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CASE REPORT

TESTICULAR FEMINISING SYNDROME

Vijay Kumar M. M[1], Prema Prabhudev[2], Kalyani C[3]

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ABSTRACT: Testicular feminization syndrome is a form of pseudohermaphroditism where phenotypic female has male gonads and is genotypically male. Androgen insensitivity syndrome (AIS), also known as testicular feminization, encompasses a wide range of phenotypes that are caused by numerous different mutations in the androgen receptor gene. AIS is an X-linked recessive disorder that is classified as complete, partial based on the phenotypic presentation. The clinical findings include a female type of external genitalia, 46-XY karyotype, absence of Mullerian structures, presence of Wolffian structures to various degrees, and normal to high testosterone and gonadotropin levels. The syndrome is illustrated by a 24-year-old phenotypic female who presented with a primary amenorrhea, female-type external genitalia, an absent uterus and ovaries, and bilateral testes at the level of the internal inguinal ring. Management includes counseling, gonadectomy to prevent primary malignancy in undescended gonad, and hormone replacement. The karyotyping of family members is advocated because of known familial tendencies.
KEYWORDS: Androgen insensitivity syndrome; Testicular feminization syndrome; Gonadectomy.

INTRODUCTION: Intersex is a condition where person has different genotypic and phenotypic sex. In phenotypic female, it is a common cause of primary amenorrhea. Testicular feminization syndrome is a form of male pseudo-hermaphrodite, where phenotypic female has male gonads and is genotypically male. This syndrome also known as androgen insensitivity syndrome was 1st described by Morris at Yale.

The phenotype is a female despite the normal male karyotyping 46 XY. The etiology of this syndrome is congenital insensitivity to androgens transmitted by means of a maternal X-linked recessive gene responsible for androgen intracellular receptors[1]. The incidence of androgen insensitivity syndrome is estimated to be 1:20, 000-64, 000 male births and with variable phenotype expression, this syndrome can present as complete or partial forms.

CASE REPORT: Patient XY, aged 22 years, graduate, from middle socio-economic class, working as school teacher, came with history of primary amenorrhea. She was referred from local doctor and for above complaints she has taken treatment for past 2 years

FAMILY HISTORY: She was last child of the three sisters. Her two elder sisters are married and having two children each.

GENERAL EXAMINATION: Phenotype female, Normal feminine looks, Height 170 cms, Weight 55 kg, Axillary and pubic hairs are sparse, Breasts were well developed (tanner III). Smell and vision were normal. Hair line was low occipital. There was no impairment of visual fields. No thyroid swelling.

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GYNAECOLOGICAL EXAMINATION: Labia majora, minora and clitoris were slightly ill developed. Per speculum examination revealed blind vaginal pouch, good rugosity, cervix not seen. One finger vaginal examination revealed blind vaginal pouch of approximate 6 cm, but cervix and uterus not felt. Per rectal examination confirmed these findings.

Examination of inguinal region revealed bilateral swellings of about 3*4cm, firm, immobile. Her routine investigations were within normal limits. Serum gonadotropin levels (serum FSH, LH) and testosterone level were within normal limits for male. Karyotype was 46 XY. She was counseled about possibility of gonadal tumor and need for the removal. She underwent bilateral gonadectomy and started on low dose estrogen therapy**.**

DISCUSSION: Testicular feminization syndrome may present as complete form (CAIS) and incomplete form (PAIS). In the complete form, there is no androgen response, therefore normal external female genitalia develop and these infants are reared as females. There may be labial or inguinal swellings which contain testis. These patients most often present in late adolescence with primary amenorrhea. There is absence of uterus and ovaries on ultrasound scan or laparoscopy. Vagina is short, develops from urogenital sinus only and ends blindly.

The partial or incomplete form of testicular feminization syndrome is associated with wide range of genital abnormalities and typically present at birth with genital ambiguity. Severe hypospadias associated with micropenis, bifid scrotum and bilateral cryptorchidism are common. In some patients, the external genital phenotype may be predominantly female with partial labial fusion and clitromegaly[2].

Ultrasonography or laparoscopy should be done in all such patients to examine internal genital organs. Measurement of serum 17-hydroxyprogesterone and its sulphate can be done to detect testosterone biosynthetic defects[3]. Management consist of appropriate counseling of parents regarding fertility and long term use of HRT needs to be discussed. Reconstructive surgery to external genitalia is not needed in the complete form but gonads need to be removed due to risk of malignancy. Gonadectomy should be done after puberty to prevent the risk of development of malignancy in the testes. For management of incomplete form both gonadectomy and reconstructive surgery of external genitalia is required[4].

REFERENCES:

1. Adachi M, Takayang R, Tomura A. Androgen insensitivity syndrome as a possible co-activator disease. N Engl J Med 2001; 344(9): 696.
2. Morris JM, Mahesh VB. Further observations on the syndrome," Testicular feminization Syndrome". Am J Obst Gynecol 1963; 15: 731-45.
3. Viner RM, Teoh Y, Williams DM, Patterson MN. Androgen insensitivity syndrome: a survey of diagnostic procedures. Arch Dis Child 1997; 77: 305-09.
4. Manuel M, Katayama KP, Jones HW. The age of occurrence of gonadal tumours in intersex patients with Y Chromosomes. Am J Obst Gynecol 1976; 124: 293.

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FIGURE 1

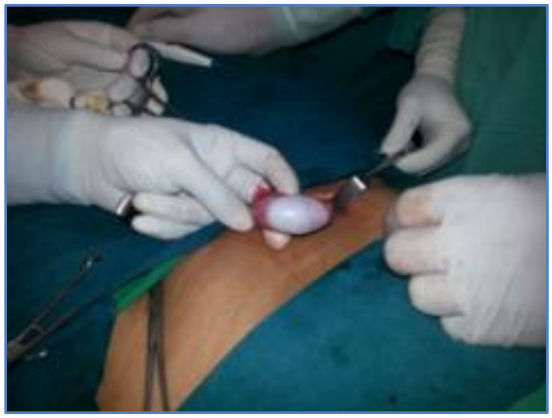

FIGURE 2

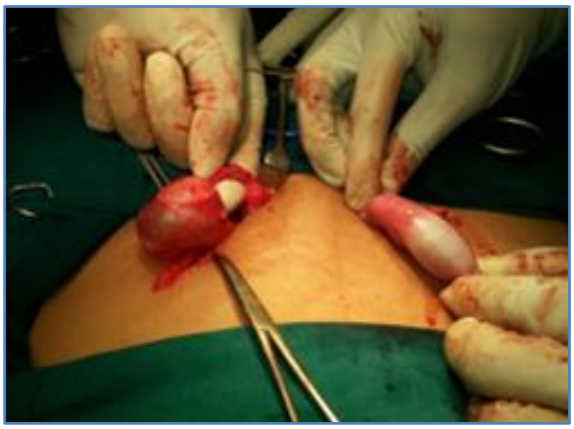

FIGURE 3

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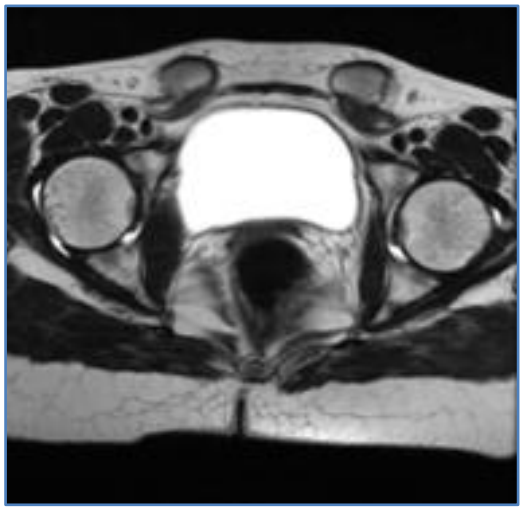

FIGURE 4

AUTHORS:

1. Vijay Kumar M. M.
2. Prema Pradhudev
3. Kalyani C.

PARTICULARS OF CONTRIBUTORS:

1. Associate Professor, Department of Obstetrics and Gynaecology, SSIMS and RC, Davangere.
2. Professor and HOD, Department of Obstetrics and Gynaecology, SSIMS and RC, Davangere.
3. Assistant Professor, Department of Obstetrics and Gynaecology, SSIMS and RC, Davangere.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Vijay Kumar M. M,
569/16, Shivkumar Swamy Nagar,
Ist Stage, IInd Cross, Davangere,
Karnataka.
E-mail: dr.vijayobg@gmail.com

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