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Case Report

Swyer syndrome presenting as primary infertility

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ABSTRACT

Swyer syndrome was first described by Jim Swyer in 1955. It is a form of "Pure gonadal dysgenesis". The affected female has 46, XY karyotype. A 21 year old married female came with complaints of primary infertility. On examination she has normal built with normal secondary sexual characteristics. She had normal vaginal opening with small uterus. Serum FSH was 71.54 mIU/ml. Thyroid and Prolactin was in normal range. Karyotype showed genotype of 46, XY. Diagnostic laparoscopy showed streak gonads, small uterus, and normal patent fallopian tubes. Diagnosis of Swyer syndrome was made.

Keywords: Swyer syndrome, Primary infertility

INTRODUCTION

Swyer syndrome was first described by Jim Swyer in 1955. It is a rare entity. The incidence of Swyer syndrome is 1:100000.[1,2] Affected individuals have XY karyotype but external and internal genialia are of the female type. The gonads are usually replaced by fibrous streaks. Patients usually present in adolescence with primary amenorrhea and lack of secondary sexual characters.

CASE REPORT

A 21 year old female, married for 3 years came to the gynaecology OPD with C/o inability to conceive. She never attained menarche. Patient took Ayurvedic and hormonal treatment from outside and on withdrawal only she had menses at age of 18 years. She had her last menses on 20/3/13. She had bleeding for a period of 4-5 days/decreased flow and with no pain. There was no h/o cyclical pain, radiation exposure, headache, visual disturbances. There was no past significant surgical history. No H/o any chronic illness.

On general examination, she was 167 cm tall, weight 60 kg and BMI was 21.5 kg/m^2. She had average built. There was no evidence of acanthosis nigricans, acne, hirsutism, goiter, cushnoid features. On examination of secondary sexual characters, breast-tanner stage 4, pubic hair present, axillary hair sparse. Her SMR was A3B5P4. Vitals were stable. Examination of external genitalia revealed a normal female genitalia. There was no evidence of clitoromegaly and no labio scrotal swelling. Vagina was well formed. Per speculum examination showed small cervix. Per vaginal examination showed that uterus was small in size and no adnexal mass.

Serum TSH and prolactin were within normal limits. FSH levels were done which was 71.54 mIU/ml and LH was 21.45 mIU/ml. Estrogen was 34 pg/ml and progesterone was 0.38 ng/ml. Ultrasound showed a hypoplastic uterus with ET 3.3 mm and B/L ill-defined adnexa and no renal abnormality was detected. Karyotype was done which showed 46XY.

Diagnostic laparoscopy was done which showed a small uterus. Left gonad was aplasia (Figure 1). On right side, small band of white tissue seen, suggestive of streak gonad. B/L tubes seen, fimbrial end slightly buried in

pelvis. There were no signs of tuberculosis or endometriosis. B/L chromopertubation test was positive (Figure 2). In view of streak gonads and genotype XY, diagnosis of Swyer syndrome was made.

Cyclical hormonal therapy was continued and patient and attendants were counselled for pregnancy by donor oocyte. But patient lost to follow up.

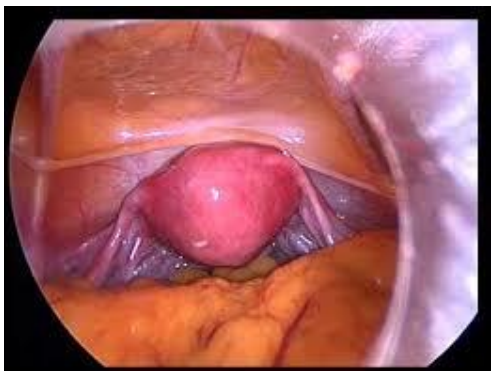

Figure 1: Diagnostic laparoscopy showing small uterus.

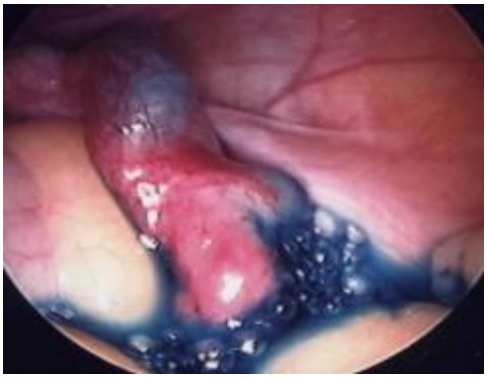

Figure 2: Positive chromopertubation test.

DISCUSSION

The Swyer syndrome, 46, XY gonadal dysgenesis, belongs to the category of sexual abnormality.[3] The syndrome is complete/pure gonadal dysgenesis. The patients with 46, XY gonadal dysgenesis are diagnosed in early adolescence with delayed pubertal development. The patients' mesonephric ducts (Wolffian ducts) are in atrophy, paramesonephric ducts (Müllerian ducts) develops to uterus, fallopian tubes and part of the vagina as a result of lacking testosterone and inhibitor of Müllerian ducts. As expected they show elevated gonadotropins, normal female levels of androgens, low levels of estrogens, female external genitalia, uterus and fallopian tubes. The patients can have normal sexual intercourse.

Minimal breast enlargement reflects peripheral aromatization of androgens. Both gonads display fibrous tissue that vaguely resembles ovarian stroma but no follicles. The etiology of 46, XY pure gonad dysgenesis is thought to be a short arm Y chromosome deletion involving SRY (putative testicular-determining factor gene), a mutation in other genes that leads to inhibition of SRY function or mutation of SRY function.[4]

Nuclear import can be disrupted by changes within the NLS (nuclear localization signals) motif itself. Clearly, if the import receptor does not recognize the NLS-cargo, then that cargo will remain in the cytoplasm, which could be deleterious if the nuclear role is critical for proper cell function. In Swyer syndrome, there is loss of nuclear localization of a key developmental protein which has been linked to developmental defects include male-to-female sex reversal. Mammalian gender is determined by the presence or absence of a dominant gene located on the Y chromosome called SRY (sex-determining region of the Y chromosome). SRY is one of many transcription factors required during early development for proper testicular formation in XY males. Mutations in SRY result in male-to-female sex reversal also known as Swyer syndrome.

In patients with gonadal dysgenesis, either "pure"(with a 46, XX or 46, XY karyotype) or associated with the somatic features of Turner's syndrome (with a 45, XO karyotype), both gonads are represented by a streak of fibrous tissue that vaguely resembles ovarian stroma.[5,6] A bilateral gonadectomy should be done especially by laparoscopy when a Swyer syndrome is discovered in order to avoid the risk of malignant transformation. There is possibility of pregnancy by oocyte donation if the uterus was not removed for a malignant etiology.

In our case, the patient did not have a bilateral gonadectomy after knowing she has a 46, XY karyotype because she wanted to be pregnant and did not believe she had the risk of suffering from malignancy of ovary. It is necessary to take the familial screening of Swyer syndrome cases. As a malignant germ cell tumor of the ovary, dysgerminoma can be found either in a pure form or mixed with other germinal elements. Therefore in premenarchal patients with a pelvic mass, the karyotype should be determined. The incidence of gonadoblastoma in a dysgenetic gonad is 25%.[7]

Swyer syndrome is an example of a condition in which an externally unambiguous female body carries dysgenetic, atypical, or abnormal gonads. The differential diagnosis could be mixed gonadal dysgenesis where one gonad is a fibrous streak and another is testes with karyotype 46, XY. Other examples include complete androgen insensitivity syndrome, partial X chromosome deletions, lipoid congenital adrenal hyperplasia, and Turner syndrome.

CONCLUSION

In addition to testicular feminization syndrome and true hermaphrodite, Swyer syndrome must be kept in mind in differential diagnosis of phenotypically female patients

with primary amenorrhea, tall eunuchoid stature and 46, XY karyotype, although it is a rare entity as is clear from the incidence. Such patients are given HRT for development of breast and to prevent osteoporosis, however conception can be done by artificial reproductive techniques and oocyte donation. Gonadectomy should be done as there are high chances of malignancy.

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REFERENCES

1. Coutin AS, Hamy A, Fondevilla M, Savigny B, Paineau J, Visset J. Pure 46, XY gonadal dysgenesis. J Gynecol Obstet Biol Reprod (Paris). 1996;25(8):792-6.
2. Behtash N, Karimi Zarchi M. Dysgerminoma in three patients with Swyer syndrome. World J Surg Oncol. 2007;5:71.
3. Guidozzi F, Ball J, Spurdle A. 46, XY pure gonadal dysgenesis (Swyer-James syndrome)--Y or Y not a review. Obstet Gynecol Surv. 1994;49(2):138-46.
4. Behzadian MA, Tho SP, McDonough PG. The presence of the testicular determining sequence, SRY, in 46, XY females with gonadal dysgenesis (Swyer syndrome). Am J Obstet Gynecol. 1991;165(6 Pt 1):1887-90.
5. Sohval AR. The syndrome of pure gonadal dysgenesis. Am J Med. 1965;38:615-25.
6. Jones HW Jr, Ferguson-Smith MA, Heller RH. The pathology and cytogenetics of gonadal agenesis. Am J Obstet Gynecol. 1963;87:578-600.
7. Speroff L, Glass RH, Kase NG. Swyer syndrome. In: Speroff L, Glass RH, Kase NG, eds. Clinical Gynaecologic Endocrinology and Infertility. 6th ed. Baltimore: Lippincott, Williams and Wilkins; 1999: 367-368.

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