

A Case of Intersex Syndrome on Ultrasonography at JIIU's Indian Institute of Medical Science and Research, Badnapur, Jalna, Maharashtra

Abhay Kasliwal^{1*}, Siddiqui M.²

^{1,2}Assutant Professor, Department of Radio-Imaging, JIIU's Indian Institute of Medical Science and Research
Aurangabad Jalna Road, Warudi, Tq. Badnapur, Dist. Jalna-431202, Maharashtra, INDIA.

*Corresponding Address:

abhaykasliwal@yahoo.com

Case Report

Abstract: Intersex Syndrome is the intermingling of characteristics of both sexes in varying degrees in one individual, including physical form, reproductive organs and sexual behaviour. A twenty one year old person brought up as male presented to Dept. of Radio-Imaging, JIIU's Indian Institute of Medical Science and Research, Warudi, Tq. Badnapur Dist. Jalna, Maharashtra with ambiguous external male genitalia. On external genital examination, rudimentary phalluses, small scrotum with impalpable testes were found. It was also found that Uterus and ovary like structure is seen in the pelvis posterior to urinary bladder and at the same time prostate like structure is seen posterior to urinary bladder. Above mention features suggested strong possibility of intersex syndrome. Ambiguous genitalia represents a true medical and social emergency which needs a multi-disciplinary team approach for elucidation.

Keywords: Intersex, Ambiguous genitalia, Ultrasonography.

Introduction

Intersex Syndrome is the intermingling of characteristics of both sexes in varying degrees in one individual, including physical form, reproductive organs and sexual behavior. It results from some defect in the embryonic development. It can be divided into four groups:

1. Pure gonadal agenesis: In this condition the testes or ovaries have never developed. The affected individuals have streak gonads [ovaries or testes].

2. Gonadal dysgenesis: In this condition, the external sexual structures are appropriate to the phenotype, but at puberty the testes or the ovaries fail to develop. Two common syndromes that are well known in this category are:

a) Klinefelter's syndrome: The phenotype is male but the sex chromosome pattern is XXY (47 chromosomes). Features that are seen are gynecomastia, azoospermia, low levels of testosterone, sterility, increased urinary gonodotrophins and increased height. Histology of the gonads shows testicular atrophy with hyalinization of seminiferous tubules.

b) Turner's syndrome: The phenotype is female and the chromosome pattern is XO. This syndrome is characterized by primary amenorrhoea, sterility, lack of development of primary and secondary sexual characteristics, increased urinary gonadotrophin excretion, pigmented naevi, a short fourth metatarsal, webbed neck and wide set nipples.

3. True hermaphroditism: [as is the case here] It is a rare condition of bisexuality characterized by the presence of an ovary and a testicle or two ovotestes in the affected individual. The external genitalia may be of either sex. The uterus may be absent or rudimentary. Phallus may be penile or clitoral. Neither gonad is completely functional.

4. Pseudo-hermaphroditism: In this condition the gonadal tissue of only one sex is seen internally, but external appearance is of the opposite sex.

Abnormalities of the external genitalia sufficient to warrant genetic and endocrine studies is said to occur in 1 in 4,500-10,000 births[1]. Sexual ambiguity is said to occur when the gender of the baby cannot be determined at birth. The gender of an individual is in question because the genitals do not appear clearly as that of male or female. It is traumatizing to the family. It also causes confusion and problems in determining the sex, in which a child would be reared. The abnormality is relatively uncommon. Disorders of sexual differentiation can arise from abnormalities in chromosomes, gonadal development or hormonal production or activity. The orderly development process may also be affected by various environmental factors. Some of these factors may ultimately lead to the development of ambiguous external genitalia.[2]

Case

A twenty one year old person brought up as male presented to Dept. of Radio-Imaging, JIIU's Indian Institute of Medical Science and Research, Warudi, Tq. Badnapur Dist. Jalna, Maharashtra with ambiguous

external male genitalia without any clinical complaints. On physical examination, the person was not having any signs of secondary sexual characteristics of either sex. On external genital examination, rudimentary phalluses, small scrotum with impalpable testes were found. On ultrasonographic examination of pelvis and abdomen, it was found that all abdominal organs were normal. It was also found that Uterus and ovary like structure is seen in the pelvis posterior to urinary bladder and at the same time prostate like structure is seen posterior to urinary bladder. Testes are not appreciated in scrotal sac and sonographically possible sites. Above mention features suggested strong possibility of intersex syndrome. Person was advised to undergo reconstructive surgery for external female organs. Person denied this treatment due to cost and social problems.

Discussion

Intersex states are associated with rare syndromes within a wide clinical spectrum.[3] Ambiguous genitalia represents a true medical and social emergency which needs a multi-disciplinary team approach for elucidation. The paediatric radiologist plays an important role in defining the genital anatomy which remains one of the most important factors in sex determination.[4]

Conclusion

Arguments which led to the ultimate choice of the sex of a child ranged from strict medical, to psychosocial and

even cultural considerations. Expert examination shortly after birth would allow an early and definite decision, avoiding more serious problems with sexual ambiguity in later life.

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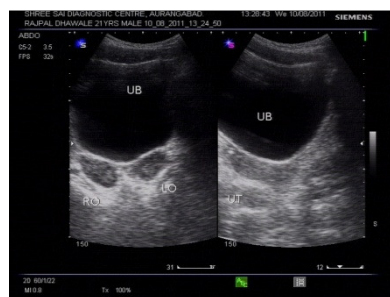


Figure 1: Urinary Bladder (UB), Right Ovary (RO), Left Ovary (LO), Uterus(UT)

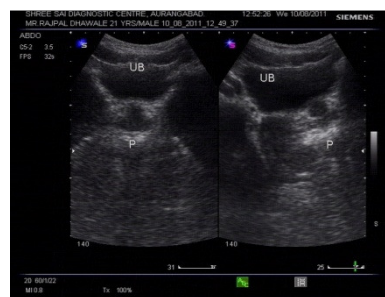


Figure 2: Urinary Bladder (UB) and Prostate (P)



Figure 3: Uterus (UT)



Figure 4: Empty Scrotal Sac