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Case Report

Rare case of complete gonadal dysgenesis 46 XY, Swyer syndrome

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ABSTRACT

Swyer syndrome also known as 46XY complete gonadal dysgenesis is a rare cause of primary amenorrhea. These patients are phenotypically females with female type of internal and external genitalia with absence of testicular tissues. They have streak gonads which have increased potential to turn into malignancy. Bilateral gonadectomy should be done as soon as diagnosis is made. In present case, 20 years unmarried female came with complaints of menstrual bleeding only on taking medication. She never attained menarche, following which she was started on treatment outside, on withdrawal bleeding for the past 3 years. She was now evaluated and diagnosed as Swyer syndrome and bilateral gonadectomy was done laproscopically. Swyer syndrome patients can get married, have normal sexual life and can get pregnant through invitro fertilisation with donor oocyte if desired.

Keywords: Gonadal dysgenesis, Primary amenorrhea, Swyer syndrome

INTRODUCTION

Swyer syndrome or 46XY complete gonadal dysgenesis is a rare disorder, which was first described by Jim Swyer in 1955. The incidence of Swyer syndrome is 1:1,00,000.[1,2] They are genetically 46 XY and phenotypically females. These patients usually present during their adolescence with primary amenorrhea with no development of secondary sexual characters.

CASE REPORT

A 20 years unmarried female came to present hospital with complaints of menstrual bleeding only on taking medication for the past 3 years. She never attained menarche, following which she was started on ayurvedic and hormonal treatment outside, on withdrawal bleeding for the past 3 years. She was not evaluated for the same outside. Her cycles were 4/35-40 days, changes 3 pads/ day. There was no history of cyclical abdomen pain/ head ache/ visual changes/ head trauma/ radiation/ chemotherapy exposure. She had no history of pelvic surgery or chronic illness in the past. On general examination, she was 172 cms tall, weight 80 kg and BMI was 27 kg/m^2. She was well built. Vitals were stable. There was no evidence of acanthosis nigricans, signs of virilisation, cushnoid features, and turner's stigmata. On examination of secondary sexual characters, breast-tanners stage 3, pubic hair-tanners stage 2, axillary hair sparse.

Examination of external genitalia revealed normal female genitalia with no evidence of cliteromegaly and no labio scrotal swelling. Gentle examination of vagina showed patent vagina of 4 cms. Per rectal examination showed small infantile uterus. Serum TSH and prolactin were within normal limits. FSH levels was 94 mIU/ml and LH was 57 mIU/ml. Estrogen was 51 pg/ml, testosterone was 37.7 ng/dl and AMH 0.2ng/ml. Ultrasound showed hypoplastic uterus, bilateral ill-defined adnexa and no renal abnormality. Karyotyping was done which showed 46XY. Diagnostic laparoscopy showed infantile uterus,

normal fallopian tubes, streak gonads. Patient was diagnosed as Swyer syndrome. Cyclical hormonal therapy was continued and patient and attenders were counselled regarding bilateral gonadectomy and marriage and pregnancy in future if desired through invitro fertilisation with donor oocyte. Laproscopic gonadectomy was done for the patient after 2 months.

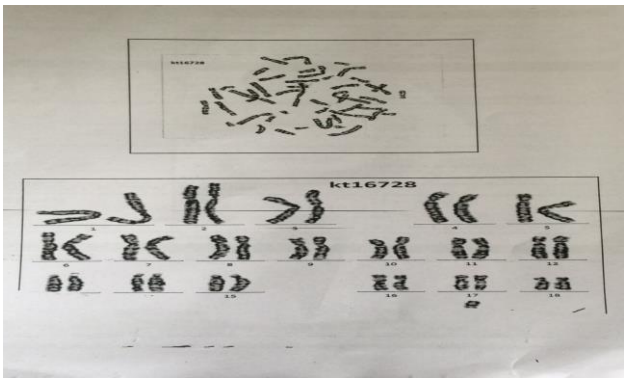

Figure 1: G-banded karyotype showing 46, XY.

DISCUSSION

In Swyer syndrome, irrespective of having 46 XY karyotype, these patients fail to develop testes as there is mutation of SRY gene. SRY gene (Sex determining region Y) expression is responsible for male sexual differentiation which leads to testicular development. When SRY gene is mutated the bipotential gonads develop into ovary and there is no development of testes. The streak gonads don't produce androgen or AMH. As testosterone is absent wollfian duct fails to develop internal genitalia. As AMH is absent, the mullerian ducts develop into uterus, fallopian tube, cervix and vagina. Uterus is usually present and is hypoplastic. In Swyer syndrome patients are genotypically males, phenotypically females, with female type of internal and external genital organs with no ambiguous genitalia.

In utero these ovaries may contain ova but these rapidly degenerate after birth and hence present with primary amenorrhea in adolescence. Streak gonads fail to function so they fail to develop secondary sexual characters. But when these patients are treated with combined estrogen and progesterone they develop secondary sexual characters and withdrawal bleeding. Due to non-functioning gonads there is elevated level of gonadotropins indicating the normal response of pituitary, low level of estrogen, and normal female level of androgen.[3]

10-15% of Swyer syndrome is due to mutation of SRY gene.[4] Most cases the cause is not identified or may be due to mutation of other genes in the sex differentiation pathway such as the autosomal genes SOX9 & WT1 and DAX1 gene on the X chromosome.[5] These patients have streak gonads which has increased chance of developing malignancy. Most common are gonadoblastomas and malignant germ cell tumours. Gonadoblastomas are seen in 20-30% of women with Swyer syndrome.[1] In Swyer syndrome bilateral gonadectomy should be done as soon as the diagnosis is made as there is increased chance of malignancy, while androgen insentivity syndrome warrants removal of both the gonads only after puberty is completed as pubertal development proceed more smoothly in response to endogenous hormone production and the risk of developing tumour is only 5-10%.[6]

Swyer syndrome is rare type of the gonadal dysgenesis when compared with Turner syndrome and androgen insensitivity syndrome. Incidence of Swyer syndrome is 1:1,00,000, incidence of Tuner syndrome is 1:2000 and incidence of androgen sensitivity syndrome is 1: 20,000 – 64,0007-10.

CONCLUSION

Swyer syndrome is a very rare cause of primary amenorrhea with increased risk of malignancy. Early diagnosis and early surgery is recommended as there is increased chance of malignancy. Absence of gonads will lead to infertility. These patients can get married, have normal sexual life and can get pregnant through in-vitro fertilization with donor oocytes if desired.

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