

Presentation on
TERATOLOGY
Veterinary Obstetrics (VGO- 421)

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LEARNING OBJECTIVES



After going through this module on
Teratology

The learner will be able to:

1. Explain the inherited lethal and semi lethal characters in domestic animals.
2. Describe the inherited and generally non lethal defects in domestic animals.
3. Explain the teratogenic agents.
4. Discuss the anomalies of various systems in domestic animals.
5. Recognize embryonic duplication, hermaphroditism and freemartin.

TERMINOLOGY

Teratology

- It is the division of embryology and pathology dealing with the abnormal development and malformations of the antenatal individual.

•Definitions

Mosaicism

- The occurrence in an individual of two or more cell populations or tissues each with a different chromosome complement derived from a single zygote.

Chimerism

- The occurrence in an individual of two or more cell populations or tissues each with a different chromosome complement derived from different zygotes, as in twins with placental anastomoses.

Pleiotropism

- It refers to certain harmful traits spread widely if they are associated with desirable traits.

Anomaly

- It refers to the malformation involving only an organ or part of the body.

Monster

- It refers to an animal with extensive deformity.

Phenocopies

- It refers to non-genetic anomalies that are similar to genetic anomalies.

Teratogens

- Variety of environmental factors or agents causing non-genetic anomalies or monsters.

INHERITED LETHAL AND SEMI LETHAL CHARACTERS IN CATTLE

Achondroplasia, or dwarf, “comprest” or “bull dog calves”

- All breeds but most commonly in the Hereford, Ayrshire, Angus and Dexter breeds.
- Most common type is, the brachycephalic “snorter” dwarf in Herefords with a short, broad head, bulging forehead, malocclusion of the jaw, prognathism of mandible, pot-belly, low viability and great susceptibility to bloat and dystocia.
- This type was generally considered to be due to a simple autosomal recessive defect with some modifiers.
- The “comprest” Hereford is the result of incomplete dominance.
- In Ayrshire, Dexter and other cattle, extreme “bulldog” calves is usually aborted about the fifth to eighth month of gestation.
- Hydramnios occurs in pregnant Dexter cattle carrying a “bulldog” calf. Normal Dexter cattle are heterozygotes.



Achondroplasia



Extreme Achondroplasia

Courtesy: Drost Project

Epitheliogenesis imperfecta

- It is a condition where skin fails to form.
- It occurs most commonly on the legs below the knees and hocks and on the muzzle, ears, tongue and mucous membranes.
- It has been described in Holsteins, Ayrshires, Jerseys, Brown Swiss and Shorthorns.



Epitheliogenesis Imperfecta



Epitheliogenesis Imperfecta-Closeup

- **Hypotrichosis congenita** or alopecia is a recessive defect characterized by degrees of hairlessness in Holsteins, Polled Herefords. It was also described in Swedish Friesian cattle, and Holsteins; in Jerseys having other defects such as a short lower jaw, short ears and anophthalmia. Streaked hairlessness reported as a sex-linked lethal in Holsteins.
- **Ichthyosia congenita** is characterized by a lack of hair and a thick scaly, horny epidermis with raw fissured skin around the body orifices. It is due to single autosomal recessive genes in Brown Swiss and Red Polled Cattle.
- **Acroteriasis congenita** or amelia and hemimelia is seen in Holsteins and Brown Swiss and other breeds. This is characterized by missing, shortened, deformed, or “amputated” limbs.
- **Cerebellar hypoplasia** and degeneration is seen in Herefords, Guernseys and Holsteins and is probably autosomal recessive in nature. This conclusion may be erroneous as BVD-MD virus can produce this defect in fetuses.

- **Paralyzed hind quarters** have been reported in Red Danish calves at birth due to a pair of autosomal recessive genes.
- **Curved limbs** with both rear and forelimbs curved anteriorly, have been observed in Guernseys as an autosomal recessive trait. Calves are usually stillborn or die promptly.
- **Muscle contractures and ankyloses**, or arthrogryposis has been reported as a recessive in Dole cattle in Norway; and a dominant with incomplete penetrance in England.
- **Hydrocephalus** in Herefords, Ayrshires, Holsteins and other breeds. It is characterized by the birth of “dummy” or “bawler” calves that are unable to nurse properly and die in several days. The heads may be enlarged or normal in size but section of the head and brain reveals distended ventricles.



External hydrocephalus



Internal hydrocephalus

INHERITED AND GENERALLY NONLETHAL DEFECTS IN CATTLE

- **Polydactylysm** in Holsteins and Herefords is possibly an autosomal dominant character with incomplete penetrance.
- **Syndactylysm** or “mule-foot” affecting one or both front feet or all four feet has been reported in Jerseys, Haryana and Holsteins and has a single autosomal recessive mode of inheritance. Affected Holsteins were unable to withstand stress due to high ambient temperatures and became hyperthermic. The front feet of the Holstein calves were much more frequently affected with syndactyly than the rear feet.
- **Muscular hypertrophy or “double” muscling** is characterized by reduced fat deposits, light bone, thin skin, and large muscles. It is seen in many breeds of cattle including Herefords, Holstein, Angus, Charolais. In the latter it may be inherited as an incomplete dominant while in the former breeds it may be a recessive with incomplete penetrance and variable expression. When fetuses are affected dystocia often occurs.

Double Muscling in Cattle



- ***Umbilical hernia*** was described as being a probable sex-limited dominant character in male Holsteins but the mode of inheritance in females was uncertain. Umbilical hernia in Holsteins was caused by one or more pairs of autosomal recessive genes of low frequency. The condition was seen more often in females but was probably not sex-linked.
- ***Osteoarthritis and hip dysplasia*** in Holsteins, Jerseys and Herefords. In dairy breeds the lesions were noted in the stifle joint at 5 to 13 years of age and were possibly due to a single autosomal recessive gene. In yearling Herefords the lesions were in the hip joint and it was possibly due to an incompletely penetrant dominant character.
- ***Spastic paresis*** in Angus, Holstein, Charolais, Shorthorn, Ayrshire, Simmental and rarely Jersey calves usually 2 to 8 months of age with straight rear legs and a contracted gastrocnemius muscle causing the rear leg to be held off the ground and swung rigidly. It is due to an autosomal recessive condition.
- ***Spastic syndrome***, Krampfigkeit, “Stretches” is a latent recessive condition developing at 2 to 7 years of age and characterized by spastic contractions of the rear limb or limbs and back that occur intermittently in the standing animal. The condition affects Holsteins, Guernseys, Charolais, Ayrshires, and occasionally other breeds.

- *Vestigial tail* (Holsteins, Angus, Shorthorns).
- *Fused teats* (Guernseys, Herefords).
- *Supernumerary teats* (all breeds).
- *Notched* or short ears : Ayrshires, Jerseys.
- *Missing phalanges* or “Creeper” calves (Swedish cattle).
- *Impacted premolars* and “parrot-mouth” (Shorthorns) Short spine (Norwegian cattle).
- *Agnathia* or *absence of a lower jaw* (Jerseys and other breeds) Opacity of the cornea is probably a recessive character in Holsteins.
- *Prognathism* (Herefords and others).
- *Dermoid cysts* on cornea, (Herefords and Guernseys).

Non-Genetic Teratological Defects

- In domestic animals, there are innumerable types and degrees of non-genetic anomalies, or monsters.
- **Anomaly** refers to malformation of only an organ or part of the body.
- **Monster** refers to an extensive deformity is extensive.
- Suspect genetic role, if a similar defect appears quite frequently in related individuals or those tracing back to a common ancestor. It would be impossible to differentiate some of these defects appearing in families or related animals in a herd due to environmental causes without a carefully controlled experiment.

Susceptibility

- The period of early differentiation in the embryo or about the time germ layers and organs are rapidly developing - Highly susceptible.
- The zygote is not as susceptible to teratogens during the period of the ovum or blastula or the period of the fetus as it is during the period of the embryo and organogenesis, especially the first half of that period.

TERATOGENIC AGENTS OR FACTORS

Nutritional deficiencies in the dam

- Vitamin A and E, riboflavin, folic acid, pantothenic acid, niacin and other vitamin deficiencies, minerals such as iodine and possibly manganese, and amino acids such as tryptophane may cause congenital defects. Hypervitaminoses A and D will also cause anomalies.

Endocrine disturbances of the dam

- Diabetes, thyroid malfunction, and large exogenous doses of glucocorticoids, ACTH, insulin, androgens, progestagens, estrogens, thyroxine and thiouracil will cause defects of the embryo. Large doses of glucocorticoids in pregnant animals at the proper stage of gestation may cause cheilo or palatoschisis. Progestagens given during pregnancy may cause masculinization of the genitalia of female fetuses.

Physical factors

- Reduced atmospheric pressures, hypothermia, hyperthermia and, anoxia cause anomalies.

Radiation

- X-ray or radioactive substances induces congenital defects.

Drugs or chemicals

- Thalidomide, quinine, sulphonamides, tetracycline, streptomycin, salvarsan, lead, mercury, nicotine, malathion, carbon tetrachloride, apholate, selenium, fluorine, cytotoxic agents including aminopterin in sheep, nitrogen mustard, actinomycin D, 6 mercaptopurine, azoserine, azo dye, trypan blue and other dyes, salicylates, histamines, ergot, "Diamox," reserpine, phenylmercuric acid, galactose, E.D.T.A. and certain plant compounds as in Veratrum californicum and locoweeds possibly containing lathyrogens, all produce fetal anomalies in animals under certain conditions.

Infections

- Blue tongue in sheep, hog cholera in swine, feline panleucopenia in cats, bovine virus diarrhea- mucosal disease virus, and toxoplasma can cause anomalies in the embryo.

Ageing of ova

- By delaying ovulation 24-48 h was characterized by a three-fold increase in chromosomal anomalies with a higher incidence of embryonic death in rats. Similarly ageing of rabbit spermatozoa before permitting them to fertilize eggs resulted in normal fertilization but greater embryonic death losses. Thus age affects the genes and chromosomes as does other agents.

MALFORMATIONS DUE TO ALTERATIONS IN TISSUE DIFFERENTIATION THAT ARISE FROM A SINGLE AREA IN THE EMBRYONIC DISC

Most of these are due to the local arrest in the normal process of tissue development and produce

- Defects due to excessive division: polydactylia, polythelia, polydontia.
- Defects due to failure of structures to fuse normally: palatoschisis, cheiloschisis, cranioschisis, spina bifida, and schistosomus.
- Defects due to arrest in division: cyclopia and syndactyly.
- Defects due to complete local failure of tissue growth: amelia or lack of limbs; ectrodactyly or absence of phalanges; epitheliogenesis imperfecta; agnathia; and anophthalmia.
- Defects due to arrest in assumption of final form or position: ectopia cordis, hypognathia, and dextro-position of the aorta.
- Defects in the persistence and disappearance of contiguous structures that normally follow a certain pattern: the aortic arches, foramen ovale, urachus, ductus arteriosus and persistence of the median wall of the paramesonephric or Mullerian duct.
- Defects due to overdevelopment of local tissues: polycystic kidneys; tumors such as sarcomas, hemangiomas and teratomas.
- Defects due to displacement of tissue: teratomas and dermoid cysts
- Defects due to fusion of sexual characters: true hermaphrodites, false hermaphrodites, and freemartins.
- Defects due to miscellaneous causes: ichthyosis, chondrodystrophies, osteogenesis imperfecta, and porphyria.

ANOMALIES OF THE ORGANS OR TISSUES INVOLVED

- Heart and blood vessels
- Digestive and alimentary system
- Urinary system
- Sexual organs and gonads
- Head and central nervous system
- Skeleton
- Trunk
- Limbs

ANOMALIES OF HEART AND BLOOD VESSELS IN DOMESTIC ANIMALS

- Atresia of the aorta with persistence of the ductus arteriosus
- Cor trilobulare batrium
- Tetratology of Fallot
- Ectopia cordis, either cervical, pectoral or abdominal
- Persistence of the right aortic arch with absence of the left causing stenosis of the esophagus, and
- Various types of acardiac monsters.

MAMMARY GLAND ANOMALIES IN DOMESTIC ANIMALS

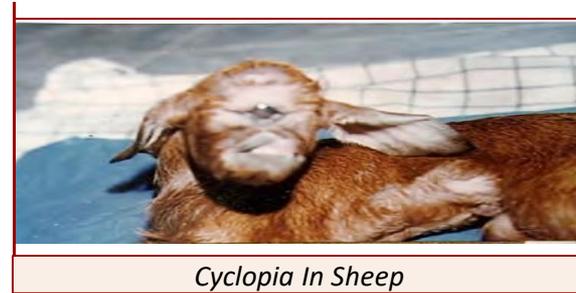
- Fused teats and supernumerary glands and teats.
- Congenital atresia, or stenosis of one or more teats or glands
- A rare case of total mammary gland aplasia has been reported in a fertile COW



HEAD AND CENTRAL NERVOUS SYSTEM ANOMALIES

Anomalies

- Microcephalus
- Cyclopia or Cebocephalus
- Hydrocephalus
- Meningocele



Microcephalus-Description

- Characterized by a small cranial cavity and brain.

Cyclopia or cebocephalus

- Cyclopia is seen most commonly in the pig and, sheep but may be present in all species.
- Characterized by a single orbit in which global tissue is absent or rudimentary or which the eyeballs vary from a single apparently normal eye through all degrees of doubling to one consisting of two complete but small adjacent globes.
- Eyelids are rudimentary or absent and the nose is usually absent or in the form of a tubular appendage placed above the centrally located eye.
- This rudimentary nose does not communicate with the pharynx.
- The skull is usually small and the lower jaw, being longer than the defective upper jaw, is curved dorsally at its cranial end.

Hydrocephalus

- Occurs due to an abnormal accumulation of fluid in the cranial cavity.
- Internal hydrocephalus is due to excessive fluid in the ventricular system.
- External hydrocephalus is rare and due to excessive fluid between the brain and dura mater.
- Internal and external hydrocephalus may be combined.
- In marked cases, the dorsal portion of the brain is encompassed only by the skin, the subcutaneous tissues, and the brain coverings.



Hydrocephalus In Buffalo



Meningocele In Buffalo

Meningocele

Cranioschisis or crania bifida

- Defects of the skull that may result in meningoencephalocele or meningocele .
- In the later instance the skull defect is large, so that parts of the brain and the meninges are extruded.

ANOMALIES OF THE HEAD

Facial fissures

- Palatoschisis, cheiloschisis, macrostomia, and schistoprosopus or divided face.

Lack of cranial or facial structures

- Acephalia, or absence of a head; acrania; hemicrania; astomia- absence of mouth; microstomia, arhinencephalia or absence of a nose, agnathis, hypognathia, anophthalmus, microphthalmia, aprosopia or partial or complete absence of a face, anotia or absence of ears, polyotia and polydontia or anodontia

Crooked calf disease

- Characterized by arthrogryposis (contracture or ankylosis of one or more joints), torticollis, scoliosis (side ways curvature of the spine) and occasionally cleft palate.

These anomalies of the central nervous system are often characterized externally by ankylosed joints, deformed fetlocks or “club feet” and other defects including-hydramnios in some severely affected fetuses. As with other markedly defective fetuses their size is usually smaller than normal.

SKELETAL ANOMALIES

Achondroplasia, Chondrodystrophy or Dwarfism

- Several types and degrees and most, or all, are hereditary.
- In achondroplastic fetuses the long bones are abnormally short. The trunk is of normal length and the abdomen is large. The head is moderately enlarged and flattened.
- “Bulldog” head and brachygnathism of the maxilla, “undershot,” or prognathism of the mandible, “overshot,” are common.

Spina bifida or rachischisis

- Absence of the dorsal portions of the vertebrae or vertebral arches often in the lumbar or sacral region with defective rear limbs and tail and paralysis of the rear parts.
- Observed in Angus, Holsteins, Bull terriers and other animals.

Taillessness, anury or Perosomus acaudus

- Common defect in dairy and beef cattle.
- Characterized by a lack of coccygeal vertebrae and deformed sacral vertebrae with a sunken perineal region and in some cases a characteristic hopping gait.

Hemivertebrae

- Most commonly in English bulldogs and Boston terriers
- Characterized by failure of the centers of ossification to unite.
- Scoliosis, kinked tails and a shortened spine are present if many vertebrae are involved.

SCHISTOSOMUS REFLEXUS

Condition

- *Schistosomus reflexus* is seen most commonly in cattle, but in rare cases may be observed in sheep, goats, and swine.
- It is marked by marked ventral curvature of the spine so the occiput of the head lies near the sacrum.
- The body and the chest walls are bent laterally and the thoracic and the abdominal viscera are exposed.
- The pelvis is deformed.
- The liver is abnormal in shape and cystic.
- The rumen is occasionally distended with fluid.
- The limbs are usually ankylosed and rigid.
- In rare cases the limbs and head may be enclosed in a complete sac of skin.



Schistosomus reflexus In Bovine

CAMPYLORRACHIS SCOLIOSA

Condition

- *Campylorrhachis scoliosa* is a fetal monster, rarely seen in cattle and swine.

Description

- It is characterized by a lateral curvature of the spine.
- The limbs are usually deformed and ankylosed.

PEROSOMUS ELUMBIS

Condition



Source: Balasubramanian et.al., (1991). Indian Vet. J. 68: July 677-678

Description

- Seen occasionally in cattle and swine.
- It is characterized by a lack of vertebrae and spinal cord caudal to the thoracic region.
- The monster has a small, flattened, deformed pelvis with strongly ankylosed and flexed hind limbs and atrophy of the muscles of the rear quarters.

PEROSOMUS HORRIDUS

Condition



Caprine Perosomus Horridus

Description

- *Perosomus horridus* is a bovine fetal monster with general ankylosis and muscle contractures.
- This is due to a marked double S-shaped lateral twisting of the vertebrae.
- It is characterized on external examination by a short spine.

LIMB ANOMALIES

- Include amelia, missing or “amputated” extremities.



Amputated Limbs in a Buffalo Calf

- Micromelia; hemimelia, or absence of the distal half of the limb; hypoplasia of the extremities; polydactyly, or the increased number of digits or claws in *cats, dogs, cattle, horses and swine*; syndactyly or the union of digits or claws, especially in *cattle and swine*; and ectrodactyly or absence of phalanges.
- Ectopia of the patella has been reported in *cattle, dogs and horses*.

- *Miscellaneous anomalies* due to displacement of tissues include teratomas, dermoids, and dentigerous cysts.
 - **Dentigerous cysts**
 - Characterized by a displaced dental follicle containing fluid and teeth, is seen most commonly in the horse.
 - May be located beneath the ear and are called an “ear tooth.”
 - **Dermoids**
 - Seen occasionally on the cornea, third eyelid or on the neck in cattle and other species.
 - In horses dermoid tumors may rarely involve the ovary or testis, especially the retained testis.
 - **Teratomas**
 - Occasionally seen in all species.

DEFINITION

- Embryonic duplications are malformations due to abnormal duplication of the germinal area giving rise to fetuses whose body structures are partially but not completely duplicated.

FREE MONOZYGOTIC OR DIZYGOTIC TWINS OR TRIPLETS

- Symmetrical twins are either monozygotic or identical, or dizygotic or fraternal.
- Asymmetrical twins consist of a normal and an acardiac or monster individual. These may include three types:
 - **Hemicardius**
 - A very imperfect individual but parts are recognizable, and
 - A rudimentary heart is present.
 - **Holocardius acephalus**
 - The cranial part of very imperfect individual is lacking.
 - No heart is present.
 - **Holocardius amorphus or Amorphus globosus**
 - The general body form is unrecognizable.
 - It may occasionally be seen, most commonly in the cow, but also in the mare, ewe and goat, attached to the placenta of the normal monozygotic or dizygotic twin.
 - These very imperfect zygotes are parasitic upon the placenta of the normal twin and are never observed in single births.

AMORPHUS GLOBOSUS

In Bovine

- Usually appears as a round or oval, edematous structure weighing 1/2 to 7 lbs.
- Covered with skin and hair and containing connective tissue, fat and other soft tissues and occasionally cartilage and bone.
- However, in cattle one acardiac monster was female and its normal cotwin was male. This confirmed Williams theory' that the *Amorphus globosus* was an imperfect zygote of dizygotic twins.
- It was demonstrated that the leucocytes of the normal male calf only contained the XY or male chromosome complement and no blood cell chimerism occurred.
- The monster composed mainly of stomach and intestinal tissues were enclosed in its own amnion.
- In another case, two acardiac monsters were attached to the placenta of a normal bovine calf.



Fig.1: Gross morphology of amorphus fetus with pigmented skin and a few hairs.

CONJOINED TWINS

Conjoined twins in which the components or component parts are symmetrical are called **Diplopagus monsters** or **“Siamese” twins**.

- Triplopagus is extremely rare.
- Conjoined twins arise from a single ovum and are monozygotic.
- Occurrence: about once in 100,000 bovine births.
- Most common in cattle but are seen rarely in sheep, pigs, dogs and cats and are exceedingly rare in horses.

Type I: Those conjoined twins in which each component is complete or nearly so include

- **Thoracopagus, sternopagus, or ziphopagus** twins are joined at or near the sternal region. The internal organs are usually duplicated. The components are face to face.
- **Pygopagus monsters** are connected at the sacrum and the components are back to back.
- **Craniopagus twins** are united at the heads. Components may be facing in the same or in the opposite direction.
- **Ischiopagus fetuses** are joined at the lower pelvic region and the bodies extend in a straight line and the heads in the opposite direction.

Type II: The two components equal one another in this group but each is less than an entire individual.

- Usually associated with lateral fusion.

- May vary from single normal individuals to those of two normal but superficially joined individuals.
- Duplication may lead to doubling of the cranial end of body while the caudal end remains single; or the caudal part may be doubled and the cranial part single.
- Duplication can occur at both cranial and caudal ends with the middle area of the monster remaining single.
- Duplication, of the cranial part of the fetus is more common than that of the caudal portion.

DUPLICATION IN THE CRANIAL REGION

Monocephalus

- Monsters with partial duplication of the frontal region, nose and mouth are referred to as Diprosopus or double face.
- Either face may be complete or one eye of each may be fused into a common medial orbit. Di-, tri- or tetraophthalmus and di-, tri- or tetraotus may be present.

Dicephalus or two heads, with distomus or monostomus occasionally is seen.



In Cow



In Cow



In Buffalo



*Feline Double Monster: Monocephalus, tetrapus
tetrabrachius*

- A Dicephalus dipus dibrachius monster has two fore- limbs and two hind limbs with partial duplication of the spine and one or two tails, dicaudatus.
- Dicephalus dipus tribrachius has 3 forelimbs
- Dicephalus dipus tetrabrachius has 4 forelimbs.

DUPLICATION IN THE CAUDAL REGION

Duplication in the caudal region is designated as Dipygus. Tripygus is very rare.

- Monocephalus tripus dibrachius has 3 rear limbs.
- Monocephalus tetrapus dibrachius has 4 rear limbs.
- Cephalothoracopagus has a single neck and more or less complete fusion of the heads to form an almost single face.
- Syncephalus has one face, four ears, and a single or partially doubled cerebrum.
- Janiceps is a monster with two faces on opposite sides of the head.

DUPLICATION OF CRANIAL AND CAUDAL REGIONS

- Duplication of both cranial and caudal regions is Dicephalus dipygus.
 - Dicephalus tripus tribrachius
 - Dicephalus tetrapus tetrabrachius
 - Dicephalus tripus tetrabrachius, or
 - Dicephalus tetrapus tribrachius

UNEQUAL AND ASYMMETRICAL CONJOINED TWINS

- Unequal and asymmetrical conjoined twins are composed of one very imperfect and incomplete twin, called the *parasite*, dependent on the other twin, the *autosite*. This is called a *heteropagus monster*. The autosite is nearly normal and the parasite is attached to it as a dependent growth.
 - The parasite may be attached to the visible surface of the autosite. Common junction sites are the back, thorax, sacrum or pelvis, and in rare cases the abdomen, head, or palate. The latter is called *epignathus*. Some of the smaller, more imperfect parasites may be called teratomas.
 - A parasite can rarely develop within the autosite usually in the abdominal, thoracic, pelvic or cranial cavity, or in the spinal canal or scrotum.
 - Teratomas in the abdominal cavity and between the mandibles have been described in two calves.

Suggested Reading

Veterinary Obstetrics & Genital Diseases- SJ Roberts Page 49-74.