

Hereditary and Congenital Anomalies of Female Reproductive Tract

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Learning Objectives

- ▶ Types of hereditary and congenital anomalies of female reproductive tract
- ▶ Incidences of these anomalies among different breeds
- ▶ Etiology and Prognosis
- ▶ Early diagnosis of these disorders
- ▶ Preventive measures, if any

Types of Reproductive Tract Anomalies

- ▶ A. Ovarian Hypoplasia
- ▶ B. Segmental aplasia (White Heifer Disease)
- ▶ C. Freemartin
- ▶ D. Hermaphrodite

Ovarian Hypoplasia

- ▶ **Definition:** Ovarian hypoplasia is a condition where the ovary undergoes incomplete development and a part or whole of ovary lacks a normal number or compliment of primordial follicles.
- ▶ Normally both ovaries in cattle have 50000 to 100000 primordial follicles but partial hypoplastic heifers have 500 primordial follicles or no follicles.

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- ▶ **Etiology:** Single autosomal recessive gene
 - ▶ **Breed Predisposition:** Polled Swedish Highland Breed with white coat color or at least white ears.
 - ▶ An incidence of 1.9 percent has been reported out of which left ovarian hypoplasia is more common.

Rectal Palpation Findings

- ▶ In heifers, the hypoplastic ovaries are so small to locate.
- ▶ Cord like thickening in the cranial border of the ovarian ligament.
- ▶ Slightly raised and firm like pea
- ▶ Kidney bean with smooth and stretched surface
- ▶ If luteal scars are present, ovary can be considered functional.
- ▶ Ultrasound reveals no follicles.



- ▶ *Tubular Genitalia*

- ▶ In bilateral total hypoplasia the genital tract remains very small and infantile.

- ▶ *General Appearance*

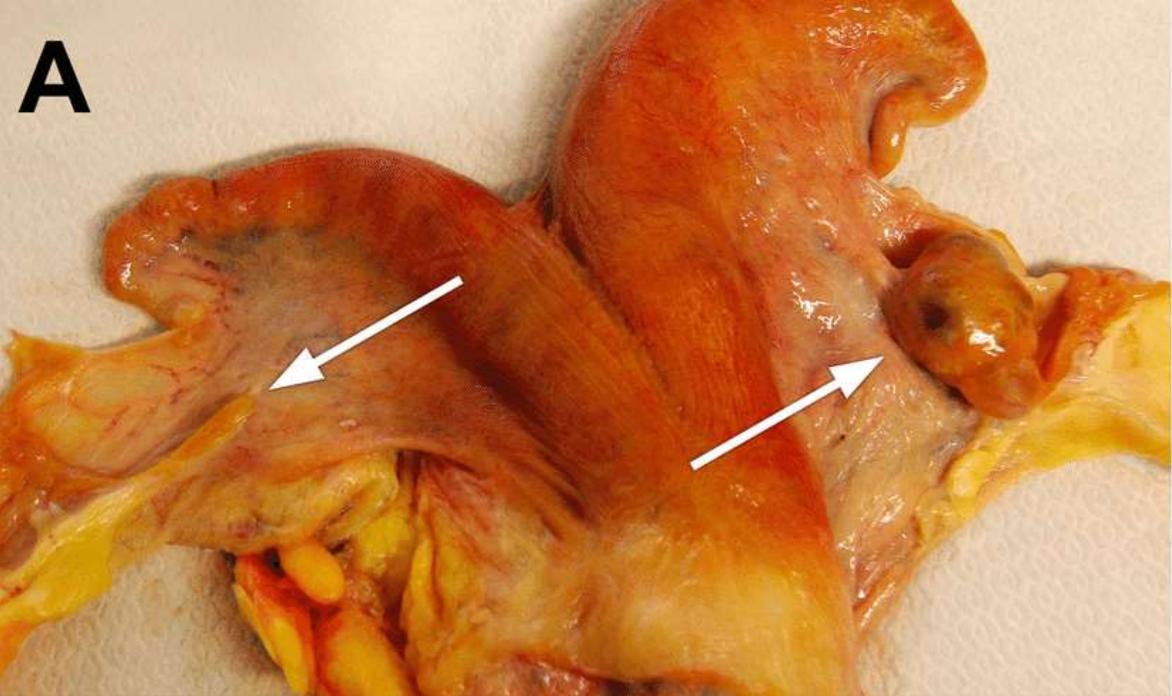
- ▶ Appears like a steer with

- ▶ Long legs

- ▶ Narrow Pelvis

- ▶ Poorly developed udder & teat

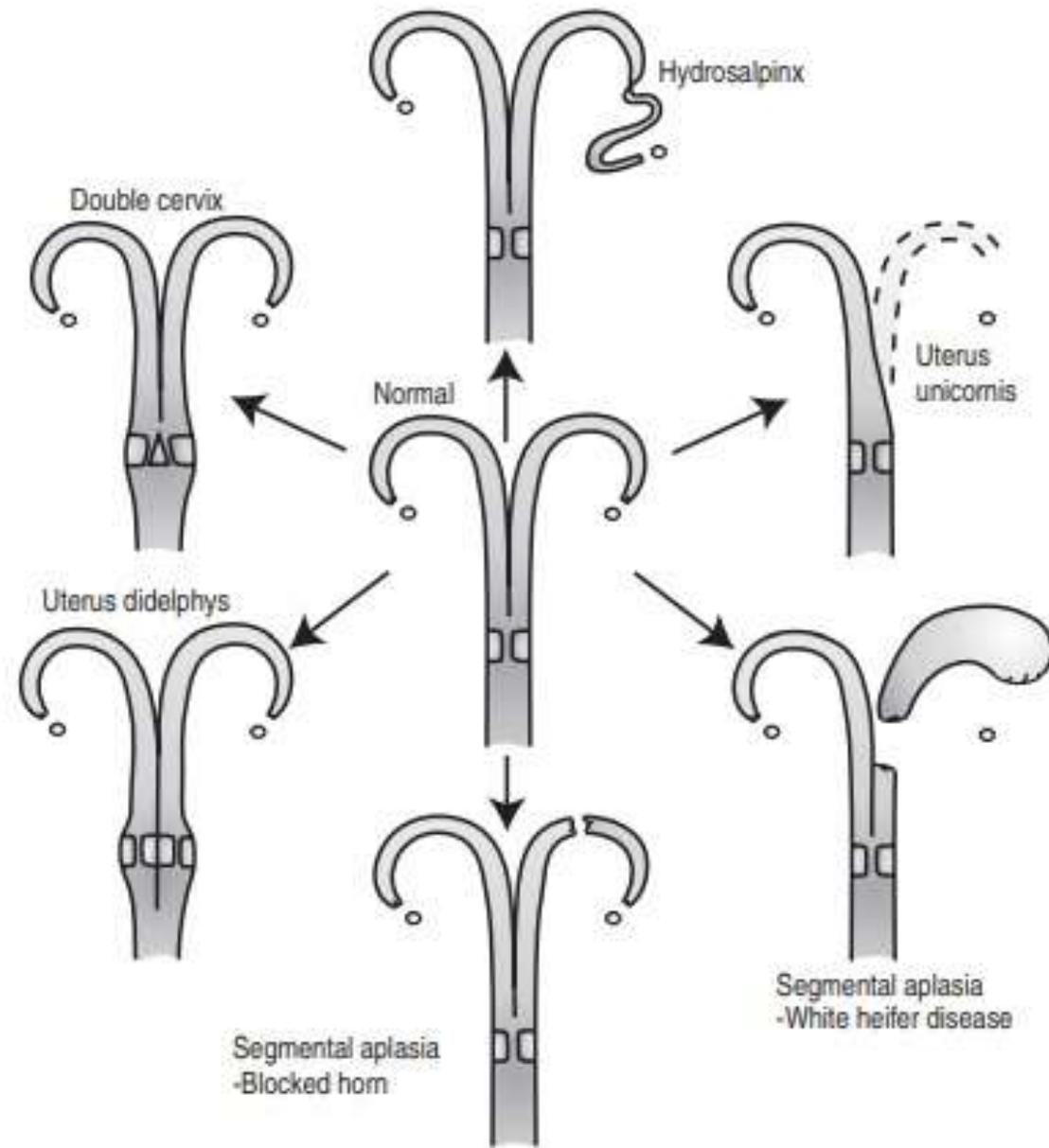
- ▶ Either totally white or at least white ears



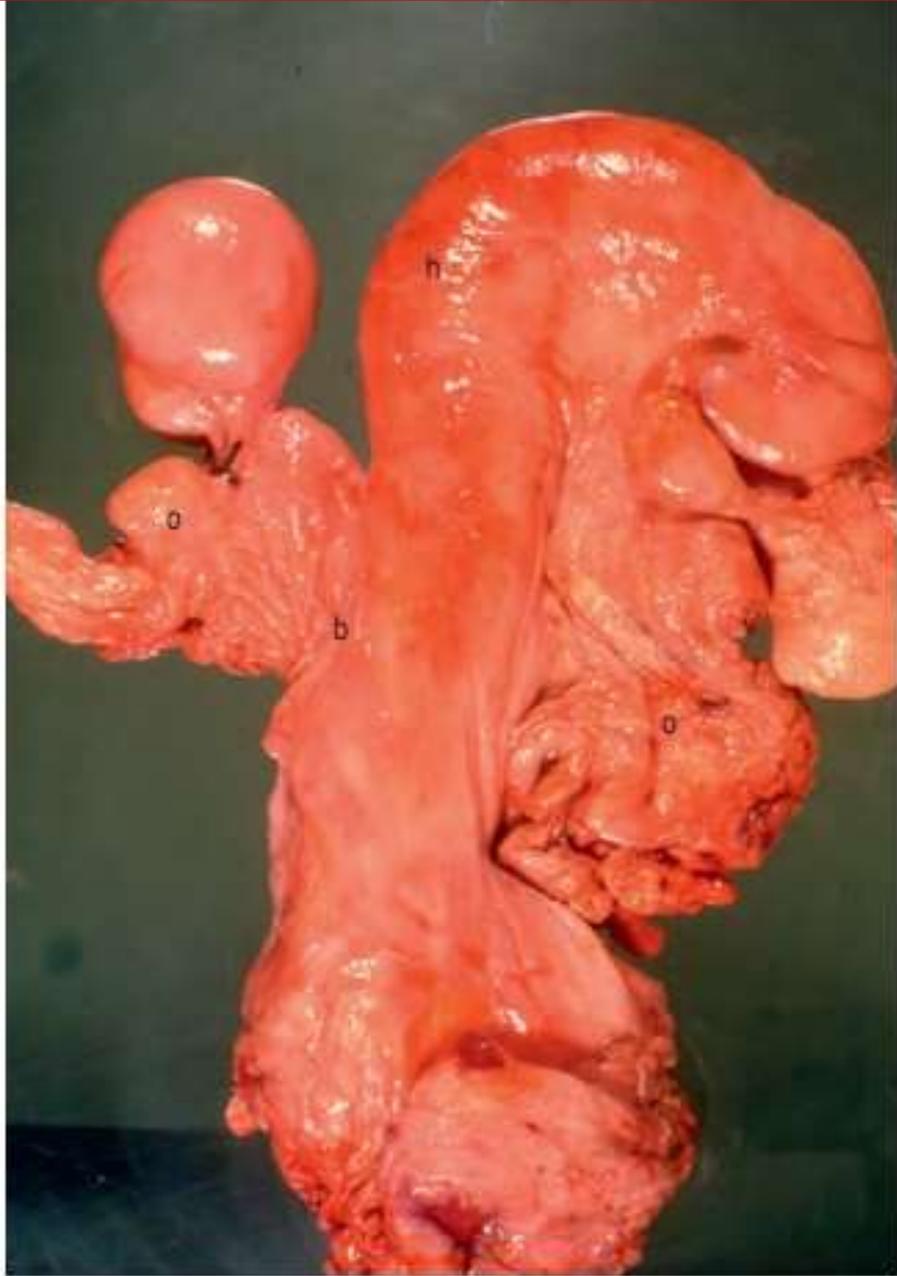
Segmental aplasia (White Heifer Disease)

- ▶ White heifer disease is a congenital defect of the reproductive tract where there is segmental aplasia of the Mullerian or Paramesonephric ducts, especially an imperforate hymen and associated with white coat color.

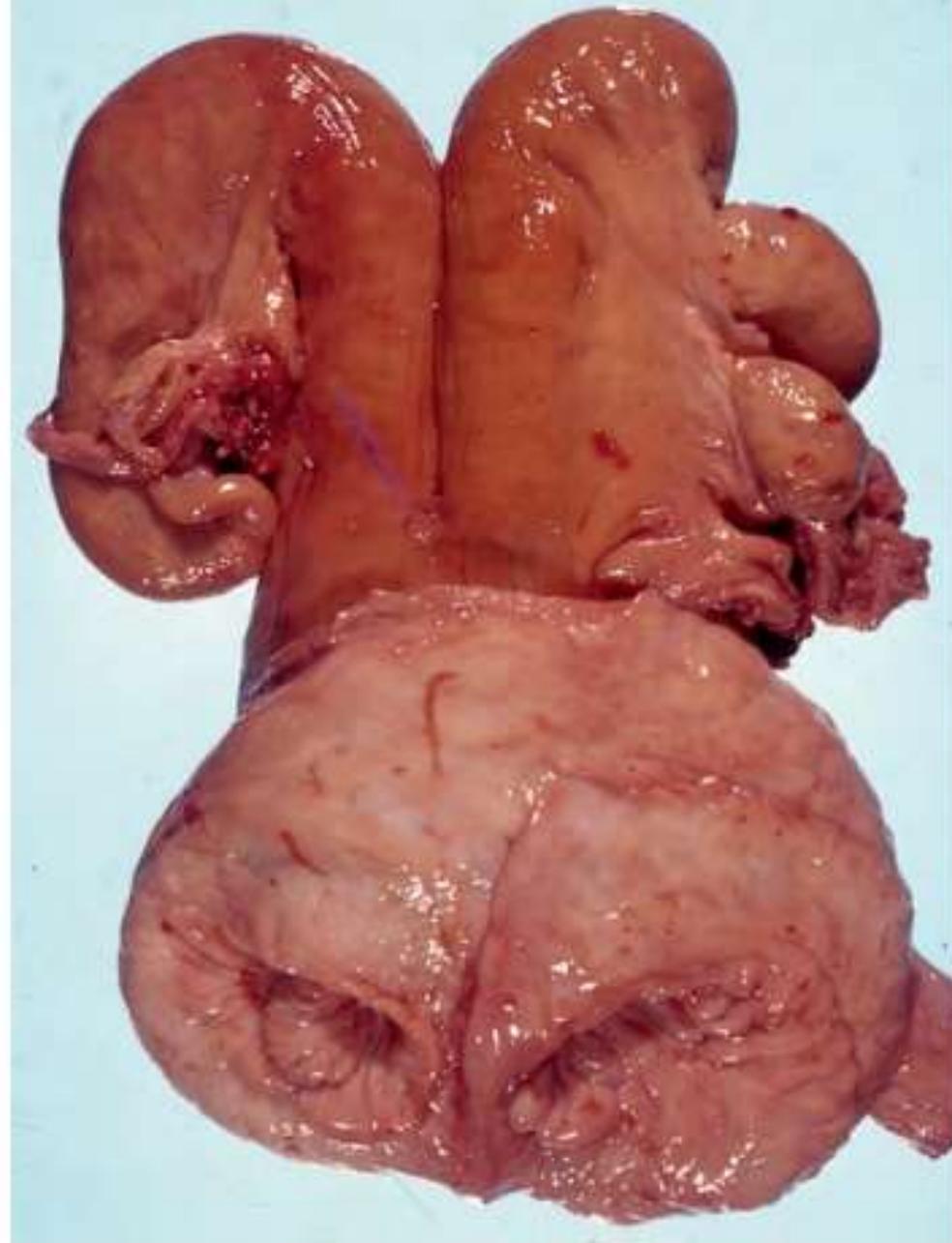
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- ▶ Incidence and Breed Pre-disposition
 - ▶ Reported to occur in 10 % of Shorthorn breed
 - ▶ Reported in Holstein, Jersey, Ayrshire and Guernsey breed
 - ▶ Caused by Single Recessive Sex Limited Gene with linkage to the gene for white coat color



Manifestations of the syndrome of uterine segmental aplasia, sometimes known as 'white heifer disease'.



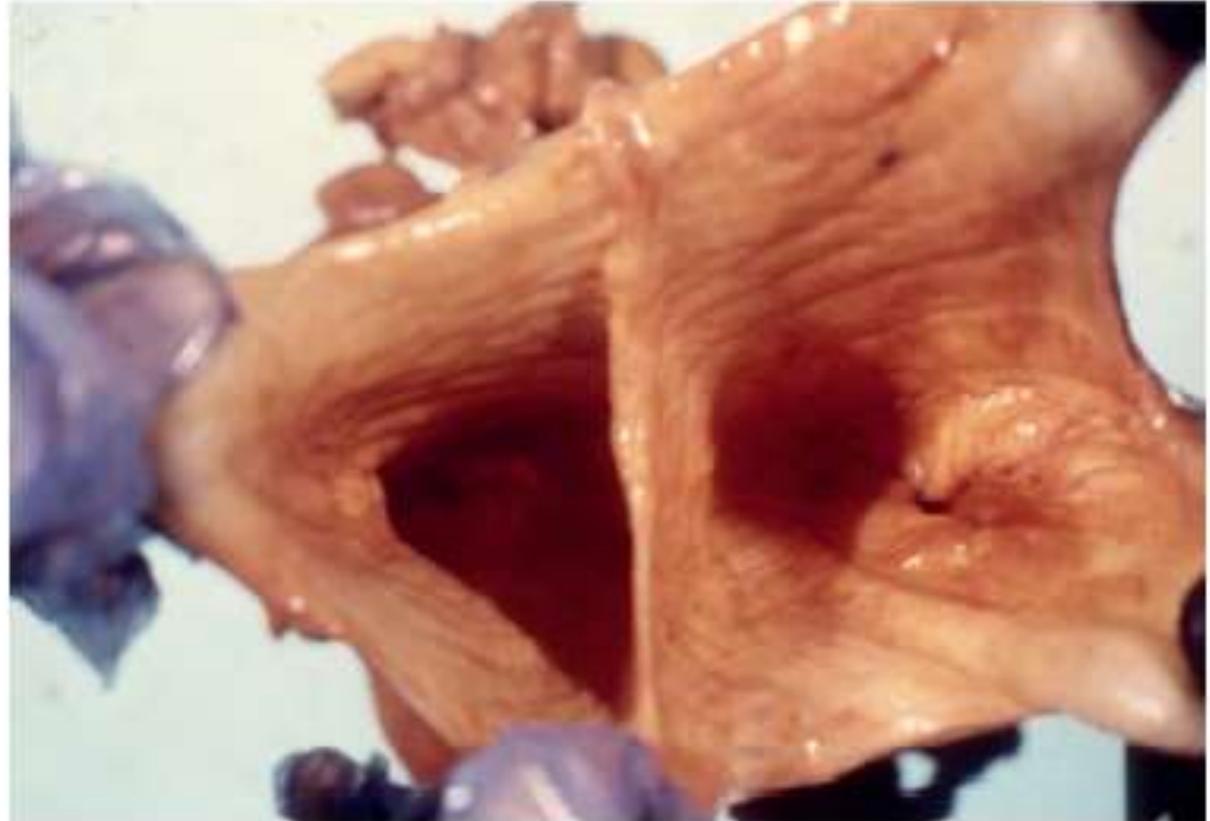
Uterus unicornis. Note normal left and right ovaries (o) and complete right horn (h). The left horn comprises a flat band of tissue with no lumen (b) and a blind residual segment.



Uterus didelphys showing two completely separate cervical canals.



Genital tract from a heifer with 'white heifer disease'. Note normal left ovary (o) and isolated portion of the right horn (h) greatly distended with accumulated fluid.



Double external os cervix showing two completely separate cervical canals.

▶ *Tubular Genitalia*

- ▶ Imperforate hymen or hymenal constriction
- ▶ Absence of either the cranial part of the vagina, cervix or the uterine body
- ▶ Uterus unicornis
- ▶ Cystic dilation of uterine horn due to aplastic body- large enough to simulate a 4 months pregnant uterus!
- ▶ Uterine horn filled with yellowish to tan dark brown mucus
- ▶ Vagina is usually short and narrow or may be enlarged or dilated with mucus or pus
- ▶ Persistence of median wall of the paramesonephric duct- double cervix, median septum in the vagina
- ▶ Rare cases Uterus Didelphys



▶ *Ovaries*

▶ Ovaries are normal and functional in white heifer diseases

▶ *Estrus and Estrus Cycles*

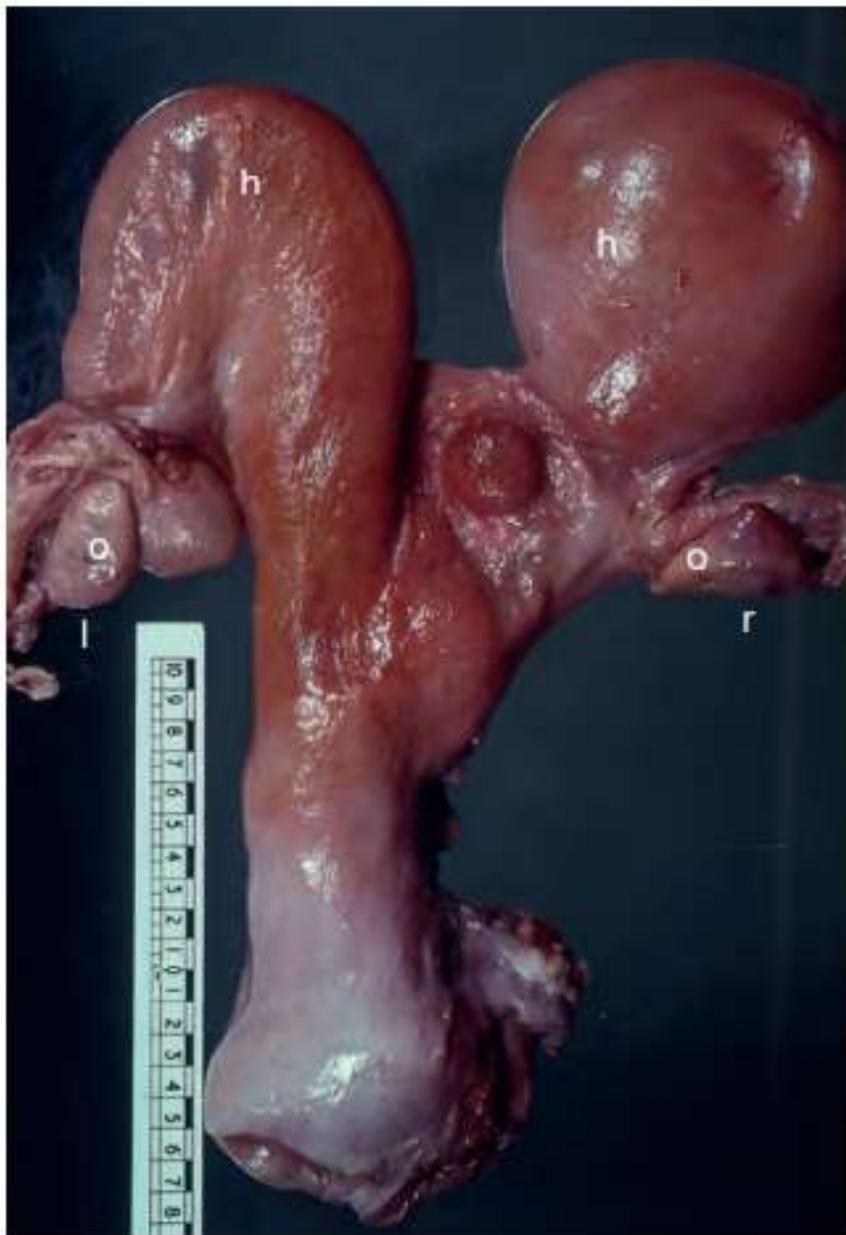
▶ If ovulation occurs in the ovary adjacent to the abnormal horn, regression of corpus luteum may not occur resulting into permanent anestrus

▶ There can be prolonged inter-estrus interval

▶ *Fertility*

▶ The animal can be sterile if the condition is bilateral and affects uterine horn, cervix and oviduct

▶ However cows may conceive in unilaterally affected cases



Genital tract from a heifer with 'white heifer disease'. Note that both ovaries (o) are normal with a corpus luteum present in the right (r), and the uterine horns (h) are distended with accumulated fluid.



Genital tract from a heifer with 'white heifer disease'. Note normal left ovary (o) and isolated portion of the right horn (h) greatly distended with accumulated fluid.

Freemartin

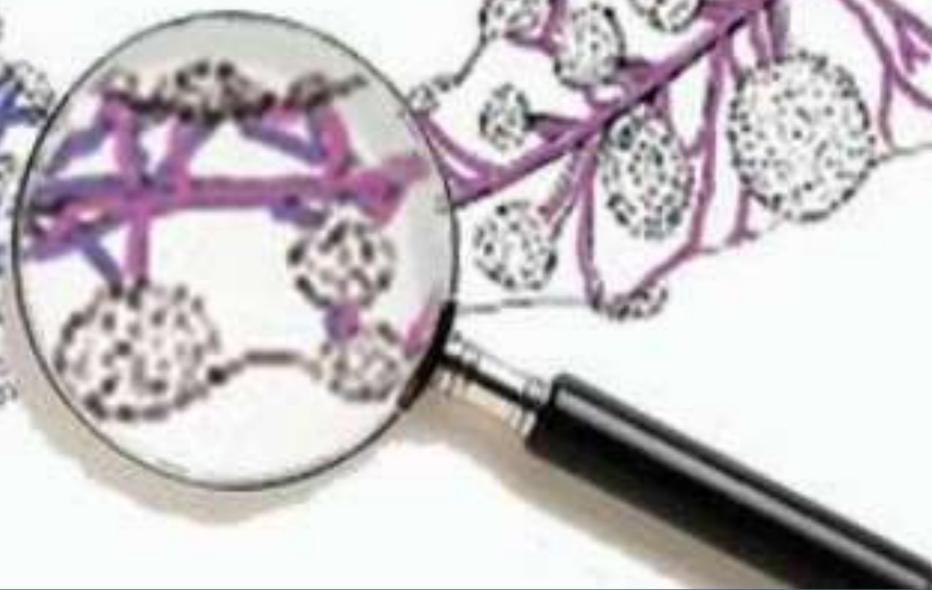
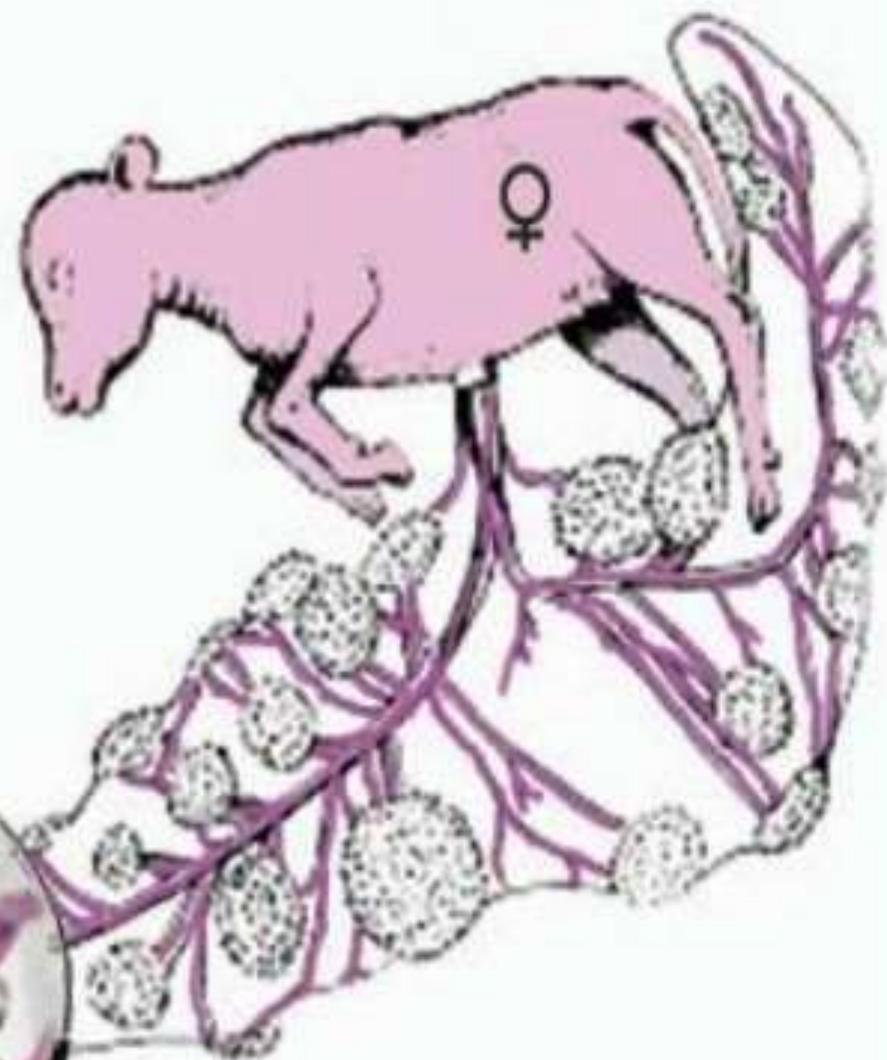
- ▶ A freemartin is a infertile female with a modified genital tract born co-twin, or in greater multiples, with a male with which it has exchanged whole blood.
- ▶ A freemartin is thus a ‘dizygotic twin’.
- ▶ 92% of female co-twins are sterile

Etiology

- ▶ Occurs due to fusion of blastodermic vesicle about 20 days of gestation and by 28 days complete fusion of chorio-allantois and blood vessels
- ▶ Two theories have been put forward for this condition
- ▶ Lille's Humoral Theory
- ▶ Fechheimer *et al.* and Ohno *et al.* Cellular Theory

Lille's Humoral/Hormonal Theory

- ▶ The anastomosis of blood vessels of chorio-allantois of the twin fetuses are complete by day 28 much earlier than sex differentiation that takes place about day 40 to 50 of gestation.
- ▶ Since the development of interstitial cells of the testes in males occur much earlier than the formation of ovaries
- ▶ Androgen or anti-mullerian substance secreted by the interstitial cells reach the female fetus by anastomosing blood vessels in the placenta
- ▶ Inhibits the development of female gonads and genitalia

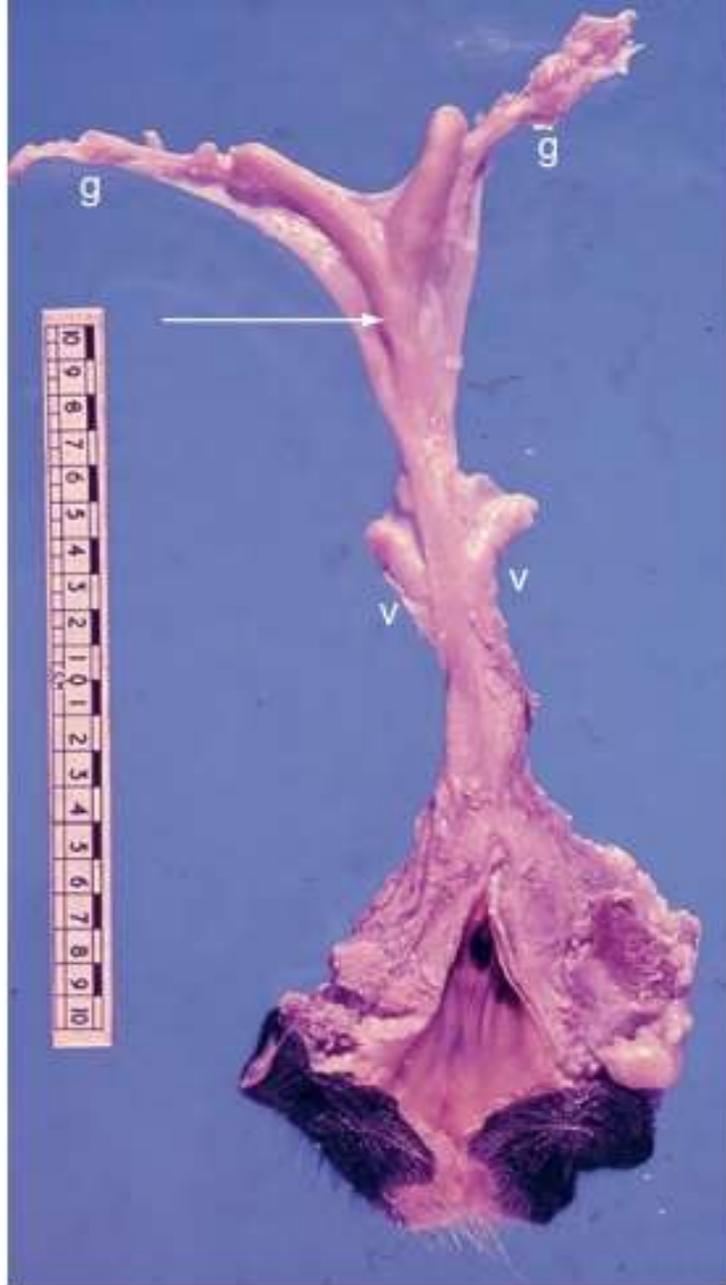




Cellular Theory

- ▶ Anastomosis of the placental vessels precedes migration of the primordial sex cells from the wall of the yolk sac to the gonadal ridge
- ▶ During the migration of primordial germ cells, germ cells from the male fetus pass to the female fetus in early pregnancy resulting in XY cells in the genital ridge and freemartinism.

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- ▶ Ovaries are small, about the size of a flattened barley grain
 - ▶ Rudimentary uterine horns
 - ▶ Lack of cervix
 - ▶ Structure resembling seminal vesicle present in the region of cervix
 - ▶ Vaginal underdeveloped
 - ▶ Vaginal opening not found anterior to the urethral opening
 - ▶ Vulva fairly normal except for the presence of prominent clitoris and tuft of hair



Reproductive tract from a freemartin heifer. Note the vestigial gonads (g), underdeveloped structures derived from the paramesonephric ducts (arrow) and rudimentary vesicular glands (v).



Vulva of a freemartin heifer showing the prominent clitoris and coarse hairs at the ventral commissure.



Normal (left) vs. Freemartin (right) External Genitalia



Normal (left) vs. Freemartin (right) Internal Genitalia

Fincher's Method for diagnosis of Freemartin

- ▶ If 0.5 inch or 1.25 cm wide glass speculum or test tube be inserted through the vulva of a freemartin heifer after proper lubrication it will progress no farther than 7.5 cm to 10 cm or 3 to 4 inches. In normal cows it should pass 12 to 18 cm or 5 to 7 inches
- ▶ The general appearance of a freemartin cow is like a steer with narrow pelvis, long legs and under-developed teats and udder

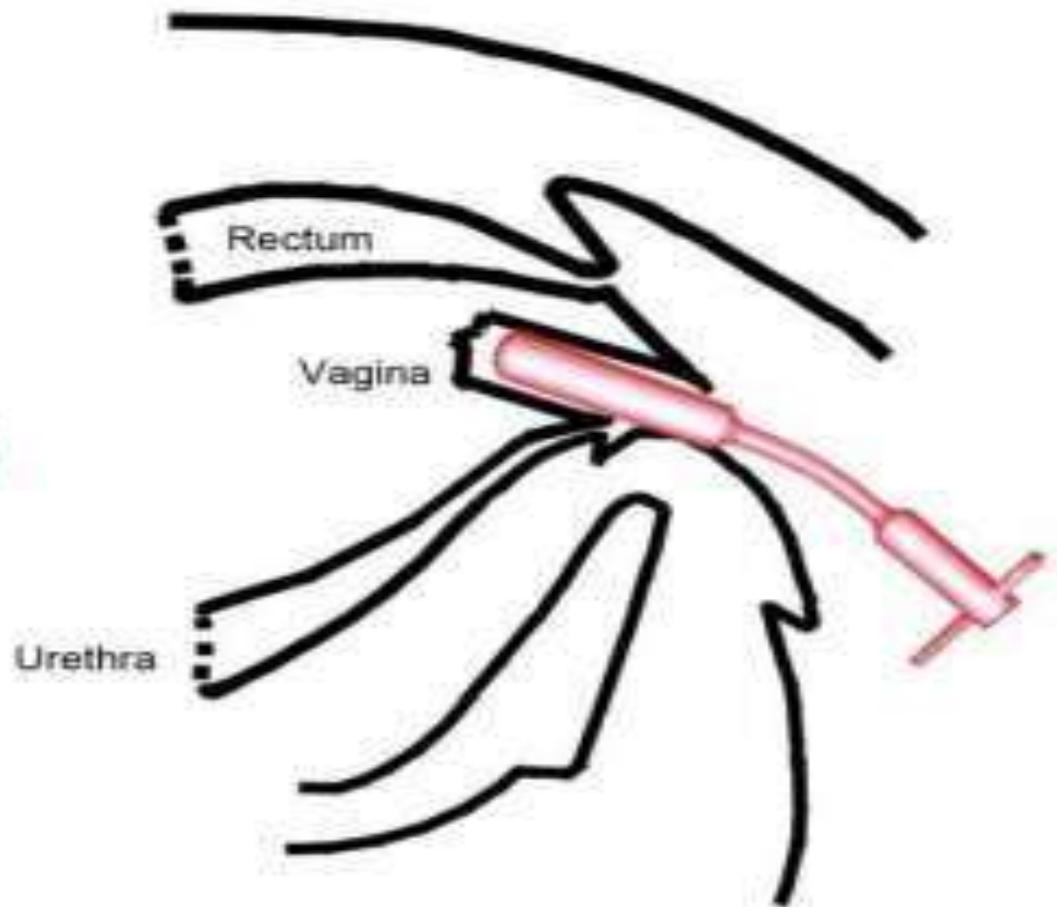
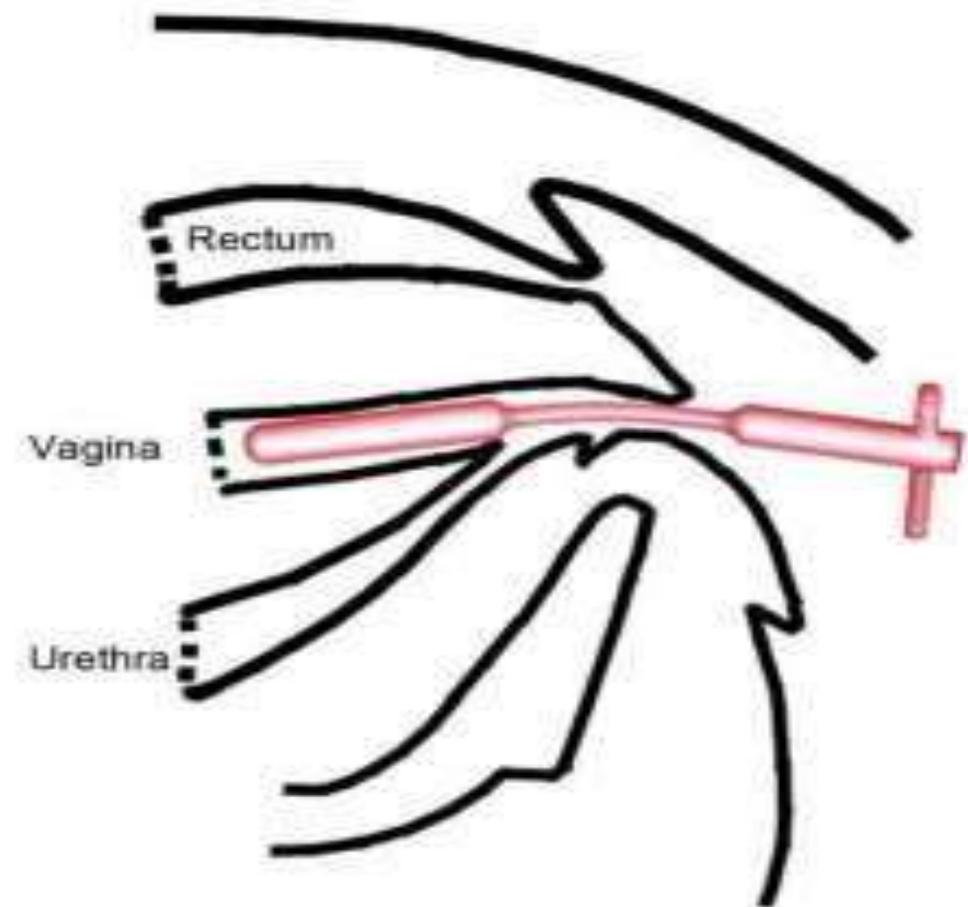
A



B

Normal heifer

Freemartin heifer



Hermaphroditism

- ▶ A hermaphrodite or intersex is an individual in which the diagnosis of the sex is confused due to congenital anatomical variation

Classification of Intersex (Biggers and McFeely)

- ▶ Hermaphrodite
- ▶ Abnormalities of accessory glands
- ▶ Gonadal dysgenesis
- ▶ Freemartin

Hermaphrodites Classified according to Gonadal Sex

True Hermaphrodite

Extremely rare, possess both male and female gonads and external genitalia either resembling male or female. Genetically they are females XX, but may be XX/XY chimera or mosaic

Pseudo-Hermaphrodite

Possess either testes or ovary, more common

Male & female pseudo-hermaphrodites are genetically females with XX/XXXY sex chromosomes

Male

- Very common
- Possess testes and external genitalia resembling female
- Testes often abdominal
- Clitoris is enlarged-Fish Hook Vulva
- Uterus is always present
- Nymphomaniac character

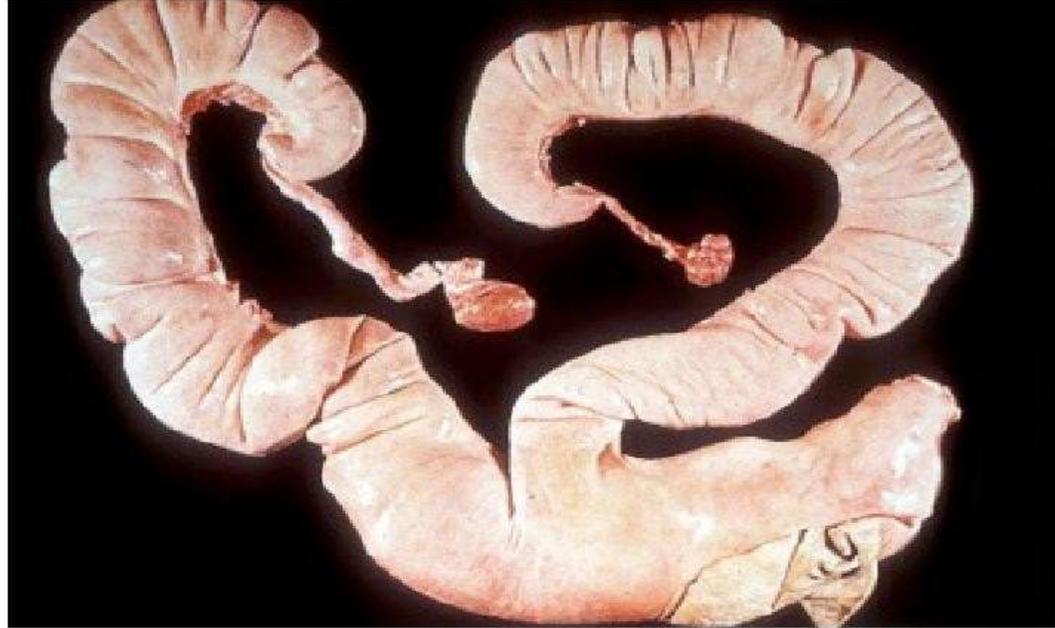
Female

Possess ovary and external genitalia of male type, extremely rare

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- ▶ Incidence and Species Predisposition
 - ▶ Common in Swine and Goats- an incidence of 6-10 percent has been reported in Saanen goats
 - ▶ Rare in Cattle and Sheep



Gilt, (unilateral) true hermaphrodite.
Ovotestis on the left, testis on the right;
well developed uterus, cervix, and vagina



Gilt, (lateral) true hermaphrodite



True Hermaphrodite.

The left gonad is a combination of testicular and ovarian tissue. The right gonad consists primarily of testicular tissue. Both ovotestes are accompanied by a uterus