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Case Report

A rare case of Swyer's syndrome

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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 46XY karyotype. A 17 year old unmarried girl came with complaints of primary amenorrhea, non development of breast. On examination she has normal built. Examination of secondary sexual characters revealed no breast development, pubic and axillary hairs were sparse. Female type of genitalia with vaginal opening. Serum FSH was 117.5 mIU/ml. Thyroid and Prolactin was in normal range. Karyotype showed genotype of 46XY. Diagnostic laparoscopy showed streak gonads, small uterus, and normal fallopian tubes. Diagnosis of Swyer syndrome was made. Patient was started on hormonal replacement therapy (HRT). In Swyer syndrome there is a mutation of SRY gene leads failure of development of testis. Mullerian duct development takes place by default. These patients can have normal sexual intercourse and can become pregnant by donor oocyte. They need to be on HRT. The risk of gonadoblastoma and dysgerminoma are very high in streak gonads so bilateral gonadectomy is advised.

Keywords: Swyer's syndrome, Primary amenorrhea

INTRODUCTION

Swyer's syndrome was first described by Jim Swyer in 1955. Affected individuals have an XY karyotype but external and internal genitalia 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. Swyer's syndrome is a rare entity incidence being 1:30,000.

Purpose of reporting this case is its rarity and importance of diagnosis of XY female as there is high incidence of gonad malignancy, in affected person.

CASE REPORT

A 17 year old girl came to OPD with her mother with complaints of primary amenorrhea and no development of breast.

There was no history of cyclical abdominal pain, hormonal intake, radiation exposure, chemotherapy or any central nervous symptoms such as headache or visual disturbances. There was no past significant surgical history. There was neither history of childhood tuberculosis nor history of Koch's in the family.

She was the first child of a non-consanguineous marriage. She has one younger sister of 15 yrs old who has got her menarche at age of 14 yrs.

On general examination she was 143 cm. tall and weighs 39 kg. Body mass index was (BMI) 18 kg/m². There was no evidence of acanthosis nigricans, acne, hirsutism, goiter, cushingoid features of Turner's stigmata, like webbed neck, cubitus valgus. Examination of secondary sexual characteristics revealed no breast development with hypopigmented areola. Pubic and axillary hair were sparse.

Examination of external genitalia revealed that they were of female type and there was no evidence of clitoromegaly. Vaginal opening was seen. After investigation, serum follicle stimulating hormone (FSH) was found to be very high -117.5 mIU/ ml.

Serum TSH and serum prolactin were normal. Ultrasound showed a rudimentary uterus and bilaterally ill-defined adnexa, no renal abnormality was detected.

A karyotype was done which showed a genotype of 46XY (Figure 1).

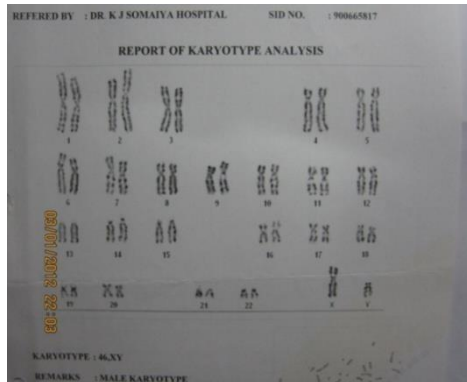


Figure 1: Karyotype report showing a genotype of 46XY.

Patient was subjected for a diagnostic laparoscopy which showed a small uterus and bilateral normal fallopian tubes. Ovaries could not be visualized; fibrous bands were seen on either side. No any other pelvic abnormality seen.

In view of streak gonads and genotype of XY diagnosis of Swyer syndrome was made.

Counseling of parents done. Patient was started on hormone replacement therapy (HRT) with conjugated estrogen 1.25 mg per day and cyclical medroxy progesterone acetate.

Three months after initiation of HRT patients attained menarche with one episode of spotting per vaginum. The secondary sexual characteristics have shown improvement. Breast development occurred Tanner stage II.

DISCUSSION

Swyer syndrome is a form of pure gonadal dysgenesis.¹

The first known step of sexual differentiation of a normal XY fetus is the development of testes. The early stages of testicular formation in the second month of gestation requires the action of several genes, most important is SRY, the sex determining region of the Y chromosome.⁴

Mutations of SRY accounts for many cases of Swyer syndrome. When such a gene is defective the indifferent

gonads fail to differentiate into testes in an XY (genetically male) fetus. Without testes, no testosterone or antimullerian hormone (AMH) is produced. Without testosterone the external genitalia fail to virilize,² resulting in normal female genitalia. Without AMH the mullerian duct develops into normal internal female organs i.e. uterus, fallopian tubes, cervix and vagina.¹

A baby who is born externally and is normal in all anatomic aspects except that the child has nonfunctional streak gonads. Because of the inability of the streak gonads to produce sex hormones (both estrogens and androgens) most of the secondary sex characters do not develop. Therefore, there is absence of breast development, widening of the pelvis and hips and menstrual periods. Adrenal gland is not affected and can produce androgens and most of these persons will develop pubic hair, though it often remains sparse.

We made a diagnosis of Swyer's syndrome because patient was a normal statured girl with primary amenorrhea with clinical features of sexual infantilism whose genotype was pure XY and the gonadal tissues are fibrous band (Figure 2).¹



Figure 2: Laparoscopic view of streak gonads.

The main differential diagnosis of sawyer's syndrome is mixed gonadal dysgenesis which is more frequently seen than the former. In this condition the gonads on histopathology will also show testicular differentiation in addition to ovarian differentiation.⁵ The genotype too is usually a mosaic pattern. The incidence of malignant gonadoblastoma in patients with dysgenetic gonads is high 25-35% and gonadectomy done for this patient.^{3,6}

These patients can have a normal sexual intercourse and they need HRT for development of breast and to prevent osteoporosis.

They can conceive using donor oocytes and artificial reproductive techniques.

CONCLUSION

Swyer syndrome is extremely rare case of primary amenorrhea. Genetic testing plays very crucial role in diagnosis of Swyer syndrome. In patients of Y chromosome there is high incidence of gonadoblastoma and dysgerminoma so gonadectomy is must.

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