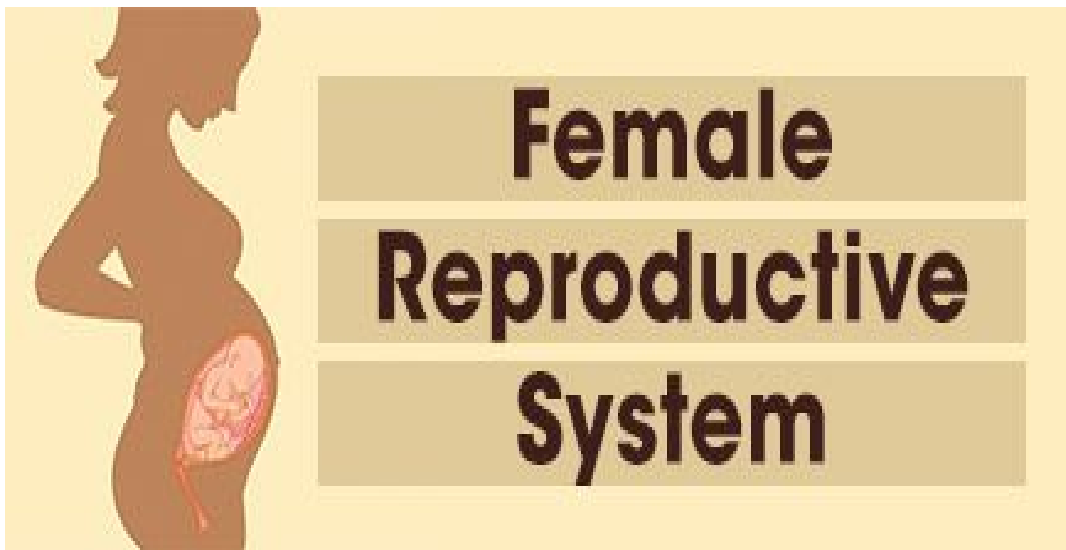




The 2nd week



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Case 1

A father of an 6 years old boy noticed that his son is having a growing mustache, and his voice is changing to a heavier one, the father said: my son's voice sounds like a broken motor bike, moreover the father says: my son's body is growing very fast, I can see his cephalic vein between the muscles. So they book an appointment in KKUH and it was after 6 months because there is a lot of people before them. They see the doctor after 8 hours of waiting, during clinical examination the physician notice the boy has enlarged genitalia in relation to his age, and he ordered some lab investigations.

Q1: What is your diagnosis ?

Congenital adrenal hyperplasia syndrome

Q2: What do you think is the most common type of this syndromes ? and give 2 other types.

Most common: 21 α hydroxylase deficiency, 11 β and 17 α hydroxylase deficiency

Q3: In 21 α hydroxylase deficiency, what is the lab test used for diagnosis ?

High level of plasma [17-hydroxyprogesterone]

Q4: What is the mode of inheritance of this syndrome ?

Autosomal recessive

Q5: In female patient, what is the signs of the syndrome ?

Virilization of girls (ambiguous genitalia -FPH-), and also rapid growth.

General questions:

What is the usual positions of the uterus ?

Anteverted anteflexed

Mention 3 structures support the uterus in position.

-Ligaments of cervix -Round ligament of uterus -Levator ani muscles

What are the derivatives of paramesonephric duct in female ?

- Upper Vagina -Cervix and Uterus -Fallopian Tubes

Mention 3 structures found in deep perineal pouch ?

- Part of urethra
- Part of vagina
- Sphincter urethrae muscle

What are the result of female sexual cycle ?

1. *Single* ovum is released from the ovaries each month
2. Uterine endometrium is prepared for implantation of the fertilized ovum.

Mention 3 possible disorders of menstruation ?

Menorrhagia: Refer to abnormally heavy or prolonged bleeding.

Hypomenorrhea: Refer to scanty flow.

Dysmenorrhea: Painful menstruation.

What is the of FSH in ovarian cycle ?

Acceleration of growth of primary follicles.

What is function of LH in ovarian cycle ?

- Extrusion of the ovum from the follicle.
- Change of granulosa and theca interna cells into lutein cells.
- Secretion of progesterone & estrogen from the corpus luteum.

Case 2

16-year-old girl is brought to her pediatrician because of an absence of menarche. She has short stature, a webbed neck, and a square chest. Physical examination reveals breast buds and female external genitalia. Her blood pressure is normal in both arms. CT scan reveals a small uterus and atretic, fatty ovaries. There is no known history of this condition in her family .

Q1;What is the most likely diagnosis?

A1:turner syndrome, characterized by gonadal dysgenesis secondary to the presence of a single X chromosome (XO). this syndrome is the most common cause of primary amenorrhea. this genetic disorder affects 3% of all conceptions, but only 1:1000 45X embryos survives to term.

primary amenorrhea	Secondary amenorrhea
Refers to the complete absence of menstruation by 16 years of age.	Refers to cessation of menstruation for more than 6 months after menarche.

Q2:What other conditions can cause primary amenorrhea?

A2: Other causes include the following:

- **absence of uterus, cervix, and/or vagina (müllerian agenesis).**
- **hypothalamic hypogonadism (secondary to anorexia, exercise, stress, or gonadotropin-releasing hormone deficiency).**
- **Ovarian failure (gonadal dysgenesis, or polycystic ovarian syndrome).**
- **pituitary disease.**
- **transverse vaginal septum or imperforate hymen.**

(compared with, which is cessation of menstruation for more than 6 months after menarche).

Q3:What diagnostic test is indicated based on the patient's clinical features?

A3:Karyotype analysis should be performed.

Q4:What other conditions are associated with Turner syndrome?

A4:• Coarctation of the aorta.

- Bicuspid aortic valve.
- hypothyroidism.
- Sensorineural hearing loss.
- renal abnormalities.
- Gastrointestinal telangiectasias.
- Osteoporosis.

Q5:What are the appropriate treatments for this condition?

A5:recombinant human growth hormone and hormone replacement therapy can initiate puberty and complete growth. treatment of other associated conditions is also advised.