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Case Report

Male Pseudohermaphrodite - A Case Report

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ABSTRACT

A 25 years old lady presented with complain of primary infertility married since 4 yrs. History revealed primary amenorrhea. Patient had not investigated for the primary amenorrhea. She got married 4 yrs ago, no history of difficulty in sexual contacts, and no history of any other systemic complaints. General examination show well developed breasts and presence of bilateral oval shaped swellings in inguinal region. Per speculum exam show evidence of well developed but blind vagina with absence of cervical pit absence of uterus. Karyotype of this patient showed 46-XY normal male chromosomal pattern .The features match with complete androgen insensitivity syndrome. Bilateral orchidectomy was advised for the risk of developing malignancy in the ectopic testes.

Key words: Male pseudohermaphrodite, Pseudohermaphroditism, karyotype, androgen insensitivity, bilateral orchidectomy.

INTRODUCTION

Male pseudohermaphroditism (MPH) is defined broadly as incomplete masculinization of the external genitalia in a male (46XY) karyotype. [1] The gonads of the male pseudo hermaphrodite, when present, are either streak gonads or testes. Complete androgen insensitivity syndrome is a syndrome in which the women often have secondary sexual characters typical of a female due to complete insensitivity to androgens however they are genetically XY and have internal testes, rather than ovaries. [2] They have the same likelihood of genetically XX women of enjoying sexual pleasure but unable to biologically reproduce.

Treatment of male pseudo-hermaphrodites includes the removal of testes i.e. bilateral orchidectomy is needed as the testes are ectopic in position and are in danger of development of malignant changes in it. [3,4]

Surgical correction of ambiguous external genitalia, if present is needed in few cases for cosmetic reasons.

CASE HISTORY

A 25 year old female presented with complain of primary infertility. Menstrual history revealed the history of primary amenorrhea too. Patient not investigated for the same. No any other chief complaint related to systemic disorder. Her sexual history was also normal.

On general examination her height was normal. Breasts, axillary hairs and pubic hairs well developed No other positive

finding on general examination except bilateral inguinal swellings were palpable.

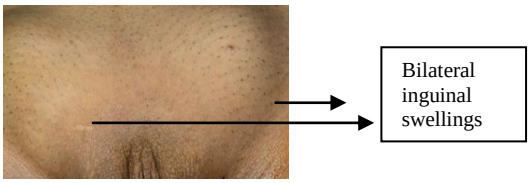

Fig no.1Showing bilateral inguinal swellings

On her per speculum examination there was evidence of blind vagina. There was absence of cervical pit .On per vaginal examination the pelvis was empty indicating absent uterus and cervix.

On ultrasound examination the findings were well correlated with per-speculum and per-vaginal examination that is there was absence of uterus and cervix and presence of inguinal testes and presence of normal vaginal canal. The karyotyping of this patient which showed the presence of 46-XY karyotype

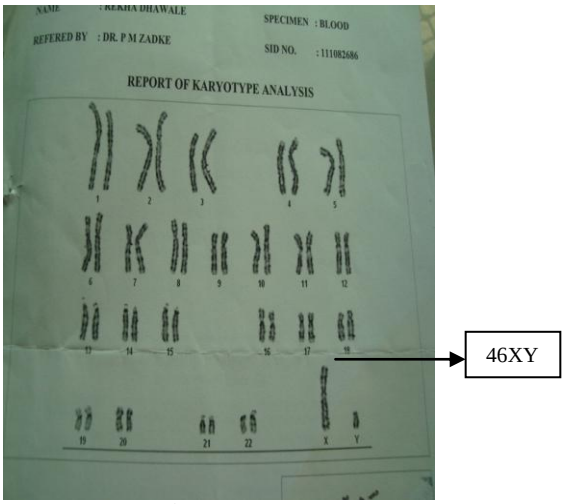

Fig. 2. Karyotype of the patient

These findings showed the male pseudohermaphroditism with normal female external genitalia and internal male gonads.

There was no evidence of any anomaly related with renal system.

This patient was referred to surgery department but patient not followed up afterward for removal of the inguinal testes.

DISCUSSION

In mammals the sexual development depends on successful completion of a series of steps under genetic and hormonal control. This involves three steps

1. Chromosomal sex determined at fertilization.
2. Gonadal sex is under genetic control.
3. Hormones secreted by the fully functional gonads play the role in defining phenotypic sex.

Abnormal sex development could result from sex determination errors i.e. discordance between chromosomal sex and gonadal sex or from discordance between gonadal sex and phenotypic sex i.e. sex differentiation process.

In first case affected individual are referred to as sex reversal whereas in the second case they are called pseudo hermaphrodite. [5]

Male pseudohermaphroditism is the most diverse type of sexual disorder which is defined as incomplete musculanization of the external genitalia in patients with normal male karyotype i.e. 46-XY.

The First cause for MPH is enzyme defect in testosterone synthesis which is very rare cause. [6-8]

The second cause is androgen receptor insensitivity which is of two types

1. CAIS: Complete androgen insensitivity syndrome
2. PAIS: Partial androgen insensitivity syndrome. [9]

In CAIS previously known as testicular feminization syndrome the patient has normal female external genitalia with bilateral inguinal or low abdominal testes.

Whereas in PAIS the patient presents with ambiguous external genitalia. Usually

in both the forms female internal genitalia are absent. [6]

Another common cause for MPH is defects in androgen action due to 5-α-reductase deficiency in which the conversion of testosterone to its most active form dihydrotestosteron is hampered. In these patients present with normal male internal genitalia and wolfian duct derivatives with severe hypospadias. [6,10]

In majority of patients with intersex disorders the defects are apparent at birth but as in our case in spite of male genotype which was a total surprise for the family members, this female phenotype made a female gender the best choice.

As in our case there is presence of XY genotype with normal female external genitalia with male internal gonads which indicates that though there is presence of internal male gonads there might be failure at the receptor site to respond to the sex hormones with subsequent failure to form male phenotype. Thus the features of our case match approximately with CAIS.

In the management of cases of male pseudohermaphroditism with complete female phenotype reconstructive surgery is not required but the male ectopic gonads are need to be removed because of risk of malignancy. [3,4]

ACKNOWLEDGEMENT

I really want to say thanks to my patient who co-operated me a lot.

REFERENCES

1. Migeon CJ.1980 Male pseudoherma-phroditism. Ann Endocrinol;41:311-43.
2. Claude J. Migeon, M.D., Amy B. Wisniewski, Ph.D. and JohnP. Gearhart, M.D. 2001 Syndromes of Abnormal SexDifferentiation page 8
3. Verp M, Simpson J.1987 Abnormal sexual differentiation and neoplasia. Caner Genet Cytogenet :25:191-218.
4. Savage M, Lowe D. 1990Gonadal neoplasia and abnormal sexual differentiation. Clin. Endocrinology : 32:519-33
5. Wilhelm D, Palmer S, Koopman P2007: Sex determination and gonadal development in mammals. Physiological reviews, vol.87 no.1, and page1-28.
6. Ahmed H. Alsalem,Hussain A. Abusrair. 2000. Etiological evaluation, surgical treatment and the incidences in relation to inguinal hernia. Annals of Saudi medicine ,vol.no.20 page 49-52.
7. Berkovitz GD, Lee PA,Brown TR, Migeon CJ1984 etiologic evaluation of male pseuohermaphroditism in infancy and childhood. Am. J Dis Child 138:755-9.
8. Taha SA Magbool GM.1995 The pattern of intersex disorder and gender assignment in the Eastern Province of Saudi Arabia Saudi Med. J. 6:17-22.
9. Wiliams DM, Patterson MN, Hughes IA1993 Androgen insensitivity syndrome. Arch Ds Child : 68:343-
10. Mohamed gad EL- Maula, Hirofimi izaki, Fathy El-Anany2008 Laparoscopy and intersex report of 5 cases of male pseudohermaphroditism. The journal of medicine investigations vol. No. 55 page 147-150.

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