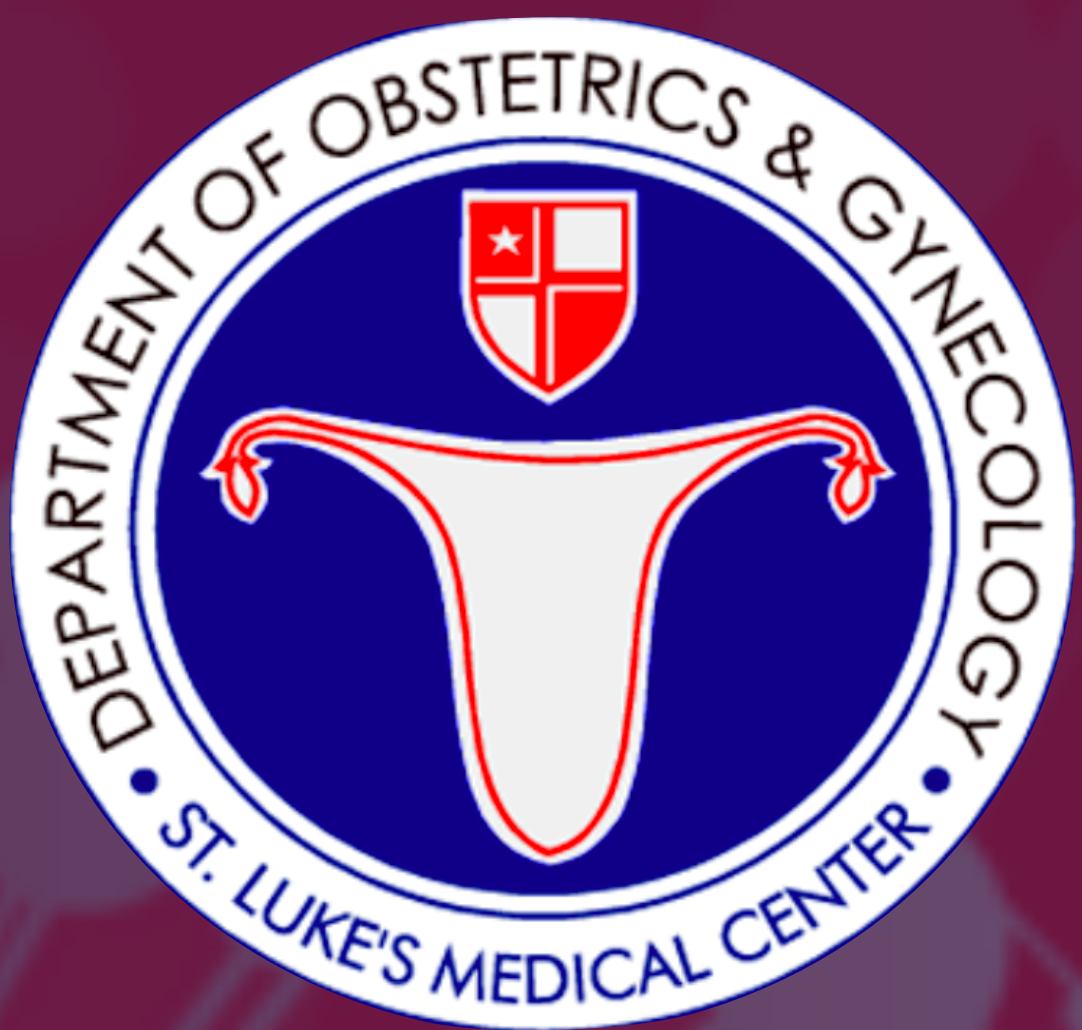




SWYER SYNDROME (46,XY COMPLETE GONADAL DYSGENESIS): A Rare Case of Primary Amenorrhea

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INTRODUCTION

Swyer syndrome is a rare type of gonadal dysgenesis wherein a 46,XY karyotype presents with a female phenotype. It occurs in 1:100,000 births. **SRY gene mutations** are the root cause of nonfunctional gonads with no hormonal or reproductive potential.

CASE REPORT



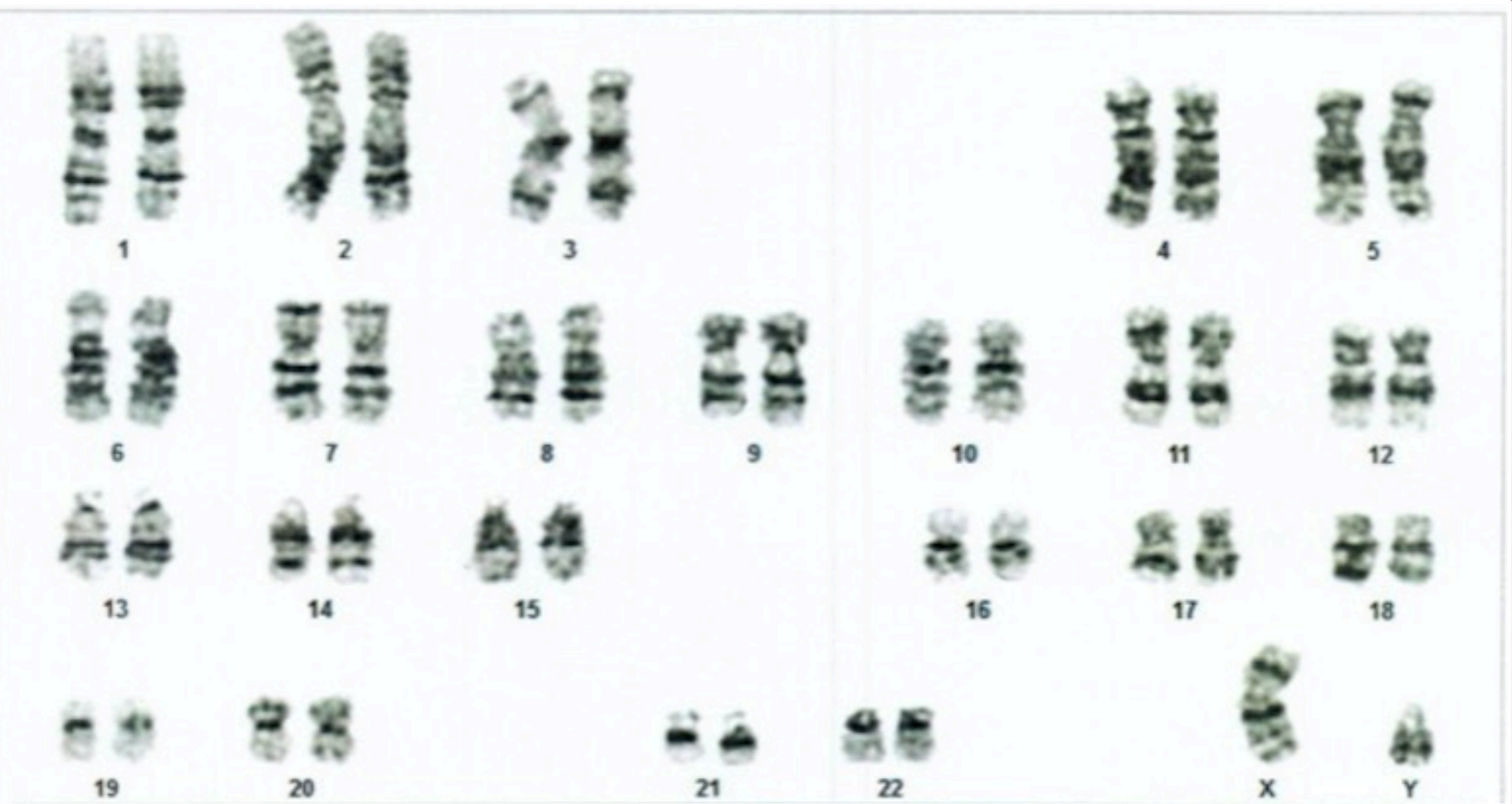
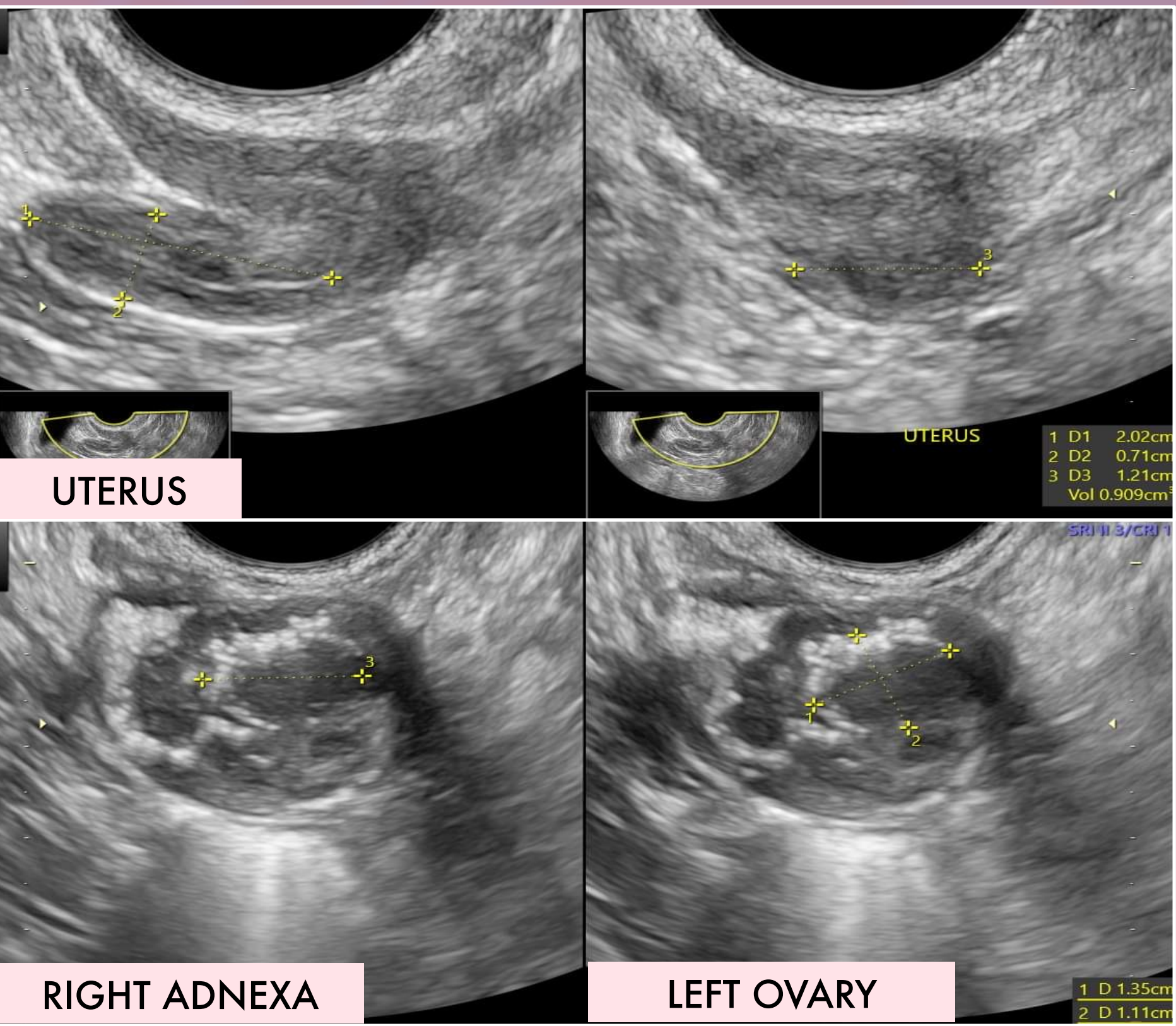
23-year-old, phenotypically female, single, who presented with **primary amenorrhea**

No associated vasomotor symptoms, headache, bowel, or urinary symptoms. On PE, she has phenotypically female features, BMI of 26 kg/m². Breasts were at Tanner stage 3 and pubic hair was at Tanner stage 2. No abdominal masses were palpable; and the abdomen was soft and non-tender. External genitalia had normal labia major and minora & slightly enlarged clitoris.



RESULTS AND DISCUSSION

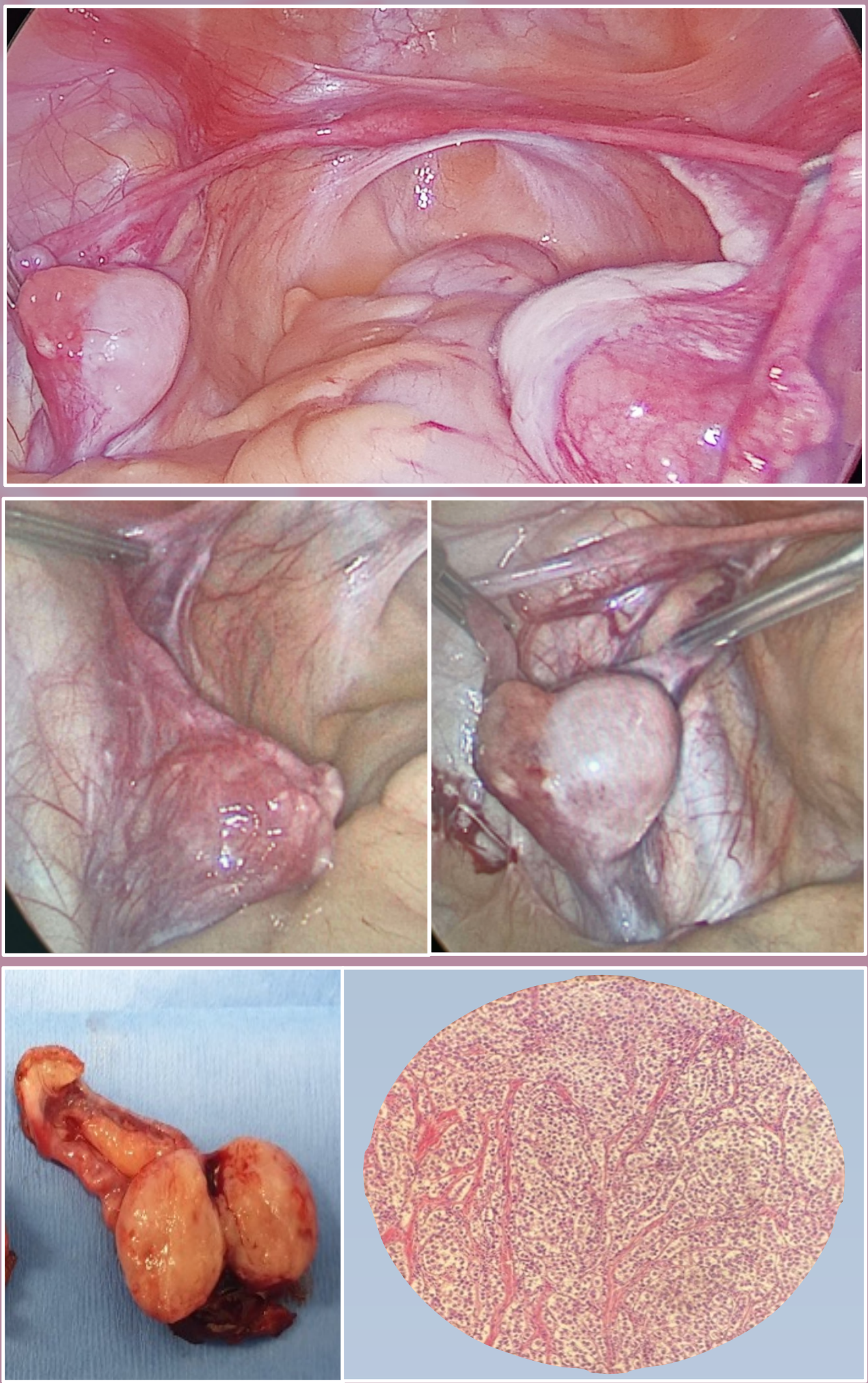
FSH	50.72 mIU/mL	↑
Estradiol	<5.00 pg/mL	↓
Prolactin	3.02 ng/mL	normal
LH	14.02 mIU/mL	normal
TSH	0.70 µIU/mL	normal
Hba1c	5.5%	normal
Creatinine	0.83 mg/dL	normal



The hormonal assays done confirmed a **hypergonadotropic-hypogonadic state**. This points to **ovarian failure** wherein the ovaries fail to produce enough estrogen leading to the absence of negative feedback on the hypothalamus and pituitary gland. **Transrectal ultrasound** showed an infantile cervix and uterus with thin endometrium (0.9cm), non-visualized right ovary, and streak left ovarian tissue. A cystic mass was noted on the right pelvic area with mixed echogenicity and no color flow measuring 3.3 x 2.9 x 2.8cm. The uterus is infantile in size due to the lack of sufficient estrogen stimulation. The gonads are dysgenetic with no hormonal function; that is why the hormonal assay would show a hypergonadotropic hypogonadic state. A **karyotype study** was advised which revealed normal **male 46,XY** karyotype. Despite having a Y chromosome, the phenotype presents as female because the gonads fail to produce AMH.

CONCLUSION

The patient underwent **laparoscopic bilateral gonadectomy with salpingectomy**. Intraoperatively, the uterus appeared flattened and infantile in size, with a pink serosal surface. The left adnexa measured 2 x 2 x 1.5 cm and the right adnexal mass was cystically enlarged to 3.5 x 3 x 2 cm. The final histopathology result: **dysgerminoma**



Management warrants a **multi-disciplinary approach**. Patient was started on hormonal therapy and monitoring every 3 to 6 months with chemotherapy with bleomycin. Successful pregnancies have been reported using ART with oocyte donation and embryo transfer. The endpoint is to give the patient, and her family, reassurance as this case is a manageable condition; a productive life is achievable.