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CASE REPORT OPEN ACCESS

Nature's Error…In Mirror He or She…Swyers Syndrome - A Rare Case

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ABSTRACT

Swyer syndrome, also known as 46, XY complete or pure gonadal dysgenesis, is a rare disorder of sex development (DSD) characterized by a female phenotype despite a male (46, XY) karyotype. The Affected individuals typically present during adolescence with delayed puberty and primary amenorrhea. Early diagnosis is essential to prevent the development of gonadal malignancies, which are common in dysgenetic gonads. Timely initiation of hormone replacement therapy (HRT) plays a crucial role in promoting the development of secondary sexual characteristics and supporting overall physical and psychosocial well-being.
This case report highlights the importance of prompt recognition and management of Swyer syndrome to ensure optimal long-term outcomes.

Keywords: Swyer syndrome, primary amenorrhea, puberty, Gonadoblastoma

INTRODUCTION

Swyer syndrome, also called 46,XY complete gonadal dysgenesis, is a rare disorder of sex development (DSD) in which individuals have a typical female external appearance but possess a male (46,XY) karyotype.

- Despite having XY chromosomes, their gonads fail to develop properly and instead form nonfunctional "streak" gonads composed mainly of fibrous tissue.
- As a result, individuals with Swyer syndrome do not produce the sex hormones which are necessary for puberty or sexual development.
- Clinically, patients usually present during adolescence with delayed puberty, primary amenorrhea (absence of menstruation) and lack of secondary sexual characteristics such as breast development.
- Diagnosis is confirmed through genetic testing and hormonal studies.
- Management involves hormone replacement therapy to induce secondary sexual characteristics and maintain bone health, and surgical removal of the streak gonads due to the significant risk of gonadal tumors, particularly gonadoblastoma.

CASE REPORT

An 18-year-old female, Miss X, presented with complaints of primary amenorrhea. She was moderately nourished and moderately built, with a height of 165 cm, weight of 51 kg, and BMI of 18.8 kg/m². There was no pallor, icterus, cyanosis, lymphadenopathy, or edema. Examination of the head, face, eyes, ears, nose, mouth, and pharynx was normal.

Secondary sexual characteristics were underdeveloped, with sparse axillary and pubic hair corresponding to Tanner stage II. Breast development was also Tanner stage II. Abdominal examination was unremarkable, with no palpable masses or tenderness.

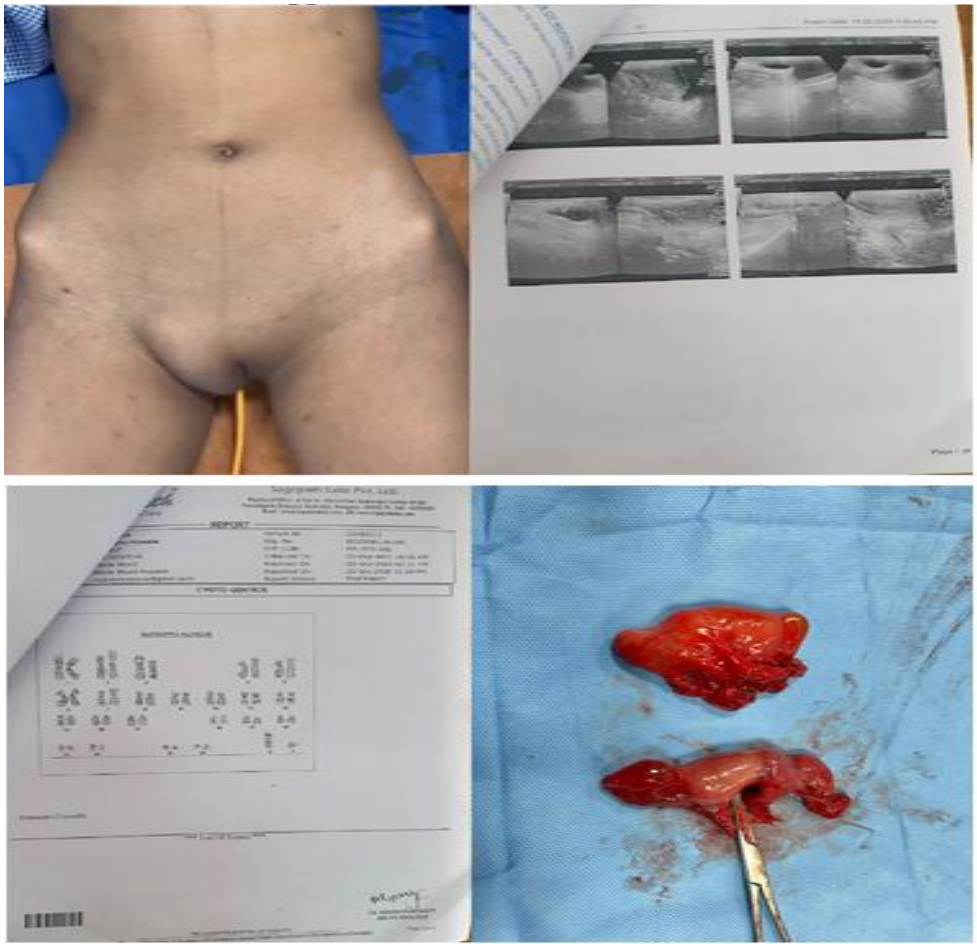
Local genital examination revealed female-type external
genitalia with a normal vulva and hymen. Sparse pubic hair was noted. A palpable mass approximately 2 × 3 cm in size was detected in the right labia majora.

Investigations:
Pelvic ultrasound revealed uterine agenesis and bilateral inguinal hernia with a diffuse hypoechoic mass in the pubic region.

Hormonal profile:
FSH: 16.77 mIU/mL (elevated) LH:
25.40 mIU/mL (elevated) TSH:
1.59 µIU/mL (normal)
Karyotype analysis showed a 46,XY chromosomal pattern.

Based on clinical findings, hormonal profile, imaging, and karyotyping, a diagnosis of Swyer syndrome (46,XY complete gonadal dysgenesis) was made.

The patient underwent prophylactic bilateral gonadectomy to eliminate the risk of gonadal malignancy, particularly gonadoblastoma. Plans for hormone replacement therapy (HRT) were initiated to induce and maintain secondary sexual characteristics and support bone health.



DISCUSSION
Swyer syndrome, also known as 46,XY complete gonadal dysgenesis, is a rare disorder of sex development in which individuals possess a male karyotype (46,XY) but develop as phenotypic females. The condition results from mutations

affecting genes critical for testicular differentiation, such as the SRY gene on the Y chromosome or other downstream genes like SOX9, DAX1, and WT1. Failure of testicular development leads to the formation of nonfunctional fibrous streak gonads that produce neither testosterone nor anti-Müllerian hormone (AMH).

Patients typically present in adolescence with primary amenorrhea and delayed puberty. Despite having female external genitalia, they exhibit underdeveloped secondary sexual characteristics due to estrogen deficiency. In this patient, sparse pubic and axillary hair and Tanner stage II breast development reflected incomplete feminization.

Absence of AMH during development allows Müllerian structures initially to form, but subsequent degeneration or agenesis often results in absent or rudimentary uterus and upper vagina, as seen in this case' s ultrasound findings of uterine agenesis. Elevated gonadotropins (FSH and LH) reflect hypergonadotropic hypogonadism, consistent with nonfunctioning gonads. Karyotyping is critical for diagnosis, revealing a 46,XY chromosomal pattern.

A crucial aspect of Swyer syndrome is the increased risk of gonadal malignancy, particularly gonadoblastoma and dysgerminoma, with reported risks ranging from 15% to 30%. This risk mandates early prophylactic gonadectomy once the diagnosis is established, as was appropriately performed in this patient.

Management includes surgical removal of the dysgenetic gonads and lifelong hormone replacement therapy (HRT) to induce and maintain secondary sexual characteristics, support bone health, and ensure cardiovascular protection. Psychological support and counseling are also essential due to potential psychosocial stress related to diagnosis, fertility implications, and gender identity concerns.

Early recognition of Swyer syndrome enables timely intervention to prevent malignancy and optimize physical and emotional health outcomes.

CONCLUSION

Swyer syndrome should be considered in any adolescent with primary amenorrhea and delayed puberty. Early diagnosis through clinical evaluation, hormonal studies, imaging, and karyotyping is essential. Timely gonadectomy and hormone replacement therapy are critical to prevent malignancy and promote normal secondary sexual development and long-term health.

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