Testicular Feminization Syndrome

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Abstract

Testicular feminization syndrome is a form of pseudohermaphroditism where phenotypic female has male gonads and is genotypically male. More than 50% cases have an inguinal hernia which was not present in our patient. In addition C.T scan could not localize the presence of testicular tumour. Reporting a rare case of testicular feminization syndrome.

Introduction

Intersex is a condition where person has different genotypic and phenotypic sex. In phenotypic female, it is a common cause of primary amenorrhoea. Testicular feminization syndrome is a form of male pseudo-hermaphrodite, where phenotypic female has male gonads and is genotypically male.

We are reporting a rare case of testicular feminization syndrome.

Case Report

Patient XY, aged 26 years, graduate, from middle socio-economic class, working in bank came with primary amenorrhoea.

She had no symptoms as such, but complained of small breast size.

Family history: She was second of the two sisters. Her elder sister, aged 30 yrs, also had primary amenorrhoea and was diagnosed as testicular feminizing syndrome, but had not taken any treatment. Hence when this girl did not attain menarche even after the age of 20, her parents presumed that she is also a case of same genetic syndrome and did not take any medical opinion.

General examination: Phenotype female, Normal feminine looks, Height 170 cms, Weight 48 Kg, Axillary and pubic hairs absent. Breasts were under developed (Tanner-II). Smell and vision were normal. Hair line was low occipital. There was no impairment of visual fields. No thyroid swelling.

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 5 cm, but cervix and uterus not felt.

Per rectal examination confirmed these findings. Examination of inguinal region did not reveal any swelling.

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. (Fig. 1).

Pelvic sonography and CT scan pelvis showed fibrous band in retroperitoneal area, but did not conclusively diagnose any mass suggestive of testis either in pelvis or in inguinal region. Patient was not willing for diagnostic laparoscopy or laparotomy.

As patient was keen on her breast augmentation, she was started on oestrogen replacement therapy (Premarin -0.375 mg/day). As this therapy did not show desired effect after six months, she was offered breast augmentation surgery in joint consultation with plastic surgeon. Augmentation breast surgery was done by plastic surgeon on 14/2/08. Her post operative course was uneventful and she was very happy with surgery results. She was counselled about possibility of gonadal tumour and was asked to follow-up regularly.

She was restarted on low dose oestrogen therapy.
She is still following up in our OPD.

Discussion

Androgen insensitivity syndrome, formerly known as Testicular Feminization syndrome, is a rare X linked recessive condition. It was described by Morris et al in 1963 therefore, was also known as ‘Morris syndrome’. This is a third most common cause of primary amenorrhoea after gonadal dysgenesis and mullerian agenesis.

The incidence of Testicular Feminization syndrome (TFS) is estimated to be 1:20,000 to 1:64,000 male births and with a variable phenotypic expression. The patient of Testicular Feminization syndrome is a form of ‘Male Pseudohermaphrodite’ i.e. genetic male with a female phenotype because of failure of normal masculinization of external genitalia in chromosomally male individuals. This failure of virilization can be either complete or partial depending on the amount of residual receptor function.

The patients with Testicular Feminization syndrome most often present in late adolescence with primary amenorrhoea. Growth and development are seen to be normal in these patients, although overall height is usually more than average as was evident in our patient. The breasts are usually abnormal with small nipples and pale

Fig. 1: Karyotype of patient showing XY chromosomes.
areolae, which was also seen in our patient. These patients with Testicular Feminization syndrome have absent body hairs specially in pubic and axillary areas. More than 50% of Testicular Feminization syndrome patients have an inguinal hernia, which was not present in our patient. Ultrasonography of inguinal region, CT scan of abdomen pelvis and even MRI are useful in localizing presence of testicular tissue. In our case, CT was not conclusive about its presence and patient refused to undergo laparoscopy or laparotomy.

These patients have underdeveloped labia minora and blind vaginal pouch, which is less deep than normal. Their plasma levels of testosterone are in the normal to high male range and the clearance and metabolism of testosterone are usually normal. But still they do not respond to androgens because of the defect at the intracellular receptor level.² These patients also have high LH levels.

According to some recent studies, the overall incidence of gonadal tumours in patients with Testicular Feminization syndrome is shown to be 5-10%.³⁻⁶ As the gonadal tumours in these patients have not been encountered before puberty, it is suggested that, once full development is attained after puberty, gonads should be removed at approximately 16-18 yrs of age, preferably laparoscopically⁷ and patients should receive hormonal therapy.

In our patient, breasts were hypoplastic and patient was very keen for breast augmentation. As oestrogen replacement therapy did not show desired effect after 6 months, patient opted for breast augmentation surgery with a Silicone Implant. Patient was very happy with the result of the surgery. Other surgical procedures that may be required for these patients are vaginal lengthening procedures and other cosmetic reconstructive surgery.

References

ON-PUMP VERSUS OFF-PUMP CABG
This comparative-effectiveness trial showed that clinical outcomes at 1 year were better with on-pump than with off-pump coronary-artery bypass grafting (CABG), and there was also better graft patency. There were no significant differences in neuropsychological outcomes.