

A Rare Case of Primary Amenorrhoea

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II year PG

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- A **26** year old female, XXX, resident of Warangal, teacher by occupation belonging to upper middle class came to gynaecological out patient department with the chief complaint of **not attaining menarche till date.**

H/O Present illness

- Patient was apparently asymptomatic and has been kept on not attaining menarche till date from the age of 15 years.
- No h/o cyclic abdominal pain.
- No h/o excessive weight gain/weight loss/excessive exercise.
- No h/o cold intolerance/ excessive sweating/ constipation/diarrhea/hairloss.
- No h/o headache, visual disturbances, hearing problems
- No h/o breast secretions.

Past history

- Development of breast at 15 years of age.
- Pubic and axillary hair 6 months later.
- She then approached a gynaecologist, was investigated and told that she had no internal genital organs.
- No follow ups were done there after.

Past history contd..

- No h/o tuberculosis , mumps
- No h/o thyroid disorders.
- No h/o any surgeries, radiotherapy ,chemotherapy.
- Not a k/c/o hypertension, diabetes, asthma, epilepsy, heart disease, or any other chronic illnesses.
- No h/o any drugs/ medication use.

Personal history

- Diet – mixed.
- Appetite – normal.
- Sleep – adequate.
- Bowel and bladder – regular.
- Not a smoker / alcoholic.
- Not sexually active.

Family history

- Her mother attained menarche at 14 years of age and conceived spontaneously for both her pregnancies.
- Patient was second in order of birth.
- Perinatal history was not significant
- Patient was good at studies.
- She had an elder brother who is pursuing his degree, unmarried and his fertility status is not known.
- No similar complaints, infertility, early menopause, genetic, metabolic, autoimmune diseases in the family.

General Examination

- Patient is conscious coherent oriented.
- Moderately build and moderately nourished.
- Height – 150 cm
- Weight – 54 kg
- BMI – 24 kg/sq m

- No pallor/icterus/cyanosis/clubbing/koilonychia/pedal edema/generalised lymphadenopathy.

- No acne/ features of hirsutism/acanthosis nigricans seen.
- No obvious neck swelling seen.
- Spine and gait were normal.
- No turner stigmata seen.
- Bilateral breast – tanner stage 2

Systemic examination

- Temperature – 98.4 F.
- PR – 80 bpm.
- BP – 110 /80 mm Hg.
- CVS – S1 S2 heard, no murmurs.
- Respiratory system- BAE+ , NVBS +, no added sounds.
- CNS – no abnormality detected.

- Abdominal examination- abdomen is flat , umbilicus central, no scars, no sinuses, no striae seen.
- No organomegaly.
- No masses palpable in inguinal region.

- Vulvo vaginal examination
 - External genitalia – labia majora
 - labia minora
 - clitoris
- } well developed
- Pubic hair – tanner stage 3.
 - Hymen is intact.
 - Vagina is 3 inch long as measured with uterine sound.
 - Per rectal examination- 4 x 2 x 1 cm sized mass felt anteriorly, probably uterus.

Working diagnosis

A 26 year old female with primary amenorrhea secondary to

- 1) turners syndrome (mosaic forms)
- 2) mullerian anomaly
- 3) gonadal dysgeneis

Investigations

- Pelvic ultrasound- both the uterus and the ovaries could not be visualised. There were no other anomalies detected.
- MRI pelvis showed the same.

- Serum TSH - 2.18 mIU/L. (normal)

- S.FSH - **72.0 mIU/ml** ↑

- S.LH - **21.11 mIU/ml** ↑

- S. Prolactin- 15 ng/ml

- S. Estradiol - **12.90 pg/ml** ↓

- S. AMH - **0.10ng/ml** ↓

- DHEAS - 120mcg/dl

- Testosterone - 54ng/dl

Normal Range

<40mIU/ml

<20mIU/ml

<25ng/ml

65-380mcg/dl

15-70ng/dl

- Karyotype - **46 XX with no Y chromosome.**
- **Diagnostic laparoscopy** - uterus of size approximately 5 × 3 × 2 cms was seen. The cervix and the body of the uterus were approximately of the same size.
- Both the ovaries were streak.
- Vaginoscopy performed through 5mm hysteroscope which revealed normal vaginal and cervical development .
- The findings were suggestive of Hypoplastic uterus and streak ovaries.

Summary

- 26 year old with Primary amenorrhea.
- Normal vagina and hypoplastic uterus.
- Bilateral streak ovaries.
- Hypergonadotropic hypogonadism.
- 46 XX with no Y chromosome.
- No somatic abnormalities.

- **Diagnosis –**

26 year old female with primary amenorrhea secondary to primary ovarian insufficiency, 46 XX, pure gonadal dysgenesis (diagnosis of exclusion).

Confirmatory – Ovarian biopsy for which the patient did not give consent.

THANK YOU