Teratology and Congenital Malformations

- Teratology is the study of abnormal development.
- The frequency of congenital malformations varies with species, breed, geographical locations and many other factors.
- Approximately 1.5% - 6% of all live-born domestic mammals show at least one congenital malformation.

Causes of Malformations:

A. Genetic factors:
   - Abnormal chromosome numbers
   - Abnormalities in chromosome structure

B. Environmental factors:
   - Mechanical factors-related to a reduced amount of fluid in the amniotic cavity causes abnormal intra-utrine pressure on fetus.
   - Physical factors-Ionizing radiation is a potent teratogen in all species.
   - Chemical factors-Any chemical substance that has the potential to alter cellular function. For example, thalidomide is highly teratogenic in humans, rabbits and some primates. The use of some antibiotics during pregnancy has been associated with congenital defects: streptomycin in high dose can result in inner ear deafness; tetracycline can cross the placental barrier and cause a yellowish discoloration of teeth.
   - Poisonous plants containing toxic or teratogenic compounds have been implicated in congenital defects in herbivorous animals.
   - Infectious agents-most infectious diseases that cause birth defects are viral.

Classification of Malformations

The major types of developmental disturbances found in domestic animals include:

1. Agenesis or aplasia: This is a developmental failure of an organ to form.
2. Hypoplasia and hyperplasia-too small or large.
3. Failure to fuse or close- cleft palate
4. Absence of normal cell death- syndactyly (webbed digit)
5. Duplication and reversal of asymmetry

Congenital Malformations of the Cardiovascular System

A: Atrial septal defect (ASD). The blood is shunted from left to right, because of the higher pressure in the left atrium. The right heart becomes overloaded and overworked from the additional volume of blood it receives. The result is dilatation and hypertrophy of the right atrium and ventricle.

B: Ventricular septal defect (VSD)- It is characterized by the presence of a small opening at the dorsal part of the interventricular septum.

C: Pulmonary stenosis (PS) - It is one of the most common cardiac defects in dogs. It can be present as a single defect or associated with other heart defects. Obstruction of the right ventricular outflow tract cause increased resistance to ejection, leading to right ventricular hypertrophy and septal fattening.

D: Aortic stenosis- obstruction of the left ventricular outfow (LVOT) is caused by a proliferative thickening encircling the aortic outlet immediately below the aortic valves. It results in a ventricular overload, which causes hypertrophy of the left ventricle.

E: Tetralogy of Fallot. It is the most common cyanotic congenital heart defect in domestic animals. It is characterized by the following four lesions:

1. A large ventricular septal defect,
2. An aorta that overrides the left and right ventricles,
3. Obstruction of the right ventricular outfow (pulmonary stenosis) and
4. Right ventricular hypertrophy.

F. Ectopia cordis-Heart does not attain its expected position in the thorax. It is most frequently reported in cattle.

G. Dextrocardia-is a malformation in which the heart lies more to the right side of the thorax than to the left.

Congenital malformations of the vascular system

- Patent ductus arteriosus-most common cardiovascular malformation in dogs
- Persistent right aortic arch - results from a failure of the right dorsal aorta to degenerate



Fig.1 Hydrocephalus in foal

Congenital malformations of the brain

- Hydrocephalus- one of the most common congenital anomalies of the nervous system. It is the abnormal accumulation of cerebrospinal fluid within the ventricular system of brain. It is most common in dogs and cattle.
- Microcephaly-abnormally small brain. Reported in calves, lambs and piglets.
- Hydranencephaly-In hydranencephaly, the cerebral hemispheres are replaced by two fuid-filled sacs. In domestic animals this condition is most commonly caused by utero-viral infection (blue tongue virus in ruminants and panleukopaenia in cats)



Fig.2 Cyclopia in piglet

Malformations of the eye

- Cyclopia (single eyes) - is a defect in which the eyes are partially or completely fused. It is characterized by a single, centrally located orbit, which contains a normal or rudimentary eye.

Congenital malformations of the Uro-genital System

- Abnormalities of testicular descent (Cryptorchidism)- Cryptorchidism is a failure of one or both testicles to descend into the scrotum. It is seen in all domestic mammals, common in stallions, boars and dogs.
- Ectopic ureter
- Patent urachus
- Double cervix-lack of fusion of the paramesonephric ducts

Congenital Malformations of the Digestive System

- Facial cleft
- Cleft lip (cheiloschesis), and
- Cleft palate (palatoschisis)

Abnormalities of the body wall

- Umbilical hernias- result of failures in closure of the muscular body wall. They occur in all species but most common in pigs. The abdominal organs protrude through the umbilicus under the skin; in cats and dogs it is usually the small intestine and calves it is the abomasums.
- Omphalocoele -hernia that occurs in the embryo. The abdominal contents protrude through the umbilicus.