Congenital and hereditary diseases to be diagnosed in the kitten

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Congenital diseases are those, visible or non visible, but present at birth. They could be from genetic origin or they can represent a teratogenic cause, in some cases also phenocopies. Phenocopies show the clinical appearance known also for classical genetic defects, but they were acquired during embryogenesis by teratogenic factors. If there is no genetic test available for the specific breed or disease, the causes will be often unspecified.

Anomalies of microanatomic or biochemical type usually go unreported and are included under stillbirths, faders, or undetermined causes, if kittens die early. Some anomalies in the brain, heart, or respiratory system can cause immediate threat to life, resulting in death at birth or within the first days, weeks or months, whereas other malformations remain unnoticed for a different period, depending on the localisation. In most cases, pathologic and histological examination is the only possibility to diagnose non visible malformations. The occurrence of a pathognomonic symptom— a typical sign on which a diagnosis or a suspicion can be made - helps in some cases to be able to recognise the problem. In other cases, X-ray, sonography , endoscopy, or blood tests can help to diagnose the defect and to make a prognosis. Some of internal defects ones can find accidentally (e.g.during surgery or necropsy). Live born kittens should be euthanised if they show an untreatable condition.

1 defects and malformations visible at birth (selection)

**Palatoschisis (cleft palate, cleft lip)**

All degrees of cleft palate are conditions that should be easily to diagnose after birth by inspection of the oral cavity.

A cleft palate is a common malformation we have to deal with. Both in puppies and kittens – esp. Simese cats - an incomplete embryonic closure of the palate occurs. There is a high degree of variability in the cleft palate complex. In some cases, only the soft and hard palate is affected, in others additionally a cleft lip occurs as an unilateral defect in the lip or in the floor of the nostril (Cheilo-Gnato-Palatoschisis). The cause may be congenital only (hypervitaminosis A, administration of corticosteroids, metronidazole, griseofulvin … during pregnancy) or inherited with a simple recessive or irregular dominant trait. A lack of folic acid like in human medicine is not very likely in kittens. A small extent of the defect allows surgical repair of a cleft palate, but not earlier than 8-10 weeks of age. Tube feeding is the only effective method of providing adequate nutrition until surgery. Especially in large defects breeders often decide to euthanize the newborn kitten. It is also recommended in combined malformations.

**Herniation (most common: umbilical-, abdominal-, inguinal hernia)**

The abnormal protrusion of an organ, parts of an organ or other body structures through a defect or a natural opening is called herniation. Umbilical hernia is a type of abdominal herniation in which parts of the intestine or omentum protrudes at the umbilicus and can be covered with skin and subcutaneous tissue. Most cases are inherited, but umbilical hernia can also be acquired. There is no genetic test available. The hernia of an intestinal loop into the inguinal canal is called inguinal hernia. In male kittens it is also called a scrotal hernia. A familiar disposition becomes often obvious. Early surgery is the best way to treat the condition. If it is no emergency, the defect should be closed between 6 and 8 weeks of age.

**Congenital anorectal anomalies**

Imperforate anus, segmental aplasia, rectovaginal fistula or rectal urethral fistula rarely occur. From all, imperforate anus is the most common. Both terratogenic and hereditary reasons are suspected, but genetic testing is not available yet. The treatment depends on the dimension and size of the malformation (anal atresia, several types). In fistulas, it is advised to wait with surgery up to the 12th week, if it is medically justifiable.

**Spina bifida (spinal dysraphism)**

Spina bifida is the most common vertebral lesion from spinal dysraphism and refers to a defective fusion of the vertebral arch. The dorsal spine and vertebral arch are absent. It is suspected a malformation due to arrested development. Some lesions can also originate from inherited cause
(sacrocaudal vertebral dysgenesis as an autosomal dominant trait e.g. in Manx cats). In severe cases the kittens die early after birth or should be euthanized.

**Hydrops universalis (anasarca)**

Anasarca is a gross edema of the entire body of a fetus/newborn kitten. The causes are usually non-infectious, it is suspected cardiac malformations, fetal kidney diseases or a disturbance of the lymphatic system/vessels. These enlarged fetusses often induce a birth problem (obstructive dystocia). Diagnosis can be made by ultrasound (liquid within the subcutaneous tissue), but not by X-ray (normal skeleton). Affected kittens usually die immediately after birth.

**Acrania /anencephalic kittens**

Anencephaly is a type of cranial dysraphism. It is a condition in which the brain is absent at birth, or only basal nuclei and cerebellum are developed. In single pup pregnancies, prolonged gestation is observed. There is no genetic test available, but in Burmese cats a simple autosomal recessive mode in craniofacial anomalies is known.

2 defects that appear within the first weeks

**Pectus excavatum**

Pectus excavatum is a result of a sternal malformation. The sternum extends into the thoracic cavity and the ventral rib ends turn medially to meet the displaced sternum. The condition is more often found in the Maincoon, but also in other breeds. Affected kittens esp. those with a flat chest are unable to walk and breath in a normal way. Such kittens mostly lay in ventral recumbancy. The most important problem is the dislocation of the heart and the lung, leading to pneumonia and difficult breathing. Surgical repair is indicated in severe cases, no specific tretment is required in mild cases.

**Hydrocephalus**

Most cases of congenital hydrocephalus in kittens are from internal type. Congenital hydrocephalus results from structural defects that either obstruct CSF outflow or impede absorption. Some cases appear to progress after birth (continued accumulation of CSF in the ventricular system e.g. in computed tomography). Clinical signs can vary from minimal up to marked neurologic dysfunctions (dome-shaped calvarium, blindness, seizures, ventrolateral strabismus). An open fontanelle is not from diagnostic value. Ultrasonography or MRT are useful tools for diagnosis. Treatment depends on the underlying cause. Clinically, affected kittens regularly show retarded growth compared to litter mates.

Degenerative neurologic diseases in cats include GM, gangliosidosis (Korat cat), congenital strabismus and nystagmus (Siamese cat), or sacrocaudal malformations (Manx cat).

**Anophthalmia, microphthalmia**

Anophthalmia, a complete absence of the globe, is extremely rare. In most cases, there is still primitive ocular tissue left. Microphthalmia, retinal dysplasia, and tapetal aplasia have been reported to be hereditary but also after intrauterine infections in Domestic Shorthair kittens. Multiple anomalies have been described in Persian cats. Administration of griseofulvin in pregnant queens can cause microphthalmia, anophthalmia, or optic nerve aplasia.

**Diaphragmatic hernia**

There are two types: peritoneopericardial hernia and pleuroperitoneo-diaphragmatic hernia. The first type is much more common in young cats. A ventral diaphragmatic defect allows abdominal viscera to intrude into the pericardial sac. The defect could be developmental and does not appear to be inherited. Although some references suspect a genetic basis, a mode of inheritance is not known for any breed. Clinical signs depend on the protruded organ(s). Gastrointestinal or pulmonary signs are most common. Diagnosis can be made by X-ray and ultrasonography. Treatment, if indicated, is usually surgical.

**Megaoesophagus**

Idiopathic forms of megaoesophagus (congenital or aquired) are caused by a dysfunction of the primary motor system of the esophagus. This results in an abnormal transport of the ingesta between the pharynx and the stomach. Siamese and Siamese-related breeds have the highest incidence. From infectious causes, viral infections and toxoplasmosis contribute to the development of megaoesophagus. The incidence is highest in special dog breeds, but in some cases it occurs also in kittens. Clinical signs (regurgitation, foetor ex ore, pneumonia…) occur mostly after the 3rd week with changes is nutrition up to weaning. Contrast radiography helps to confirm the diagnosis. Treatment depends on prognosis and usually consists of conservative therapy (feeding in standing position and keeping the head up, vitamin B administration, antibiotics in the case of aspiration).
Malformations in kittens usually visible beyond 4 weeks

Ectopic ureters
An ectopic ureter is a congenital anomaly in which the ureter terminates abnormally outside the urinary bladder (intramural, extramural with termination into the urethra or vagina). Unilateral and bilateral ureteral ectopia occurs both in male and female kittens, although the incidence is much lower compared to dogs. In cats, familiar or breed predispositions have not been found. Usually it becomes obvious after weaning. Excretory urography or ultrasonography help to diagnose the problem. The treatment is surgical.

Malformations that appear at any time (usually within the first year) after birth
Liver shunts (Himalayan cat, Persian cat, mixed cats) or congenital heart diseases are common, some with breed predispositions and inherited traits. For several diseases, special diagnostic tools or genetic testing is available.

In general, most of the anomalies are unpredictable and unavoidable. So they count for a large part of losses and euthanised puppies and kittens. The estimated number of unreported cases is possibly much higher than the proportion in the literature, especially concerning internal defects.

Literature