CVE – AGMOOCS - TANUVAS

LESSON IV- MISCELLANEOUS HEREDITARY FORMS OF INFERTILITY

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FREEMARTINISM

- Freemartinism is a distinct form of **intersexuality due to vascular anastomosis of the adjacent chorioallantoic sacs of heterozygous fetuses in twin pregnancies.**
- The majority (~95%) of female fetuses in male/female twin pregnancies are affected.
- Vascular anastomosis **occurs as early as 30 days of gestation**
- Occurs when the blastodermic vesicles from each developing zygote meet and fuse in the uterus about day 18 to 20 of gestation

FREEMARTINISM

- It is possible for singleborn freemartins to occur if there is death of the male twin of a heterozygous pair after the time of vascular fusion with the other being carried to term.
- This has been demonstrated as a cause of infertility in heifers with apparently normal external genitalia but with **sex chromosome chimerism**.
- The external genitalia of freemartin heifers may appear relatively normal
- The clitoris is characteristically enlarged and prominent, and coarse hairs are present at the ventral commissure of the vulva.
- The internal genitalia are grossly abnormal.
- The **gonads are typically vestigial**, although they undergo masculinization in mild cases

FREEMARTINISM

- The structures derived from the paramesonephric ducts are almost **entirely absent or are grossly hypoplastic.**
- In animals with a significant degree of masculinization, **the gonads resemble testes to the extent that their parenchyma contains recognizable tubules and interstitial tissue.**
- Development of the mesonephric (Wolffian) ducts is related to the degree of masculinisation of the gonad.
- In extreme cases- well developed epididymides, vas deferentia, and vesicular glands.
- More typically, **the vestigial gonads of freemartins are devoid of oocytes and follicles and have parenchyma that consists largely of degenerating sex cords**.

THEORIES OF FREEMARTINS

1. Lillie/Humoral theory

- **Interstitial cells of the testes of the male** developed earlier than the female ovaries and secreted an **androgen** that reached the female embryo by way of the **anastomosing blood vessels in the placenta and this inhibited the development of the female gonads and genital tract**
- **H-Y antigen of the male fetus** governs the development of initially indifferent gonads of the female twin toward the male mode
- Subsequent **androgen and anti-mullerian hormone produced by the male gonads-** suppress the development of the uterine tubes, uterus and cephalic portion of the vagina

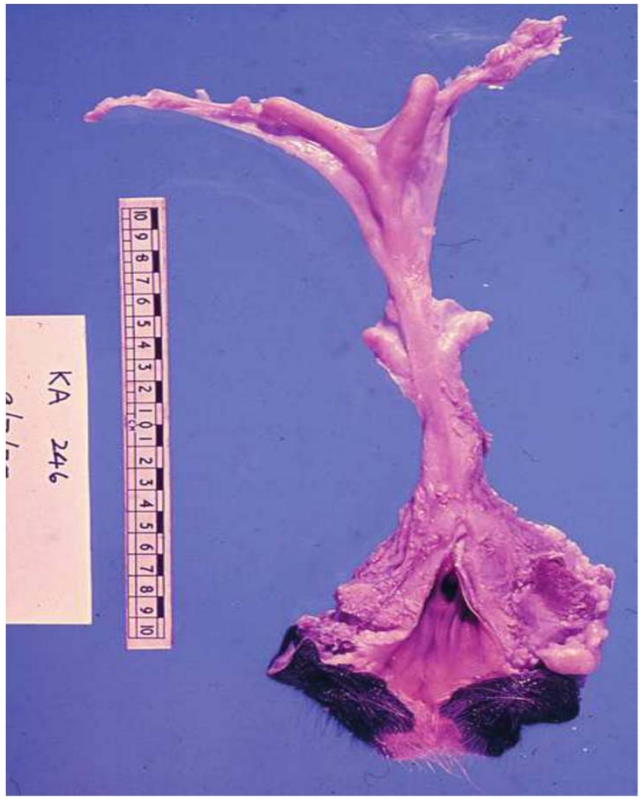
2. Cellular theory

- **Migration of primordial germ cells from male fetus to female fetus-** inhibited the development of the female gonads and genital tract.

CLINICAL SIGNS

- The newborn freemartin - **prominent clitoris and tuft of hair at the ventral commissure of the vulva** but these signs are not always reliable.
- Freemartins can be identified on the **basis of the length of the vagina and the absence of the cervix.**
- In the adult, the vagina is normally 30 cm in length compared with 8 to 10 cm in the freemartin female.
- Rectal palpation will fail to identify the cervix.
- In calves of 1 to 4 weeks of age, the vagina is normally 13 to 15 cm in length compared with 5 to 6 cm in a freemartin.
- Diagnosis at this age can be made using **Fincher's test-** a **blunt probe, which should be inserted initially at an angle of 45 degrees below the horizontal for 5 cm and then angled downwards to avoid impinging on the hymen**

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



- Reproductive tract from a freemartin heifer.
- Note the vestigial gonads, underdeveloped structures.

DIAGNOSIS

- Most accurate method of diagnosis- the **demonstration of sex chromosome chimerism in cultured lymphocytes.**
- Unfortunately, the distribution of male cell percentages in freemartins appears to be random.
- Thus animals with low male percentages in the blood are as common as those with high male percentages.
- Diagnosis by a **polymerase chain reaction (PCR) test for the presence of Y chromosomes in blood cells or hair**
- This is a definitive test for the presence of the Y chromosome

INTERSEXES OR HERMAPHRODITES

- Intersexes or hermaphrodites occur **most commonly in goats and pigs, and less commonly in horses and dogs.**
- It is **occasionally present in sheep and cattle and is rare in the cat.**
- Intersexes are **individuals in which the diagnosis of the sex is confused because of congenital anatomical variations and abnormalities of the genital organs**.
- Conditions in intersexes include hermaphroditism, abnormalities of the accessory genital organs, gonadal dysgenesis, and freemartinism.
- Other forms of intersexuality - **pseudohermaphroditism** have been reported, as have rare cases of **XY sex reversal and true hermaphroditism**
- Intersexes or hermaphrodites are commonly observed in naturally polled goats.
- These intersexes are genetic females and although the polled condition is dominant the **hermaphroditic defect is recessive, sex-limited and incompletely penetrant**.

INTERSEXES OR HERMAPHRODITES

- Most of these hermaphrodites are **phenotypic females most with ovotestes and many exhibit a projecting vulva and enlarged clitoris**.
- The **more masculine phenotypes have a penile-like clitoris, hypospadias and hypoplastic testes often in the inguinal region**.
- Variable development or lack of development of the mesonephric and paramesonephric ducts are seen at autopsy.
- The **vagina** in these hermaphrodites is **very hypoplastic and a small glass rod will only penetrate about an inch past the vulva**.

SUMMARY OF DAY 4 LECTURE

- Freemartinism
- Theories of Freemartins
- Clinical Signs and Diagnosis
- Intersexes or Hermaphrodites