

A case of Swyer syndrome with spontaneous breast development and menstruation possibly related to gonadoblastoma formation



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INTRODUCTION:

Swyer syndrome (46, XY pure gonadal dysgenesis) is characterized with normal female genitalia at birth, underdeveloped müllerian structures and fibrotic, primitive and non-productive gonads. It is estimated to occur in 1:80.000 births.

Patients are usually diagnosed in adolescence with delayed puberty and primary amenorrhea. Classically, hypergonadotropic hypogonadism leads to the absence of spontaneous breast development and menstruation. However, in the literature three cases with spontaneous breast development and a few numbers of assisted full-term pregnancies have been reported. Additionally, a unique case with spontaneous menstrual cycles and a fertile woman with a predominantly 46, XY karyotype that gave birth to daughters with gonadal dysgenesis have also been described.

AIM:

We aimed to present an apparently female adolescent with Swyer syndrome, who had spontaneous breast development and menstruation considered to be due to the active hormone secretion from gonadoblastoma.

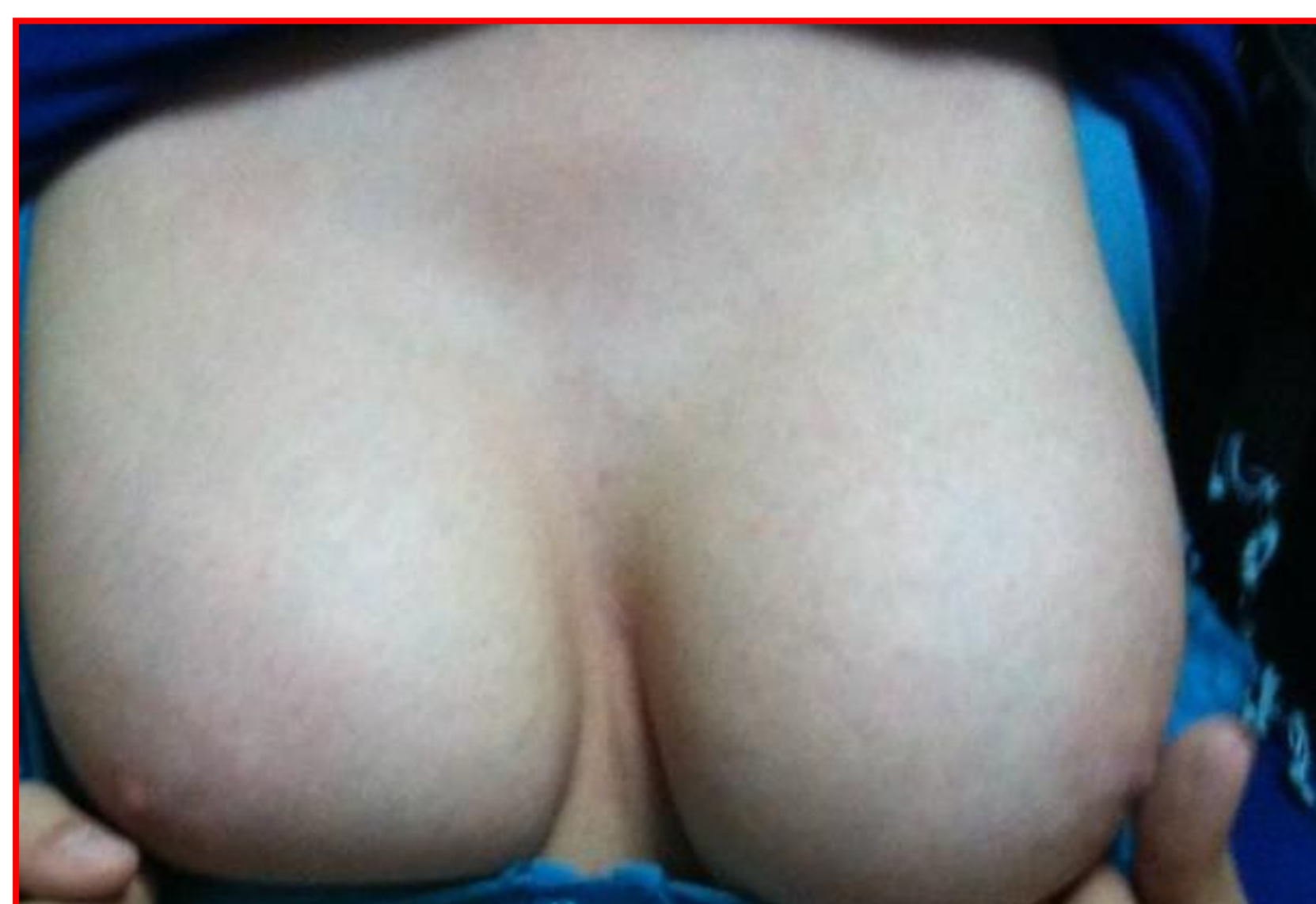


Figure 1. Tanner stage V breast development

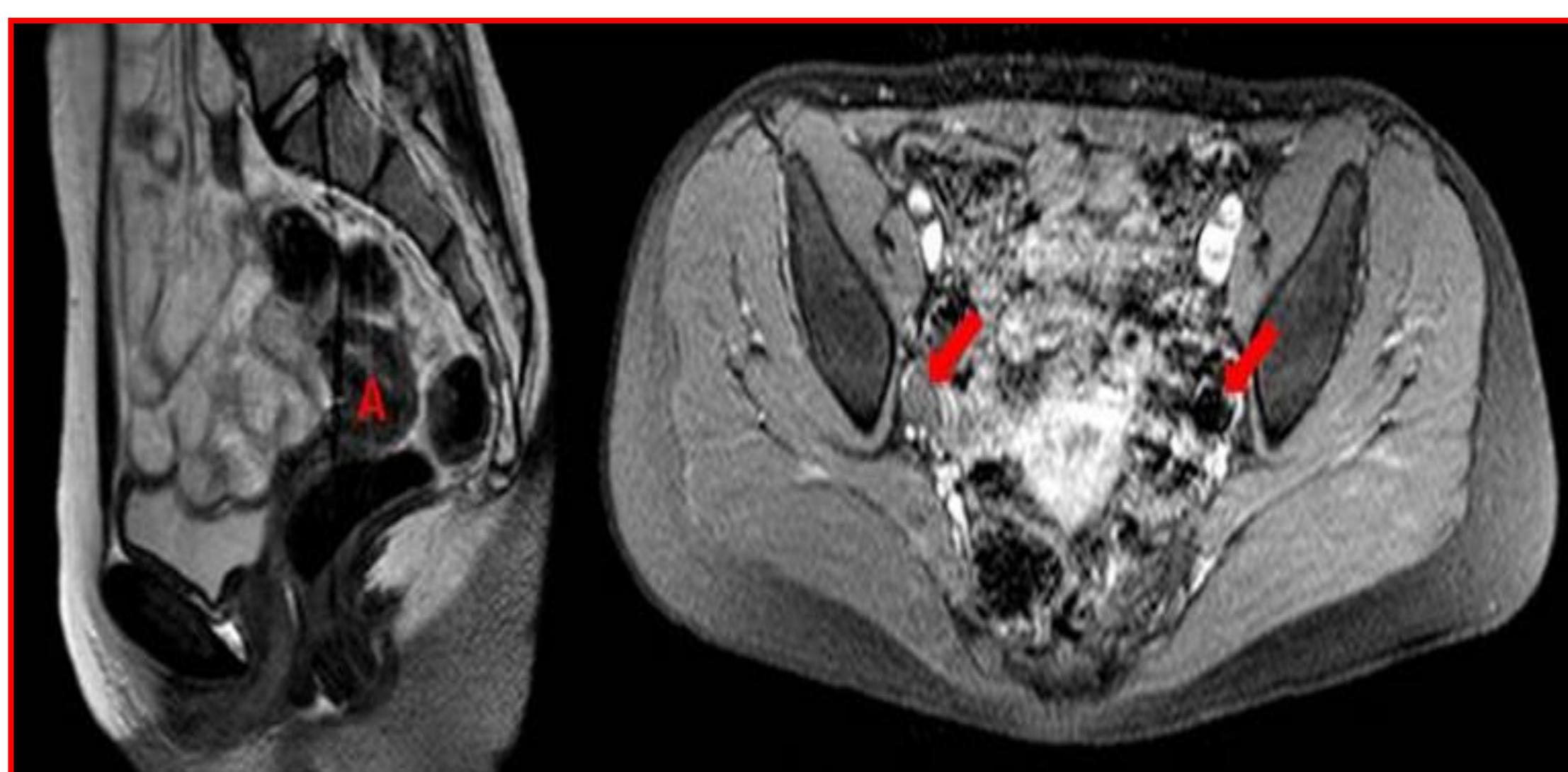


Figure 2. Magnetic resonance imaging of uterus (A) and ovaries (Red arrows)

CASE REPORT

A 15-year-old girl with primary amenorrhea

Past medical history:

- ❖ Thelarche: 10 years old
- ❖ Pubarche: 11 years old

Physical examination:

- ❖ Weight=55.2 kg (0.25 SD)
- ❖ Height=165.5 cm (0.95 SD)
- ❖ BMI= 20.1 kg/m² (0.10 SD)

Her external genitalia was completely female with a breast development and pubic hair compatible with Tanner's stage V (Figure 1).

Laboratory Evaluation;

- ❖ FSH: 121 mIU/mL (N, 0.3–10), LH: 13 mIU/mL (N, 0.3–31),
- ❖ E₂: 66 pg/mL (normal, 15–350)
- ❖ Total testosterone: <20ng/dL (N, 15–181)
- ❖ Prolactin: 5.3 ng/mL (N, 1.9–25)
- ❖ β-HCG: <1 mIU/mL (N, 0–10)
- ❖ ACTH: 50.3 pg/mL (N, 0–46), Cortisol: 14 µg/dL (N, 5–25)

Radiologic Evaluation;

- ❖ Pelvic USG: right ovary: 1.4 cm³, left ovary: 2.4 cm³; uterus of 60x25x23 mm, endometrial thickness of 7 mm
- ❖ MRI: Small ovaries (Figure 2)

Karyotype analysis

- ❖ 46, XY SRY (+)
- ❖ No mutation in the SRY, SF1 and WT1 genes

Swyer Syndrome (Diagnosis)

Clinical Follow-up

- ❖ Temporarily the patient has started to have spontaneous menstrual cycles
- ❖ After nine months the patient underwent diagnostic laparoscopy

1. Biopsy

- ❖ Frozen examination of multiple biopsies from gonad tissues revealed gonadoblastoma

2. Treatment

- ❖ Bilateral gonadectomy
- ❖ Hormone-replacement therapy

CONCLUSION:

- ❖ Swyer syndrome is a rare condition
- ❖ It can be easily diagnosed if presented with classical symptoms
- ❖ Patients can also present with spontaneous breast development and/or menstruation, which may delay the diagnosis
- ❖ Swyer syndrome patients have a high incidence of neoplastic transformation and germ cell tumors

We suggest that normal pubertal development in patients with Swyer syndrome may be associated with the presence of a hormonally active tumor (gonadoblastoma?)