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# Swyer Syndrome in 18-Year-Old: A Rare Clinical Entity Case Report

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**Abstract:**

*Swyer syndrome, a rare disease of gonadal dysgenesis is associated with 46XY karyotyping. A18-year-old patient presented with complaints of primary amenorrhea and absence of breast development. Ultrasound revealed hypoplastic uterus with lack of endometrium-myometrium differentiation. Both the gonads were streaky. Following laparoscopic gonadectomy & hormone replacement therapy follow up ultrasound showed increased uterine size.*

***Key words:*** *Swyer Syndrome, ultrasonography*

## Introduction:

Swyer syndrome is caused by abnormal sex differentiation during the embryonic period, resulting pure gonadal dysgenesis associated with 46XY karyotype. It is characterized by bilateral streak gonads, normally developed Mullerian structures, female appearing external genitalia, primary amenorrhea, lack of secondary sexual characteristics and hypergonadotropic hypogonadism.[1]

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## Case Report:

- An 18-year-old patient was referred to Radiology department for sonographic evaluation who had complaints of primary amenorrhea and absence of breast development.
- On physical examination patient had a height of 162 cm, weight of 49 kg, BMI of 19.08Kg/$m^2$, phenotypically female with masculine body built.
- Breast development tanner stage I, absent axillary hair, female pattern pubic hair-tanner stage II with female external genitalia and intact vaginal orifice.
- Her endocrinological assessment revealed.

| | | |
|---|---|---|
| Follicle stimulating hormone | 116.16 | mlU/ml |
| Luteinizing hormone | 28.96 | mlU/ml |
| Estradiol | 10 | pgm/ml |
| Prolactin | 240.74 | mlU/ml |
| Free T4 | 13.48 | pmol/ml |
| ACTH | 14.40 | pgm/ml |
| Cortisol | 190 | nmol/L |
| 17-OH progesterone | 2.19 | ng/ml |
| Testosterone | 0.08 | ng/ml |
| Anti-Mullerian hormone | 0.02 | ng/ml |
| Serum thyroid stimulating hormone | Normal | |

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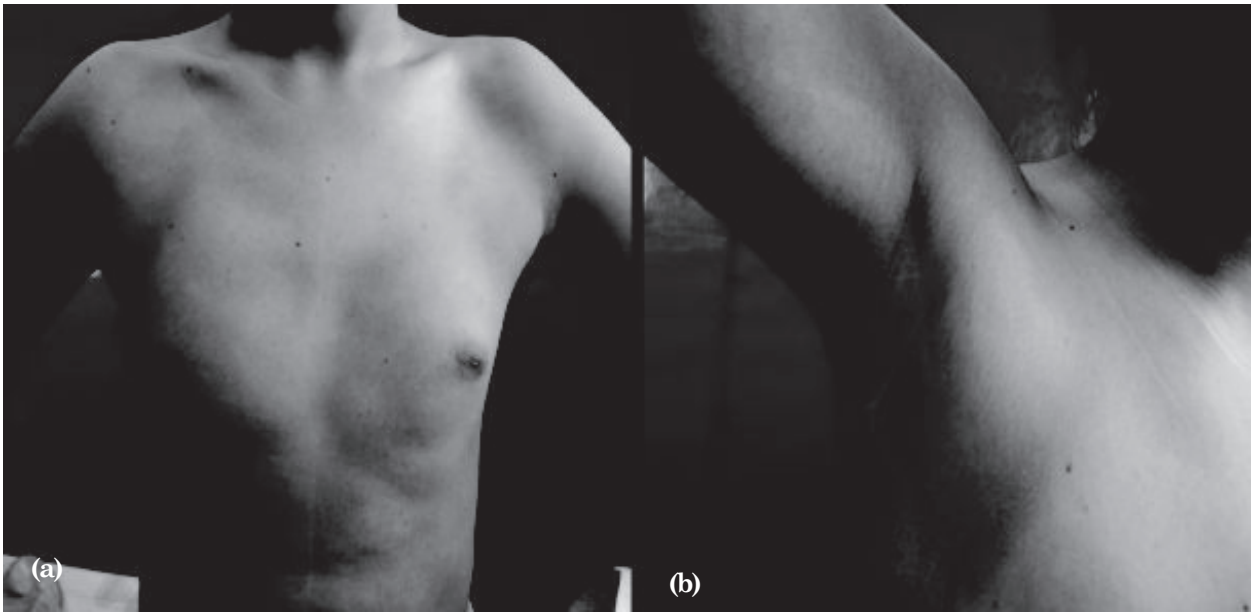


**Fig 1(a):** *Under developed breast* **(b)** *Absence of axillary hair*

- Ultrasonogram showed small, hypoplastic uterus, length - 38.3 mm and AP diameter - 7.2 mm with no differentiation between endometrium and myometrium and bilateral streaky gonads without any follicle (Figure: 2-a).
- Karyotyping revealed 46 XY with no mutation of SRY gene by Fluorescence in situ hybridization. Bilateral gonadectomy was done by laparoscopy and advised hormone replacement therapy with conjugated estrogen.
- Follow up ultrasonogram after one year revealed improvement of uterus size (Figure 2-b: length 58.7 mm and AP diameter -15.4 mm) and noticed for menstrual cycles and breast development.

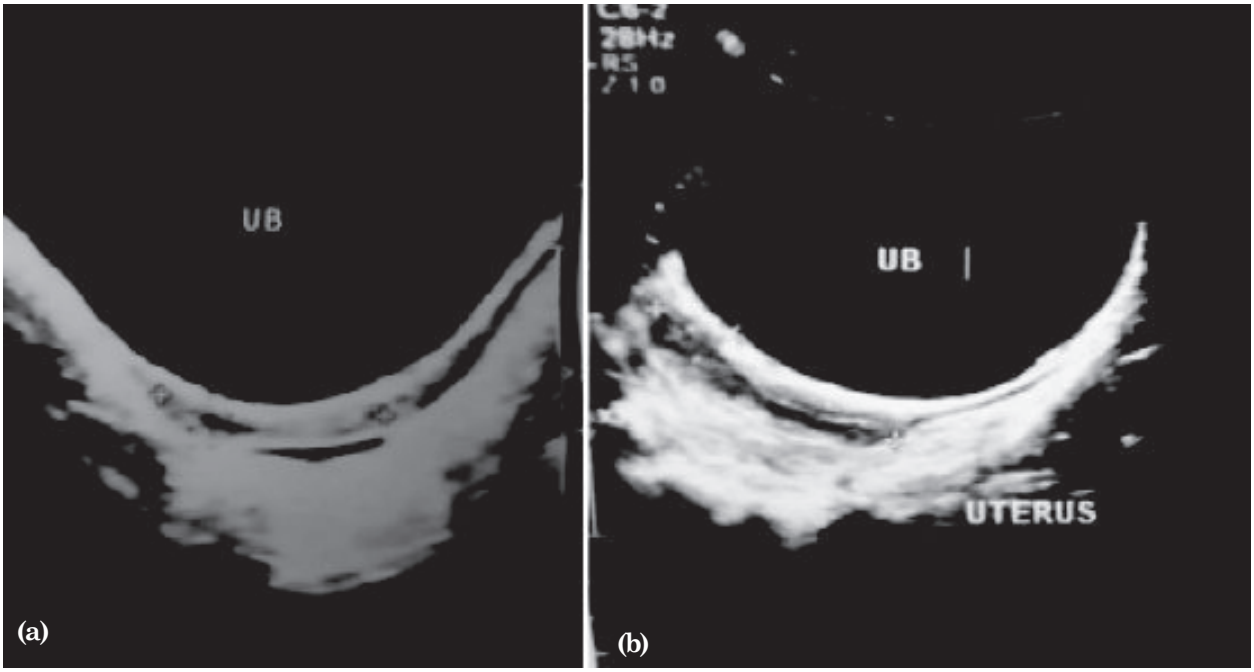


**Fig 2:** *Ultrasonogram of uterus* ***(a)*** *during first follow up and* ***(b)*** *after one year*

**Discussion:**

Swyer syndrome was first described in 1955 which is a rare disorder of sexual dysgenesis. Multiple mutations in the SRY gene are the main factor to complete gonadal dysgenesis and the retention of Mullerian ducts in a genetic XY male.[2] Mutations in other genes, such as ZFY, SOX9, SF1, WT1, DYZ1, and DAX1, are possible causes in the interfering of sex determination. Complete gonadal dysgenesis is associated with an absence of testicular differentiation in a phenotypic female with a 46XY karyotype.[3] Normally, anti-Mullerian hormone is synthesized by Sertoli cells, which are activated by the SRY gene found on the Y chromosome. Anti-Mullerian hormone then causes regression of Mullerian structures. If anti Mullerian hormone secretion fails to occur by six to eight weeks of intra uterine life, because of a defect in testicular differentiation or a mutation of the SRY gene, external female genitalia form. Female internal genitalia development varies according to the exact timing of secretion of anti-Mullerian hormone. Women with complete gonadal dysgeneses have an overall smaller uterus size when compared to normal controls which is the important diagnostic and therapeutic implications in these patients.

Mayer-Rokitansky-Küster-Hauser syndrome (XX), characterized by varying degrees of Müllerian duct abnormalities and a rudimentary or absent uterus and complete androgen insensitivity syndrome XY with normal breast and vaginal development, but with no uterus are common differential diagnosis of primary amenorrhea.[4]

**Conclusion:**

Swyer syndrome is very rare disorder of sexual dysgenesis with increased risk of malignancy. Accurate diagnosis, early surgery can reduce mortality and morbidity and experienced normal sexual life by hormone replacement therapy.

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