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CONGENITAL ABNORMALITIES
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INTRODUCTION

Congenital abnormalities (CA) may be conspicuous macroscopically or only microscopically; they may be visible to the naked eye or affect the internal organs; they may be anatomical or functional. CA can be genetically determined or develop during pregnancy due to environmental influences. Often, a combination of both is involved. Usually, genetic defects affect only individual littermates, whereas environmental influences (e.g., infections, toxins, etc.) affect the whole litter. During the first four weeks of pregnancy, when organogenesis takes place, the fetus is most susceptible. The same clinical syndrome (e.g., megaesophagus, cleft palate) can be caused by genetic or environmental influences. Age at detection of a CA varies. While some are obvious at birth (e.g., hare lip), others may be detected at days to weeks of age (e.g., atresia ani) or even later in life (e.g., ectopic ureters in male dogs). As multiple malformations are frequently found in the same animal, meticulous examination is recommended.

SELECTED CONGENITAL ABNORMALITIES

Dysraphism

Dysraphism, including encephalocele and spina bifida, refers to malformations caused by incomplete closure of the neural tube. The condition may be associated with butterfly vertebrae or hemivertebrae. Usually, animals with malformations of the vertebrae are asymptomatic. Thoracal hemivertebrae are common in brachycephalic breeds. Transitional vertebrae are associated with the cauda equina syndrome in the German shepherd dog.

Dermoid sinus

A dermoid sinus is an epithelialized fistula connecting skin and vertebral canal. The condition is caused by incomplete closure of the embryonal neural tube. The fistula may end in subcutaneous fat, the meninges, or in the spinal canal. Clinical signs vary with degree of disease. If a dermoid sinus, which is connected with the dura mater, gets infected, neurological symptoms result. Diagnosis is made by fistulography (risk of infection!), myelography, ultrasonography or MRI. Surgical correction is necessary. Various breeds are affected, but most commonly the Rhodesian ridgeback and its crossbreeds.

Hydrocephalus

The congenital form of hydrocephalus is usually caused by a stenosis of the aqueduct of Sylvius. The condition is frequently an incidental finding in asymptomatic toy and brachycephalic breeds. In the Siamese cat, hydrocephalus is inherited as an autosomal recessive trait. Clinical signs are caused by neuronal loss and intracranial pressure rise and are dependent on the affected brain area. Typical findings are a domed cranium, ventrolateral strabismus, blindness, somnolence, compulsive walking and seizuring. If the fontanelle is still open, diagnosis can be established by ultrasonography, otherwise by CT or MRI. Surgical correction is necessary. Various breeds are affected, but most commonly the Rhodesian ridgeback and its crossbreeds.

Anasarca

Puppies affected with anasarca are usually delivered by caesarean section because the extensive subcutaneous oedema prevents a natural birth. The cause of the condition is unknown, a genetic disposition is suspected in the Bulldog, Bichon, Chow Chow, Labrador and Schnauzer. Administration of furosemide and KCl may be curative in some cases. Urination should be stimulated every 30-60 minutes. If treatment is still ineffective after three cycles of furosemide, it should be stopped.

Swimmer syndrome (also flat puppy syndrome), pectus excavatum, tarsal hyperextension in cats
Puppies affected with swimmer syndrome are unable to walk and lie in sternal recumbence. Forward movement is accomplished by lateral “swimming” motions. The cause is unknown. The syndrome occurs more often in brachycephalic breeds, a genetic background is possible. Possible consequences are dorsoventral compression of the thorax and skeletal joint and diaphyseal abnormalities. Prognosis is favourable if therapy is initiated before the age of four weeks. Treatment aims at muscle strengthening.

Because of similar clinical signs, swimmer syndrome is often mixed up with pectus excavatum, a congenital deformity in which the sternum intrudes into the thorax. The condition is more common in brachycephalic dog and cat breeds. The aetiology is unknown. External as well as internal fixation improved clinical symptoms in dyspnoeic patients.

Tarsal hyperextension (also twisted leg syndrome) is a rare malformation in cats, where one or more limbs are twisted and stunted. The cause is unknown; treatment is by splinting, followed by physiotherapy.

**Brachycephalic syndrome**

Brachycephalic dogs, and, more rarely, cats may suffer from exercise and heat intolerance, stridor, dyspnea, cyanosis, hyperthermia and collapse. The upper airways are too narrow, leading to increased airway resistance and consequently to lengthening of the soft palate, eversion of laryngeal sacculae and eventually to laryngeal collapse. Usually, first clinical signs are noted before 6 months of age. Diagnosis is made by endoscopy. Treatment consists of surgical correction of the stenotic nares and the elongated soft palate. Early correction improves the prognosis.

**Primary ciliary dyskinesia**

Ciliary dysfunction decreases mucociliary clearance and results in recurrent rhinitis, sinusitis and bronchitis. The condition is more prevalent in the Bobtail and the Springer spaniel, and may be inherited as an autosomal recessive trait. Usually, affected dogs have nasal discharge already at the age of a few weeks. Symptoms can be temporary relieved by antimicrobial therapy. Kartagener’s syndrome (situs inversus, bronchiectasis and rhinitis/sinusitis is found in about 50% of affected animals. Treatment consists of intermittent or permanent use of antimicrobial agents and supportive care such as inhalation therapy and coupeage.

**Cleft palate, cleft lip**

In the Brittany spaniel and the Great Pyrenees cleft palate is inherited as an autosomal recessive trait, in other breeds, polygenetic inheritance is suspected. Teratogenic agents (e.g. griseofulvin) may also cause cleft palate. Drainage of milk through the nostrils during and after nursing is characteristic. Other clinical signs include slow growth, sneezing while eating and coughing. Aspiration pneumonia is common. Surgical repair is usually attempted at the age of 6-9 weeks. Prognosis depends on extent of the cleft palate and feeding management. Cleft lip (hare lip) usually is associated with cleft palate.

**Atresia ani**

Atresia ani is due to incomplete or absent perforation of the anal membrane. The condition encompasses 4 types: anal stenosis (type 1), imperforate anus (type 2), imperforate anus associated with a blindly ending rectum at a variable distance from the anal membrane (type 3) and normal distal rectum and anus associated with agenesis of the proximal rectum (type 4). Clinical signs are noticed during the first few weeks of life. Animals affected with type 1 have tenesmus and constipation shortly after weaning. Animals affected with type 2-4 are uneasy and do not defecate. The abdomen is distended and the perineum may bulge out. Vomitus may be present. Diagnosis is by imaging (rectal barium contrast study). Prognosis for type 1 and 2 is good after balloon dilatation and surgical repair, respectively. After surgery, risk of faecal incontinence remains.

**Urogenital anomalies**

Ureteral ectopia (UE) is more common in Retrievers, Poodles, Huskies, Collies, Spaniels, Briards, Entlebucher mountain dog and some Terrier breeds. One or both ureters do not terminate normally in the trigone region of the urinary bladder, but more caudally in the bladder neck, urethra, vagina or uterus. Intramural EU contact and enter the bladder wall normally but continue submucosally and terminate further distally. Extramural EU totally bypass the bladder. Permanent urinary incontinence is the predominant clinical sign and is usually observed during the first year of life in bitches, whereas in male dogs, it may not become apparent until later in life or after castration. If localization or structure of the ectopic orifice impedes urinary flow, hydronephrosis and megaureter result. Recurrent cystitis is common.
Diagnosis is established by ultrasonography, IVP, retrograde contrast urethrography, urethrocystoscopy or spiral-CT. Surgical correction results in continence in 70% of the patients. EU are often associated with other urogenital anomalies such as congenital USMI, ureterocele, hypoplastic bladder or kidneys, vestibulo-vaginal malformations or cryptorchidism. Affected dogs should not be neutered, thereby avoiding a further decrease in urethral sphincter pressure. Clinical signs and treatment of congenital USMI are similar to spaying-induced USMI. Concurrent cystitis is common and should be treated accordingly.

Incomplete atrophy of the urachus results in persistent urachal patency or in formation of a diverticula. Persistent urachus is associated with permanent loss of urine through the umbilicus and is often accompanied by dermatitis of the umbilical region and cystitis. Permanent licking of the umbilical region by the bitch usually is the first clinical sign. Treatment consists of surgical correction and therapy of concurrent cystitis. Presence of a bladder diverticula may predispose the animal to or complicate urinary tract infections and urolithiasis. However, about 30% of cases are asymptomatic. Diagnosis is established by positive contrast or double contrast radiography. Treatment consists of antimicrobial therapy of the urinary tract infection and resection of the diverticula if clinical signs recur.

Congenital phimosis is most often caused by a stenosis of the prepuce. Affected dogs are usually presented for signs of urinary incontinence or dysuria because the failure of the penis to be exteriorized leads to urine pooling within the prepuce. The condition is surgically corrected. Paraphimosis occurs more frequently and is associated with more severe clinical signs. The underlying cause is most often congenital preputial stenosis or hypoplasia. Failure to retract the extruded penis into the prepuce leads to congestion, swelling and severe irritation. If treatment with cold compresses, hyperosmolar solutions, lubrication and removal of entrapped hairs fails, immediate surgical intervention is indicated.

Persistent frenum is a string of remnant connective tissue between the prepuce and the tip of the penis. No hereditary basis has been identified so far. Clinical signs include excessive licking of the penis and prepuce, dermatitis associated with urine pooling within the prepuce, and painful and abnormal erections. Treatment includes surgical transsection of the frenulum.

Cryptorchidism is very common in dogs (0.8 – 15%). Incidence is increased in purebred dogs, especially toy and brachycephalic breeds. In litters with many males, more individuals are affected. Genetic, epigenetic and environmental factors are likely to be involved in the failure of testicular descent. The defect is probably inherited as a sex-linked recessive trait with variable penetrance. A definitive diagnosis is made at 6 months of age, after closure of the inguinal canals. Cryptorchid testes are able to produce hormones, but spermatogenesis is disrupted. As the risk of becoming neoplastic is 9-13 fold increased in cryptorchid testes, surgical removal before the age of 5 years is strongly recommended. In cats, cryptorchidism, although being the most common urogenital malformation, is much less frequent than in dogs. A cryptorchid tomcat can be easily distinguished from a neutered one, as presence of penile spines is dependent on testosterone.

References available from author upon request