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Case Report

Testicular Feminization Syndrome: A Case Study

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ABSTRACT

We report a rare case of testicular feminization syndrome in a 24 years old patient. This is a syndrome due to androgen insensitivity. The patient is phenotypically female with male Karyotype (46XY). The patient is completely feminine with well-developed breasts, female external genitalia, blind vagina, absent mullerian structures, undescended testes and sparse axillary and pubic hair. The gonad (undescended testes) may be intra-abdominal, inguinal or labial. The patient was surgically treated with bilateral orchidectomy and vaginal reconstruction. The incidence of testicular feminization syndrome is reported to range from 1 in 2,000 to 1 in 62,400.

KEYWORDS: Cryptorchidism, Androgen insensitivity syndrome, Orchidectomy, Malignant transformation, X linked recessive

INTRODUCTION

Testicular feminization syndrome is a rare condition where the patient with (46XY) genotype is associated with a female phenotype. This condition can arise (1) if there is a failure of testicular development, as in true gonadal agenesis or leydig cell hyperplasia, (2) if there are enzymatic errors in testosterone biosynthesis, or (3) most commonly, if there is androgen insensitivity at the target organs commonly known as testicular feminization syndrome[1]. In 1970, Lyon and Hawkes[2] reported an X-linked gene for testicular feminization in the mouse. The syndrome is an X-linked recessive condition, the taint being transmitted maternally. Here we report the clinical features diagnosis and treatment for the patient who presented with testicular feminization syndrome.

CASE REPORT

A 24 year old, an apparently female patient presented with on and off hypogastric pain for the past 3-4 years and was not associated with nausea and vomiting. She gave history of primary amenorrhoea. On examination, she had well developed breasts, normal vulva with mildly enlarged phallus and blind vagina. She had very sparse axillary and pubic hair. All the routine investigations were with in normal limits. S.FSH level was raised to post menopausal level of 53.6 /ml. Total testosterone level was done, and found to be 180 ng/dl. MRI pelvis showed B/L undescended hypo plastic testes seen at both inguinal regions. Uterus and ovaries are absent. Histopathological examination of B/L undescended testes after orchidectomy showed testicular tissue with prominent leydig cell hyperplasia. There is no spermatogenesis. Features consistent with those of undescended testes. Chromosomal analysis showed 46XY and revealed a male karyotype with no numerical and structural chromosomal abnormalities in the analyzed cells at the band resolution achieved [Figure 1].

The patient is diagnosed to have bilateral undescended testes present at the inguinal canal and blind vagina with enlarged phallus. The patient was operated and B/L Orchidectomy of undescended testes, labial and vaginal reconstruction[3] and penectomy were done. Both testes were sent for histopathological examination.

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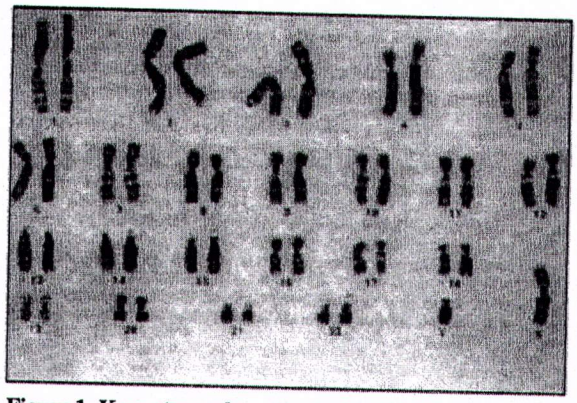

Figure 1: Karyotype of a patient with 46XY karyotupe (1)

DISCUSSION

The testicular function in-utero is normal to the extent of production of Anti Mullerian Hormone (AMH) to which the ducts are sensitive and to steroid hormones. After birth, it is similar to that of any undescended testis. The syndrome results because, with the exception of the urogenital sinus (which may be oversensitive), the target organs of the hormone such as breasts, hair follicles, vocal cords and phallus are inherently insensitive to androgens. The androgen receptors may be completely absent or they may be present in normal numbers but insensitive to androgens, no matter how large a dose of testosterone is administered to them.

The failure of virilization is either (1) complete androgen insensitivity syndrome (CAIS)[4–6]. The patient is an apparent female, with well developed breasts and a normal vulva who presents with primary amenorrhoea. The tubes and uterus are absent but urogenital sinus component of vagina is invariably present. The chromatin pattern is negative and the chromosomal arrangement is 46XY. The gonads are always testes and are found intra–abdominally or in hernial sacs. The plasma level of testosterone and other androgens are above the normal level for the male due to increased leutinizing hormone (LH) production and there is an associated increase in testicular oestradiol. Peripheral conversion of androgens to oestradiol further promotes breast development. (2) Partial androgen insensitivity syndrome (PAIS) or Reifenstein syndrome is the result of partial insensitivity to androgens[7]. These patients have reduced binding affinity of testosterone to the receptors, or there may be a defect in transcription. The mode of inheritance and endocrine profiles are similar to the complete form. Some of these men may have an enlarged phallus with hypospadias and a blind vaginal pouch at birth. There may be cryptorchidism and gynaecomastia. The testes are azoospermic.

The third form of androgen insensitivity is (3) 5-alpha–reductase deficiency[8]. Here, there is failure of conversion of testosterone to dihydrotestosterone (DH) at the target tissues. It is transmitted as an autosomal recessive trait. The presentation and management are as described for partial androgen insensitivity.

CONCLUSION

Testicular feminization syndrome represents well-defined form of made pseudohermophroditism .The patients are at risk of undergoing malignant transformation of the undescended gonad[9]. So we recommend that post pubertal patients should be assessed for possible orchidectomy because of the aggregate risk for testicular malignancy .The androgen levels fall when testes are removed. Management of PAIS depends on the degree of ambiguity of genitalia and the sex of rearing. Some men may respond to high dose administration but it is difficult to predict response. Gonadectomy and hormone replacement therapy is recommended for those assigned the female sex.

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