Setgene Lab
PATHOLOGY • GENETICS • MICROBIOLOGY

Title: Clinical and Diagnostic Challenges of Swyer Syndrome: A Dual Case Study

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Introduction

Swyer Syndrome, also known as 46, XY gonadal dysgenesis, is a rare genetic condition that results in a female phenotype with a male genotype, implying that individuals have one X chromosome and one Y chromosome in each cell, as is typical in male; however, they have female reproductive structures. This condition is commonly linked with primary amenorrhea and infertility, which presents considerable diagnostic and treatment issues. Typically identified during adolescence, the syndrome has a frequency of 1 in 80,000. Affected individuals have female external genitalia, and some have female internal reproductive systems such as the uterus and fallopian tubes, but their gonads are dysfunctional. Babies with Swyer syndrome are typically treated as female based on their physical appearance until they reach adolescence. Because they lack functional ovaries that produce hormones, affected people frequently undergo hormone replacement therapy in early adolescence to initiate puberty, which causes the breasts and uterus to expand and eventually leads to menstruation. In this poster, we discuss two cases with Swyer syndrome, emphasizing the clinical presentation, diagnostic findings, and the significance of genetic counseling.

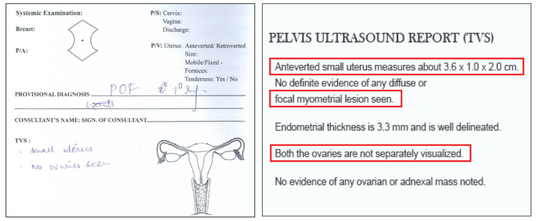
Aim & Objective

Objective: To investigate and compare the clinical and genetic characteristics of two patients with Swyer syndrome.
Aim: To enhance understanding of Swyer syndrome for better diagnosis, management, and genetic counseling of affected individuals and their families.

Materials & Methods

Clinical Data:

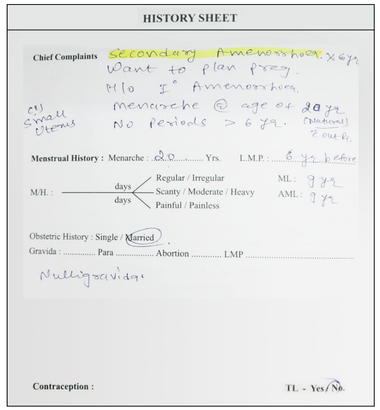
Case 1: A 39-year-old female, married for 8 years, presented with primary infertility and menstruation only induced by medication. Ultrasonography revealed a small uterus and absent ovaries.


Systemic Examination:
Breast:
P/A:
P/S: Cervix:
Vagina:
Discharge:
P/V: Uterus: Anteverted/ Retroverted
Size:
Mobile/Fixed -
Fornices:
Tenderness: Yes / No
PROVISIONAL DIAGNOSIS ... POF @ 19 y/o
CONSULTANT'S NAME: SIGN. OF CONSULTANT.....
TVS :
- Small uterus
- No ovaries seen

PELVIS ULTRASOUND REPORT (TVS)
Anteverted small uterus measures about 3.6 x 1.0 x 2.0 cm.
No definite evidence of any diffuse or
focal myometrial lesion seen.
Endometrial thickness is 3.3 mm and is well delineated.
Both the ovaries are not separately visualized.
No evidence of any ovarian or adnexal mass noted.

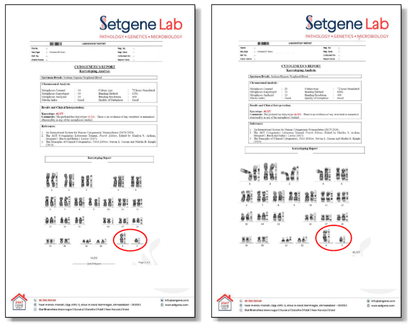
Case 2: A 27-year-old female, married for 9 years, had a history of primary amenorrhea with menarche at age 20, followed by six years of secondary amenorrhea with primary infertility. Imaging showed absent ovaries.

Based on the details, karyotyping was suggested to the patients. Karyotyping involved culturing cells from blood, arresting them in metaphase using colcemid, and then treating them with a hypotonic solution. Fixed cells are stained with Giemsa, and chromosomes are analyzed under a microscope to identify genetic abnormalities. Hormonal analysis was indicative of hypergonadotropic hypogonadism.


HISTORY SHEET
Chief Complaints: secondary Amenorrhoea X 6 yr. Want to plan preg. H/o I° Amenorrhea, menarche @ age of 20 yr. No periods > 6 yr. (Medical) Small uterus
Menstrual History : Menarche : 20 Yrs. L.M.P. : 6 yr. before
M/H. : days / days — Regular / Irregular; Scanty / Moderate / Heavy; Painful / Painless
ML : 9 yr
AML : 9 yr
Obstetric History : Single / Married
Gravida : ... Para ... Abortion ... LMP ...
Nulligravida
Contraception :
TL - Yes / No

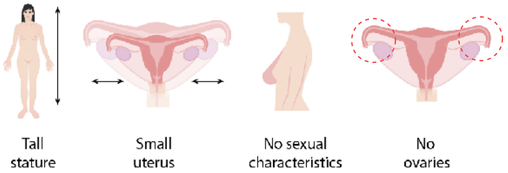
Results

Both the cases showed karyotype 46, XY. The genotype and phenotype both were consistent with the diagnosis of Sywer syndrome.



Discussion/ Conclusion

Swyer first reported two cases of sex reversal in 1955. These cases were distinct from the other known cases of what was then referred to as "male pseudohermaphroditism." The etiology of syndrome is unknown, but mutations in one of several genes have been linked to the disorder in certain cases.Variants in the SRY gene have been identified in 15–20% of Swyer syndrome patients. The SRY gene on the Y chromosome encodes a protein known as sex-determining region Y. The sex-determining region Y protein initiates activities involved in male-typical sex development. Swyer syndrome is caused by SRY gene mutations that prevent the sex-determining region Y protein from being produced or produce a nonfunctioning protein. A significant incidence of gonadoblastoma and germ cell cancers has been reported; hence the current practice recommends a gonadectomy following the diagnosis. Swyer syndrome is rarely inherited, and most occurrences occur in people who have no family history of the disorder. These situations are frequently caused by de novo variations in a gene that arise during the production of reproductive cells (eggs or sperm) or early embryonic development.



Swyer syndrome must be diagnosed early due to the significant risk of dysgerminoma developing at such a young age. Genetic counseling played a pivotal role in managing these cases. It provided patients with information about their condition, the importance of regular follow-up, and the implications of their diagnosis for their reproductive health. Counseling also addressed psychological and emotional support, ensuring a holistic approach to patient care. These case studies underscore the importance of a multidisciplinary approach in diagnosing and managing Swyer Syndrome, aiming to optimize clinical outcomes and improve the quality of life for affected individuals.

References

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