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Congenital anomalies and inherited disorders of the horse include all of the physical abnormalities which are present upon birth of the foal and those that are diagnosed later in life. Some anomalies may be acquired during fetal development while others may be inherited. There have been a few recent breakthroughs in discovering the etiology of some diseases because of their similarity to a human condition, e.g., *Hyperkalemic periodic paralysis (HYPP)* in Quarter horses or crosses.

It is often difficult to determine what event(s) may have resulted in an anomaly. The prevalence or rate at which these anomalies occur in a given population is unknown since very few cases are reported. This information sheet will highlight some of the recognized congenital anomalies of horses (indicated in *italics*). They are categorized under the various body conditions, with brief descriptions where necessary.

**Online Mendelian Inheritance in Animals (OMIA)** is an excellent database and served as a primary source for this information sheet. It categorizes and compiles genetic conditions in horses and other species. It is maintained by the Australian National Genomic Information Service and can be viewed on internet at <http://www.angis.su.oz.au/Databases/BIRX/omia/> <sup>(1)</sup>.

### **Congenital Problems of the Autoimmune System**

- *Severe Combined Immunodeficiency (SCID)* is a fatal disease of Arabian and part-Arabian foals. It is caused by a genetic defect transmitted as an autosomal recessive trait. Affected foals that attain colostral antibody transfer are clinically normal until the colostral antibodies decrease. No functional B and T lymphocytes are produced which leads to a complete lack of antibody production and defective cell-mediated immunity. Affected foals are lymphopenic (less than 1,000 lymphocytes per mm<sup>3</sup>), develop infections and die by 4.5 months of age <sup>(2,3,4)</sup>.
- *Neonatal Isoerythrolysis* is not truly an inherited disease. However, absorption of colostral (maternal) antibodies result in the destruction of the red blood cells in the newborn foal. For it to occur, the dam's blood group must be negative for certain blood types, e.g., Aa- or Qa-. Also, the mare must become sensitized to the offending antigen (Aa or Qa) by exposure through previous pregnancies, blood transfusion or transplacental contamination and the foal must inherit from the sire the antigens (Aa or Qa) to which the mare has been sensitized. Foals become sick after nursing. Jaundice (icterus) is preceded by lethargy (drowsiness) and weakness, and the presence of haemoglobin in the plasma and urine (haemoglobinemia, haemoglobinuria) rapidly follows. A severe anaemia (low red blood cell count) ensues. Prompt recognition and therapy contribute to a good outcome; however, the prognosis in advanced cases is poor. Mares can be screened before parturition (birth of the foal) for alloantibodies against a panel of known erythrocyte alloantigens <sup>(5)</sup>.

### **Congenital Defects of the Eyes and Ears**

- *Colobomas* are defects and, especially, a fissure of any part of the eye.
- *Absence of the nasal punctum*. Tears flow across the eye and drain from the puncti, which are located at the medial canthus (near the nose) of the eye, into the nostrils. Blockages or absence of any part of this drainage system results in overflowing of tears from the lower eyelid(s).
- *Entropion* is the inversion (turning inward) of the margin of the eyelid. It results in chronic irritation of the cornea from the eye lashes turning inward.
- *Congenital cataracts*

### **Congenital Heart Defects**

- Various holes or defects between the atria or ventricles occur as a result of improper fetal development. Other defects are due to failure of various parts of the fetal circulation to close after birth. These include *patent ductus arteriosus*, *patent foramen ovale*, or *persistent fetal circulation*. *Persistent right aortic arch* is a failure of regression in the embryo, resulting in a ring structure that can occlude the esophagus and creating megaesophagus (dilation of the esophagus).

### **Congenital Defects of the Gastrointestinal (GI) System include:**

- *atresia* or blockages of various areas along the intestinal tract. This condition is uncommon in foals and it is not related to the Lethal White Disease. The segment of intestines most often involved is the colon (*atresia coli*), although it has also been described in the rectum (*atresia recti*), anus (*atresia ani*), ileal and jejunal segments. The affected animals develop signs of colic during the first 24 hours of life. Depending on the location of the atresia, scant mucous (lighter than meconium) may be present in the rectum.
- *Lethal white disease* is seen in horse breeds that have white spotting and has been recognized in the following breeds: the Paint horse, Pinto horse, Quarter horse, Miniature horse and Thoroughbred. Horses often nurse vigorously at birth and are fine until their GI tract proximal to the lesion fills up, then they get colicky and look "bloated". At that point, they don't nurse any more. The cause of the intestinal obstruction is a lack of nerve cells in the distal portion of the large intestine (aganglionic megacolon). This is thought to be due to a fault in the proliferation and/or migration of nerve stem cells from the neural crest of the developing embryo. The disease is similar to the human Hirschsprung Disease. It is caused by a mutant allele. Homozygosity for the Overo allele (both alleles are OO) results in white or nearly white foals. Heterozygous animals (Oo) can be tested for the presence of this allele and therefore prevent heterozygous carriers being mated. Lethal white foal syndrome is recessive (i.e. the only horses to show this disorder are homozygotes for the Overo allele).
- *Brachygnathia* refers to a malocclusion between the mandible and the maxilla. Mandibular brachygnathia is most common and refers to an abnormal shortening of the mandible which results in an overshot upper jaw or *parrot mouth*.
- *Cleft palate* is a longitudinal opening in the hard palate which separates the mouth from the nasal pharynx. Signs are usually evident at birth or shortly after nursing when milk drains from the nostrils. An incident level of 0.1 to 0.2 % is reported. A physical exam must be done in a neonate with this sign. Aspiration pneumonia is a common sequel. Surgical therapy is the only option but it may not be recommended.

### **Congenital Disorders of the Musculoskeletal System**

- *Flexural, Contractural and Angular limb deformities* include flexor laxity (flexor ligaments are too loose), flexural deformities of the fetlock and coffin (*club foot* where the deep digital flexor tendon is too tight), varus (e.g., bowlegged) and valgus (e.g., knock-kneed), affecting mostly the carpus (knee), fetlock and tibio-tarsal (hock) regions. These problems are often self-limiting and often resolve with adequate treatment and support.
- *Patellar luxation*. There is a congenital form of this condition which has been described, most commonly in miniature horses and Shetland ponies. The luxation occurs laterally (the patellar can be displaced or moves to the outside of the knee) because of hypoplasia of the femoral trochlea and shallowing of the intertrochlear groove. In newborns, it becomes apparent when the foal tries to stand but fails to extend the stifles, so the foal acquires a crouching position. Femoral nerve paralysis may also produce this sign.
- Skeletal malformations include *wry nose* (twisted nose), *torticollis* (twisted neck), *scoliosis* (lateral deviation of the back), *lordosis* (ventral dorsal deviation of the back).

- Incomplete closures of the bony spinal canal (*cervical meningocele, spina bifida*)
- *Hydrocephalus* (abnormal accumulation of fluid in the cranial vault with resultant enlargement of the head)
- Digital malformations - supernumerary (*polydactyly*) digits, hypoplasia of phalanges, e.g., navicular
- *Malignant hyperthermia syndrome* (hyperthermia of anaesthesia) is a progressive increase in body temperature, muscle rigidity and metabolic acidosis, leading to rapid death, seen when an anaesthetic is administered.
- Hernias (are defects in the muscle wall which permit intestines/organs to move into an abnormal location)
  - ▶ *Diaphragmatic hernia* is a congenital opening in the thoracic diaphragm, permitting the displacement of abdominal organs into the thorax.
  - ▶ *Umbilical hernia* is the incomplete closure of the abdominal wall at the umbilicus.
  - ▶ *Inguinal hernias* are commonly seen in certain breeds, particularly Standardbred and draft horses. Usually they happen in male foals and a large scrotal sac will be seen. They will rarely present colic signs. It is recommended to reduce the hernia on a daily basis and wait for 4-6 months as most of them will resolve. In cases presented with colic or when edema appears in the inguinal/ventral area (ruptured hernia), it is advisable to surgically repair it.
- *Dwarfism* refers to the failure of appropriate growth resulting in a smaller horse. A dwarf horse can be proportionate or disproportionate. Proportionate dwarfs are a result of a deficiency in growth hormone while disproportionate dwarfs result from abnormal thyroid hormone levels. The latter results in a foal with musculoskeletal immaturity, characterized by delayed cuboidal bone development, a large head, silky hair coat, floppy ears and mandibular brachygnathia. Determination and interpretation of either growth hormone assays or thyroid hormone function is not entirely developed or understood in the equine, hence the importance of clinical diagnosis. Efforts towards characterizing thyroid function and growth hormone levels should be undertaken to prevent overdiagnosing this condition. A nitrate toxicity theory has been confirmed in certain foals born with “congenital hypothyroid syndrome”.
- *Tying-up Syndrome (Equine rhabdomyolysis syndrome, Exertional rhabdomyolysis, Myoglobinuria)*. Some forms of this disorder are thought to be inherited as an autosomal recessive trait. However, the published data is inconclusive on this point <sup>(6)</sup>.
- *Hyperkalemic periodic paralysis or HYPP* is marked by sudden attacks of paralysis which, in severe cases, may lead to collapse and sudden death. It is an inherited mis-sense mutation in the gene encoding the alpha chain of the adult skeletal muscle sodium channel, resulting in increased sodium permeability across the skeletal muscle cell membrane. Quarter horse, Paint horse and Appaloosa progeny tracing back to the Quarter horse sire, “Impressive”, can be affected with this disease and must be eliminated from any reproductive program.

### **Respiratory Abnormalities**

- *Guttural pouch tympany* is a distinct soft swelling at the throat latch area. Palpation of the swelling resembles the palpation of a balloon. Radiographs revealed a large air-filled cavity. Horses may also have a respiratory noise, dyspnea (difficult breathing), a cough, dysphagia (difficult swallowing) and aspiration pneumonia.
- *Choanal atresia* is a failure to regress of the bucconasal membrane, producing an airway obstruction at the junction of the nasal cavity and the pharynx. Bilateral cases usually die unless an emergency tracheotomy is performed. Unilateral cases may go undiagnosed; however, asymmetry of airflow can be detected. Surgical treatment by a transnasal or laryngotomy approach or through an endoscope has resulted in some success.

### **Congenital Sex Determination Disorders**

- The intersex disorders *hermaphrodite* and *pseudohermaphrodite* occur when an individual has a mixture of male and female characteristics. It is often due to an abnormality of the sex chromosomes. The disorders include gonadal hypoplasia (small testes or ovaries), gonadal dysgenesis. XY female type, Swyer syndrome, Sry -XX hermaphroditism, and XX male pseudohermaphroditism.

An example is a horse with stallion-like behaviour with gonads retained in the abdomen which histologically were testes but, when karyotyped (looking at the chromosomes), the stallion had the normal 64 chromosomes, two of which were XX (female) chromosomes. This stallion was a 64 XX male pseudohermaphrodite.

### **Congenital Skin or Integument Disorders**

- *Hereditary junctional mechanobullous* is a condition in Belgian foals where the basement membrane in the hoof separates and results in sloughing of the hooves by 12 days of age <sup>(7)</sup>.
- *Undermined skin or Hyperelastosis cutis* (Ehler's-Danlos syndrome in humans) is seen in Quarter horses and Haflingers. Ehlers-Danlos syndrome in humans is an autosomal dominant trait with variable expression.
- *Dilute Lethal or Lavender Foal Syndrome* occurs in Egyptian and part-Egyptian Arabian horses and is usually fatal within 48 hours of birth. Foals are born with difficulty (dystocia), fail to stand or nurse and have neurological problems (intermittent joint rigidity and rapid eye movements). The foals are called 'lavender' because the hair coat has a diluted lavender or pink colour. This may be due to abnormal clumping of the pigmentation in the hair, but could also be attributed to cyanosis (lacking oxygen) caused by the long and difficult birth. The foal is often larger than normal. On postmortem, vacuolations of the neurons are found <sup>(8)</sup>.
- *Roan coat colour* occurs at a low frequency (generally less than 5%) in many horse breeds. Hintz and van Vleck (1979) provided convincing evidence that roan coat colour in Belgian horses is due to heterozygosity for an autosomal gene that is lethal when homozygous <sup>(9)</sup>.

### **Congenital Diseases of the Urogenital Tract**

- *Ruptured bladder*. The etiology of this condition is believed to be traumatic and occurs most commonly in male foals during parturition, although the signs of uroperitoneum (urine in the abdomen) do not appear until 2-5 days of age. These animals can be suffering from very severe metabolic disturbances and their correction previous to surgical repair is of paramount importance. Therefore, ruptured bladder is not a surgical emergency but a medical one. Clinical signs involve lethargy, abdominal distention, decreased appetite and mild colic. Affected foals may urinate normally although commonly they are seen to strain and make frequent attempts to urinate. The back is seen ventroflexed (flexed downwards) as a difference with meconium impaction where the back is dorsoflexed (flexed upwards). Serum electrolytes and metabolites will be affected and are characterized by increased potassium (hyperkalemia), decreased sodium (hyponatremia), decreased chloride (hypochloremia) and presence of urea in the blood (azotemia). Diagnosis of uroperitoneum is based on comparison of levels of creatinine in peritoneal fluid and serum.
- *Patent Urachus* is where urine is detected leaking from the umbilicus. This could be a normal finding in foals up to 5 to 7 days of age. However, failure of the urachus to close could result in septicemic (blood infection) foals. It is not a surgical emergency and time and medical treatment should be considered before engaging in a surgical procedure.
- *Recto-vaginal, urethro-rectal fistula* are characterized by a bypass of urine into the rectum or feces into the vagina or urethra. These types of abnormalities are usually a part of a larger picture where other congenital abnormalities related to the urogenital tract are present. In addition, in rare occasions, some of these cases may also present an atretic segment of bowel (section of bowel

without an opening).

- *Ectopic ureter* is unusual in horses. There is no predisposition of sex or breed. It may go undetected for years or the foal may present signs of urinary incontinence because the ectopic ureters usually open distal to the trigone of the bladder. Unilateral or bilateral cases may occur and some cases may also present with hydro-ureter (dilated fluid filled ureter) or hydronephrosis (dilated kidney). Surgery has been successful in some cases.
- *Uterus unicornis* signifies the presence of a single uterine horn rather than two.

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