

Congenital malformation of FGT

Normal genetic differentiation

Following fertilization the normal embryo, contain 46 chromosomes including 22 autosomes and 1 chromosome derived from each parent.

46 xx develop as a female and 46 xy develop as a male.

Y chromosome contain a region known as SRY region (sex determining region of the chr. Y), in males this gene triggers testis formation.

Ovarian differentiation determined by the presence of two x chromosomes and ovarian determinant gene located on short arm of X chromosome.

Normal anatomical differentiation

- Embryologically mesonephric (wolffian) develop into male genital structures
- The two-paramesonephric (mullarian) ducts develop as upper female genital structures including fallopian tubes and after fusion of both tubes develop into uterus, upper third of vagina.
- Urogenital sinus develop into lower two thirds of vagina.
- Ovaries differentiated from coelomic epithelium.

Disorders of sexual development

Heamphroditisim

In which ovarian and testicular tissue present at the same time in a various degree result in ambiguous genitalia.

Most common karyotyping 46xx but may be 46xx/xy or 46 xy.

Management

The sex will be determined at puberty by gender role or functional capability of the external genitalia and removal of inappropriate organ.

Congenital adrenal hyperplasia

- Most common disorder.
- Deficiency of 21-hydroxylase enzyme lead to increase 17 hydroxyprogesteron.
- Karyotyping 46xx
- Autosomal recessive disease.
- Present at birth as ambiguous external genitalia like labial adhesion and clitoral hypertrophy and salt losing syndrome, normal internal organs

- Management by replacement of mineralocorticoid at birth and cortisol administration long life.

Turner syndrome

- Karyotyping 45O
- Phenotype female with features of short stature, webbed neck, and shield shape thorax, elbow deformity, primary amenorrhea due to gonadal dysgenesis, cardiac and renal diseases.
- Management by hormone replacement therapy by estrogen and growth hormone to achieve height of 150 cm and breast development and after age of 11 years by estrogen and progesterone .
- Pregnancy can be obtained by ova donation.

5 alpha reductase deficiency

- Karyotyping 46 xy
- The defect in 5 alpha reductase which responsible for conversion of testosterone into dihydrotestosterone which is important in development of male external genitalia.
- Presented as ambiguous genitalia at birth and some degree of virilization at puberty.
- Sex determined by gender role

Androgen insensitivity syndrome

- Karyotyping 46 xy
- Receptors not respond to testosterone hormone .
- Presented with female external genitalia at birth and absence of menstruation (no uterus) and sparse axillary and pubic hair at puberty, blind end vagina, with presence of testis in the abdomen or inguinal region.
- Management by gonadectomy after puberty because of risk gonadoblastoma and creation of vagina for sexual satisfaction.

Mayar-Rokatinsky-Kuster-Hauser syndrome

- Karyotyping 46 xx
- Mullarian agenesis result in absent uterus and tube and upper vagina.
- Female present with normal external genitalia ,blind end vagina and normal secondary sexual charechteristics with primary amenorrhea.
- Associated with urinary tract anomalies in 40% and skelatal abnormality in 25%.
- Management by psychological counselling,vaginal cration and pregnancy by surrogate uterus.

Other structural malformation

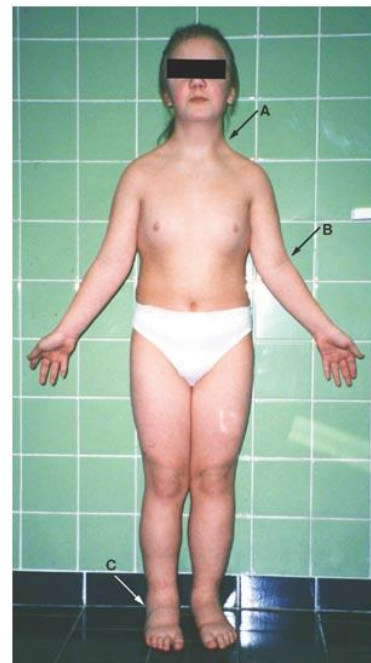
- **Imperforated hymen** due to incomplete canalization of vagina, managed by crciate incision of hymen.

- **Septate uterus** due to incomplete failure of fusion of two mullarian ducts may present with recurrent miscarriage and malpresentatin during labor and management by removal of septum
- **Uterine didyphylus** dut to complete failure of fusion of two mullarian ducts (pregnancy occur in one cavity)
- **Bicornuate uterus** when there is deep indentation of fundus present with recurrent miscarriage and malpresentation.
- **Unicornuate uterus** when there is failure of development of one mullarian duct associated with recurrent miscarriage and ectopic pregnancy.
- **Longitudinal vaginal septum** presented with dyspareunia managed by removal of septum.
- **Transverse vaginal septum** presented with primary amenorrhea managed by surgical removal of septum.

Slide

The girl shown in the picture of 16 yers and 140 cm length old age present with delayed onset of menarche

- What is the most likely diagnosis?
- What do you suspect the result of karyotyping?
- Give 4 main other clinical features of the condition.
- How can this patient get pregnant in the future?



Slide

A 14 years old girl present with periodic abdominal pain and delayed onset of puberty, perineal examination showing in this picture.

- What is the diagnosis?
- What are other clinical findings you suspect?
- What are the ultrasonic findings you suspect?
- What is the management?



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