Primary Amenorrhea

Learning Objectives

By the end of this lecture students should be able to:

1. Define **primary amenorrhoea**.
2. Describe the **physiology of normal menstruation**.
3. List the **common causes** of primary amenorrhoea.
4. Understand the **clinical evaluation and diagnostic algorithm**.
5. Identify **important investigations**.
6. Outline **management according to the cause**.

Definition

Primary amenorrhoea is defined as:

• **Absence of menarche by age 15 years with normal secondary sexual characteristics**, OR
• **Absence of menarche by age 13 years with no secondary sexual characteristics**.

Evaluation should also begin **3 years after thelarche if menstruation has not started**.

Normal Physiology of Menstruation

Normal menstruation requires integrity of the **Hypothalamic-Pituitary-Ovarian-Uterine axis**:

1. Hypothalamus → GnRH secretion
2. Pituitary → FSH & LH
3. Ovaries → Estrogen & progesterone
4. Uterus/endometrium → menstrual shedding

Failure of any component results in **amenorrhoea**.

Causes of Primary Amenorrhoea

Primary amenorrhea is broadly divided into **four groups**:

1. Outflow Tract Abnormalities

- Imperforate hymen
- Transverse vaginal septum
- Müllerian agenesis (MRKH syndrome)
- Cervical agenesis

2. Ovarian Causes

- Gonadal dysgenesis (Turner syndrome – most common)
- Primary ovarian insufficiency
- Enzyme defects in steroidogenesis

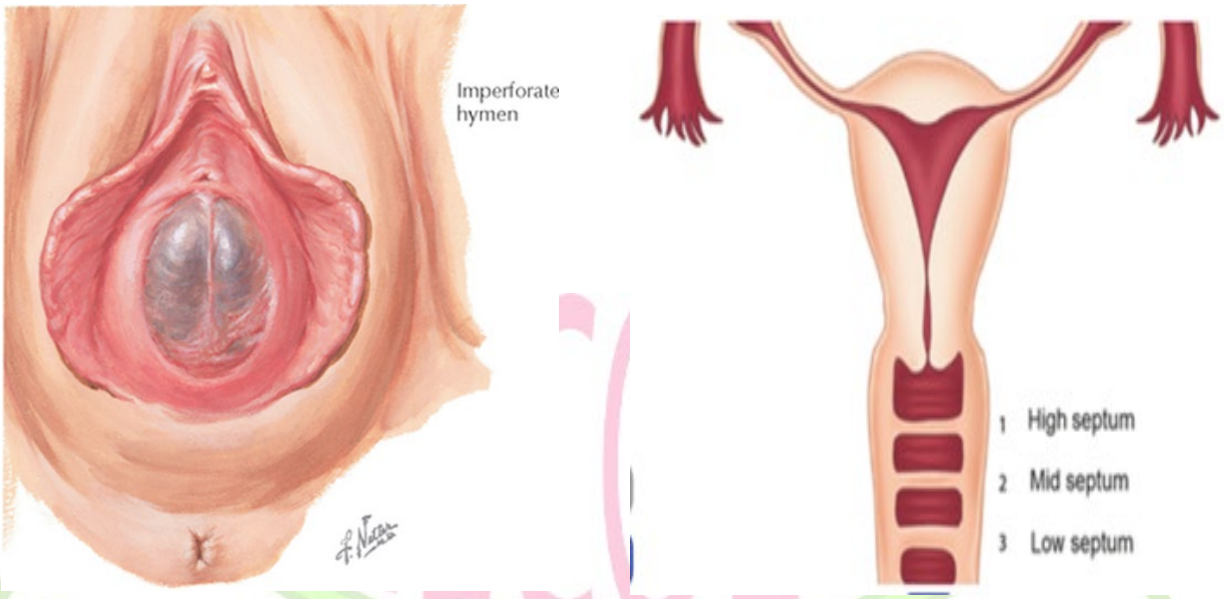
3. Hypothalamic or Pituitary Disorders

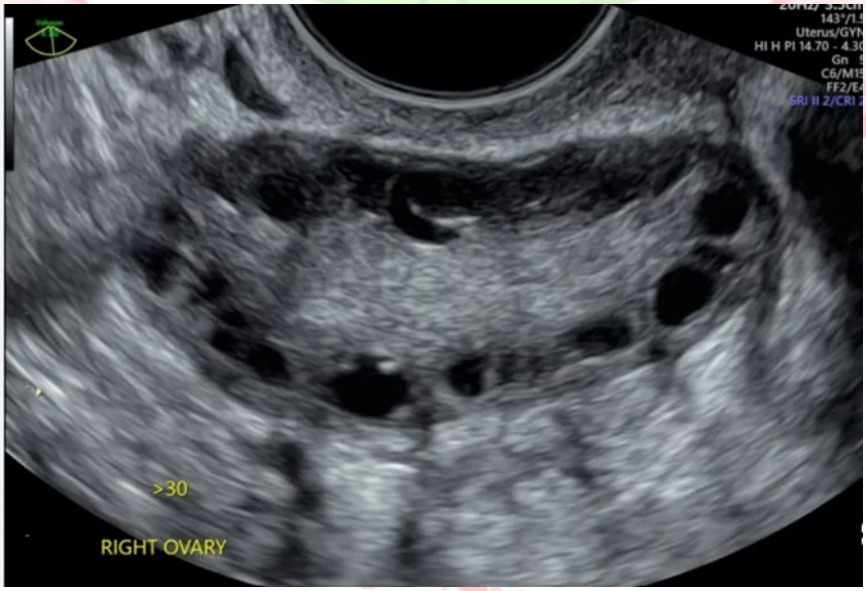
- Constitutional delay of puberty
- Kallmann syndrome
- Hypothalamic dysfunction
- Pituitary tumors
- Hyperprolactinemia

4. Endocrine Disorders

- Hypothyroidism
- Congenital adrenal hyperplasia
- Polycystic ovary syndrome

Common etiologies include **gonadal dysgenesis, Müllerian agenesis, and constitutional delay of puberty**.





Clinical Presentation

Patients may present with:

- Primary amenorrhea
- Delayed puberty
- Cyclic pelvic pain (suggests obstruction)

• Short stature (Turner syndrome)
• Hirsutism (PCOS or androgen excess)

History Taking

Important points:

1. Age and pubertal development
2. Breast development (thelarche)
3. Family history of delayed puberty
4. Cyclic abdominal pain
5. Chronic illness or weight loss
6. Exercise and nutritional status
7. Drug history.

Physical Examination

General examination:

- Height and weight
- Tanner staging
- Signs of virilization
- Dysmorphic features (Turner)

Pelvic examination:

- Vaginal patency
- Presence of uterus
- External genital abnormalities

Initial Investigations

Initial tests recommended in guidelines include:

• **Pregnancy test**
• **Pelvic ultrasound**
• **FSH, LH, Estradiol**
• **Prolactin level**
• **TSH**

Additional tests depending on findings:

- Karyotype
- Testosterone
- 17-hydroxyprogesterone
- MRI brain (pituitary lesions)

Diagnostic Approach

Step 1: Assess secondary sexual characteristics

A. Breast development present→ Evaluate uterus

If **uterus present**
- Outflow obstruction
- PCOS
- endocrine disorders

If **uterus absent**
- Müllerian agenesis
- Androgen insensitivity syndrome

B. No breast development

Check **FSH**

• High FSH→ Gonadal failure (Turner syndrome)

• Low or normal FSH→ Hypothalamic or pituitary causes

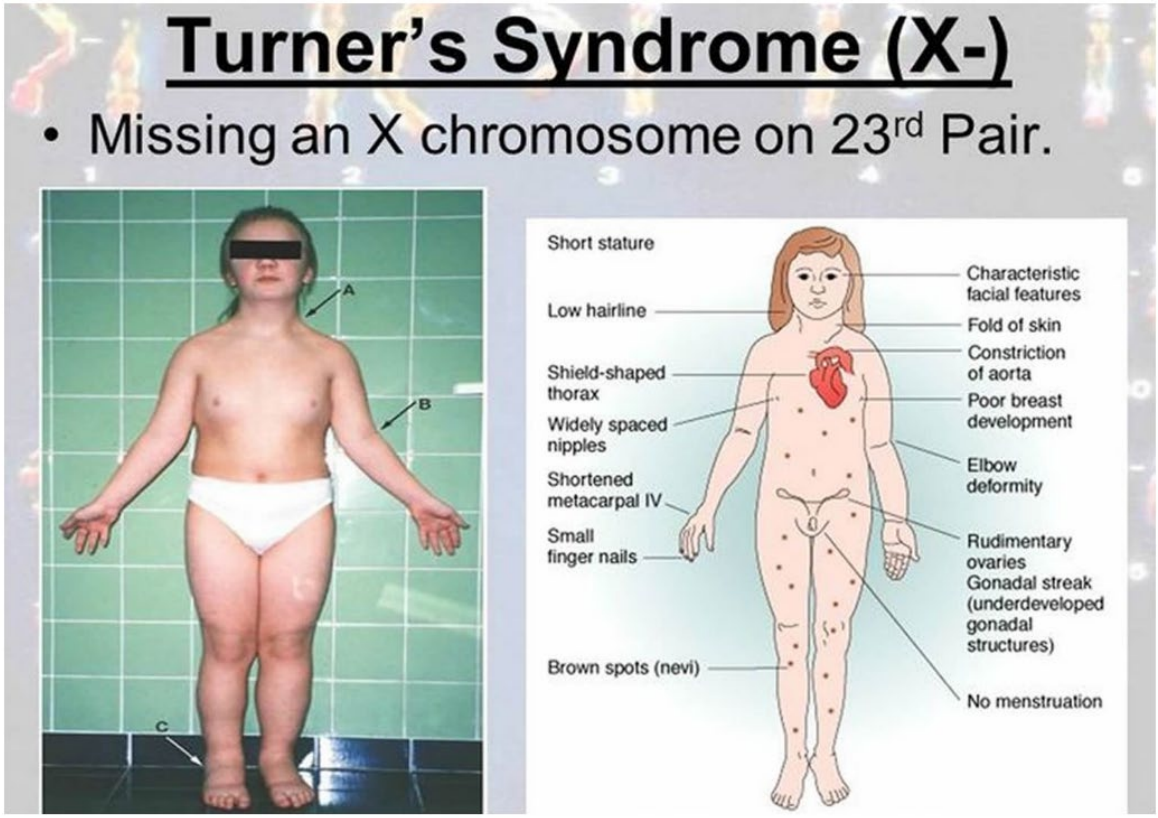
Important Causes to Know

1. Turner Syndrome

Features:
- 45,XO karyotype
- Short stature
- Webbed neck
- Streak ovaries
- High FSH

Management:

- Estrogen replacement
- Fertility counseling



Müllerian Agenesis (MRKH)

Features:

- Normal secondary sexual characteristics
- Absent uterus
- Normal ovaries
- Normal karyotype (46XX)

Management:

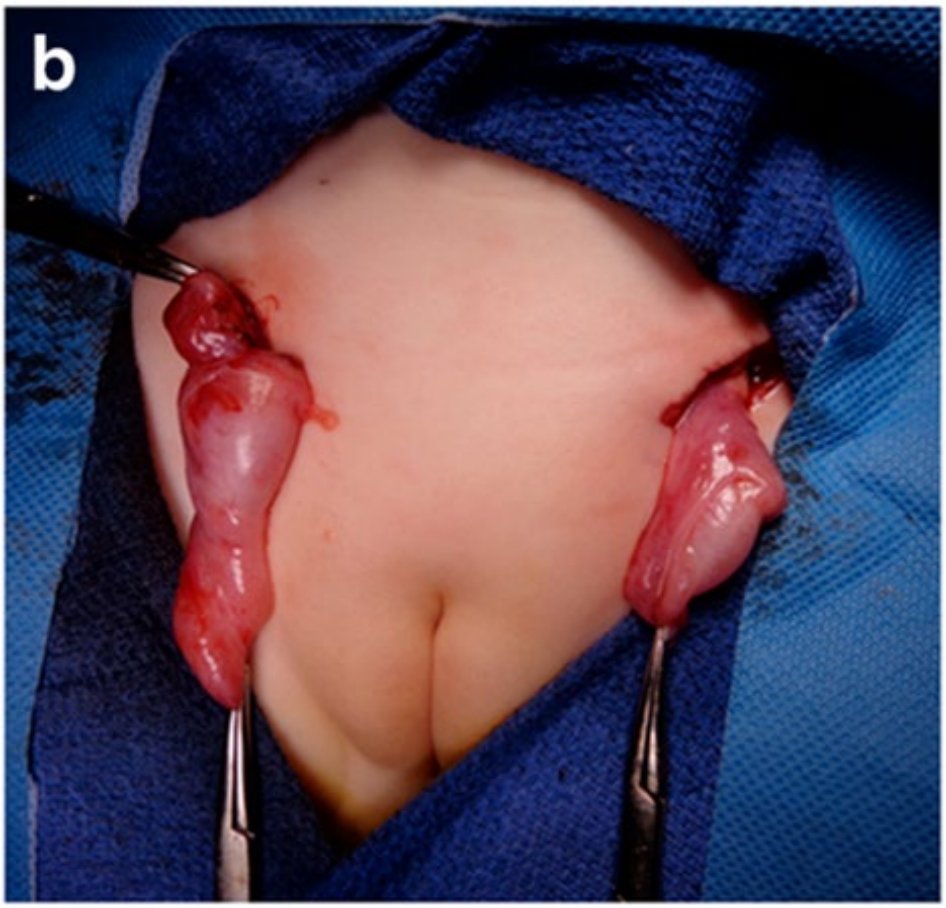
- Vaginal reconstruction
- Fertility via surrogacy

Androgen Insensitivity Syndrome

Features:

- 46 XY
- Female phenotype
- Absent uterus
- Undescended testes
- Breast development present

Management:gonadectomy after puberty



Management of amenorrhea : Treatment depends on the **underlying cause**.

Medical Management

- Hormone replacement therapy
- Treatment of thyroid disease
- Dopamine agonists for hyperprolactinemia

Surgical Management

- Hymenectomy
- Vaginal septum resection
- Vaginal reconstruction

Supportive Care

- Psychological counseling
- Fertility counseling

Complications

Untreated causes may lead to:

- Infertility
- Osteoporosis
- Psychological distress
- Endometrial pathology

Key Points for Students: Stepwise approach:

Secondary sexual characteristics present?

- Yes → think outflow obstruction / Müllerian agenesis / AIS
- No → think gonadal failure / hypothalamic causes

2. **Uterus present?**
    - Yes → endocrine causes
    - No → MRKH or AIS
3. **Hormonal profile**
    - High FSH → ovarian failure
    - Low FSH → hypothalamic/pituitary