the right side, the vas ended in a small, sac-like dilatation. The spermatic arteries and veins were so small they were found with difficulty.

Moorthy *et. al.* (1991)^[17] studied eleven teenage boys with bilateral anorchia and 12 with gonadotrophin deficiency were treated by injections of testosterone ester (enanthate) at an initial dose of 100 mg every six to eight weeks, rising to 250mg every four weeks after three to four years. In the anorchic boys average adult height was 177.1cm, compared with a mean mid-parental height of 174.4cm, and mean predicted adult heights of 177.0cm and 178.0cm. In the patients with gonadotrophin deficiency, mean adult height was 176.9cm, compared with a mean mid-parental height of 176.1cm, and mean predicted adults heights of 174.0cm and 177.3cm. Moorthy *et. Al.*^[17] came on conclusion that the testosterone regimen allows achievement of full growth potential in such.

In our study our findings are similar to various workers but recent Karyotyping technology adds uniqueness in our articles.

CONCLUSION

The 46, XY DSD may be raised as males or females. Treatment involves surgery and hormone replacement therapy. The patients have increased risk for gonadal tumors and may benefit from regular surveillance or surgery to remove abnormally developed gonads. The reconstructive surgery to external genitalia is advisable to be raised as males or females on maturity.

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