

ABNORMALITIES IN PEDIGREE CATS

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Categories : [Vets](#)

Date : June 16, 2008

CATHERINE F LE BARS focuses on neurological and musculoskeletal conditions in the first of a five-part series

CONGENITAL defects are anatomical or functional abnormalities that are present in a neonate and may occur as a result of any factor that influences the normal development of the foetus.

Such causes may include infectious or environmental agents, pharmacological substances, nutritional supplements and genetic defects.

The feline genome project is now in its third decade and more than 250 genetic diseases are recognised, with more identified each year.

This is the first of a fivepart series of articles that will examine conditions found in domestic cat breeds that have a suspected or proven genetic basis, with emphasis on neurological and musculoskeletal conditions. Not all these conditions are congenital, but many are expressed in young animals. Where a definite mode of inheritance has been confirmed, it will be described.

Ataxia

Ataxia may be seen in a variety of conditions, including cerebellar abiotrophy, which presents as severe ataxia, a wide-based stance, symmetrical hypermetria and spasticity.

Other signs include intention tremors, postural reaction deficits, vertical nystagmus, absent menace

response and decreased pupillary light reflexes. Clinical signs are slowly progressive. Histopathology reveals a marked reduction in the Purkinje cell population and attenuation of the granule cell layer. Other cases, seen in younger cats, have exhibited multifocal Purkinje cell degeneration with reactive gliosis and cerebellar foliar disorganisation. A recessive mode of inheritance is suspected.

Cerebellar hypoplasia results in similar signs shortly after birth. A similar condition, cerebellar degeneration (seen from seven weeks of age onwards), has been proven to have an autosomal recessive mode of inheritance. Although clinical signs progress over a couple of months, the condition is not fatal.

Congenital unilateral peripheral vestibular disease has been documented in Siamese, Burmese and Tonkinese cats. Clinical signs are evident at birth or within the first few months, and may be severe initially. As with other forms of vestibular disease, the cats may learn to compensate and enjoy an acceptable quality of life.

Feline hereditary neuroaxonal dystrophy is characterised by the early development of progressive ataxia during infancy and is associated with an abnormal coat colour. An autosomal recessive mode of inheritance has been demonstrated. Histopathological findings include a neuronal swelling within the brain stem, changes within the spiral ganglia and organ of Corti in the inner ear, and atrophy of the cerebellar vermis.

Deafness

Congenital deafness in cats is caused by degeneration of the auditory apparatus of the inner ear. There is also a proven relationship between coat and eye colour. The gene responsible is the autosomal dominant gene W, which has complete penetrance for white coat colour, incomplete penetrance for deafness and incomplete dominance for blue iris colour. Environmental factors and other genes may affect the expression of deafness and eye colour. Deafness may be unilateral or bilateral and occurs in less than 1.5 per cent of the cat population across a number of countries. A cat with blue eyes is three to five times more likely to be deaf compared to other cats, and a cat with one blue eye is twice as likely to be deaf. Coat length is not significant in unilateral deafness, but long-haired cats are three times more likely to exhibit bilateral deafness.

In addition to being deaf, photophobic and having reduced vision in low-light conditions, affected cats are also believed to have reduced resistance to disease and semi-sterility.

Many cat breeds are known to have the W gene and a number of breeds, such as the Norwegian forest cat, are checked for deafness using brainstem auditory evoked response testing in an attempt to remove the gene from the population.

Inherited polyneuropathies

Sphingomyelinase-deficiency polyneuropathy, or Niemann- Pick disease, is an autosomal recessive lysosomal storage disease characterised by a deficiency of sphingomyelinase. An associated primary polyneuropathy has been described in three Siamese kittens.

Hyperchylomicronemia-associated neuropathy is a suspected autosomal recessive disease characterised by fasting hyperlipaemia, lipaemia retinalis and peripheral neuropathy. Clinical signs rarely occur before eight months of age.

Hypertrophic polyneuropathy has been described in two unrelated 12-month-old cats. The affected cats had intention tremors, decreased postural reactions, hyporeflexia and mild sensory loss.

Birman cat distal polyneuropathy is a degenerative polyneuropathy that has been reported in related litters of Birmans. A recessive mode of inheritance is suspected.

Lysosomal storage diseases

Lysosomes are intracellular structures that contain enzymes involved in the metabolism of cellular products.

Disease occurs when enzyme deficiencies lead to the accumulation of a specific cellular product. Many lysosomal storage diseases have an autosomal recessive mode of inheritance. The diseases are rare and diagnosis can be difficult, leading to under diagnosis.

Certain disorders have been associated with specific breeds (listed in [Table 1](#)) and all have been identified in domestic short and long-haired cats.

Most affected animals show clinical signs from an early age and neurological abnormalities are common. Certain types of these conditions may be identified by genetic testing and the detection of abnormal metabolites in the urine.

Spongiform degeneration

Spongiform degeneration is a disorder of myelin synthesis, characterised by widespread vacuolation of the white matter and, to a lesser degree, the grey matter of the brain and spinal cord. A genetic cause is suspected in Egyptian mau kittens, although some cases may result from exposure to certain toxins. Clinical signs are evident from a few weeks to a few months of age, and consist of pelvic limb ataxia and hypermetria, progressing to depression, inactivity and seizures. Prognosis is poor and histopathology is the only means of diagnosing the condition. A similar condition has been seen in a Persian kitten.

Anatomical abnormalities

Polydactyly can affect the front or hindpaws and has been seen in many breeds, notably the Manx and the Maine coon, in which it is usually inherited as an autosomal dominant trait, sometimes with variable gene expression. More than one gene may be involved, affected genes may vary between breeds and the condition can occur as a result of teratogenic exposure of the pregnant queen.

Split foot (ectrosyndactyly) is characterised by the fusion or absence of digits on the forepaws and is not specific to a particular breed. It is believed to be a monogenic dominant trait.

A short bent tail (brachyury) is part of the breed description for certain breeds, such as the Japanese bobtail, and is believed to be an autosomal recessive trait. It may also be seen in the Scottish fold.

Kinked tails are seen in many breeds, particularly Siamese, Burmese and oriental breeds. They usually occur as a result of bony deformities and are often of aesthetic relevance only. The mode of inheritance is, as yet, unknown.

Curled ears are part of the breed description for the Scottish fold and occur as a result of a simple dominant gene. All cats carrying the gene suffer from degenerative joint disease; those with two genes are always severely affected, raising ethical questions regarding the breed.

Umbilical hernias of varying severity can be seen in all cat breeds and may result from excessive tension placed on the umbilicus immediately after birth and teratogenic exposure of the pregnant queen (such as methotrexate). Brachycephalic cats are over-represented, perhaps because the queen has difficulties chewing through the umbilical cord. A genetic basis is suspected but has not yet been proven.

Arthritis

Radiographic evidence of degenerative joint disease is present in 70 to 90 per cent of all older cats. This appears most commonly in the elbows and hips. Arthritis has been seen in breeds predisposed to the development of hip dysplasia and/ or patellar luxation.

Burmese cats may develop arthritic changes in their elbow at a relatively early age. Severe osteoarthritis often results in reduced mobility, altered social interactions and loss of litter tray training.

Dwarfism and chondrodystrophic disorders

Dwarfism may occur as a result of genetic, hormonal or environmental factors. Some of these cats are in normal proportion and may have a growth hormone deficiency or may simply be the offspring of small cats.

Disproportionate dwarfs have a normal-sized body with short legs and occur in any breed, as a result of different gene defects. They tend to exhibit joint pain and arthritis.

Many cases are familial and inheritability has been shown to be autosomal recessive in a few cases. These traits have been selectively bred into the munchkin breed.

Hip dysplasia

Hip dysplasia is poorly recognised in cats compared with dogs, and is often an incidental finding - as many affected cats may be asymptomatic. It is not congenital.

Studies indicate a high prevalence in certain breeds, such as Maine coons, Himalayans, Persians and the Devon rex, with the condition in these breeds often occurring concurrently with patellar luxation.

The acetabulum is commonly malformed, although misshapen femoral heads are occasionally recognised. It is thought to be inherited. However, the role of environmental influences is unclear.

Muscular dystrophy

Hypertrophic muscular dystrophy is a rare disease, most commonly occurring in male domestic shorthaired (DSH) cats as an X-linked autosomal recessive condition. In this disease, muscles in affected cats are deficient in dystrophin, leading to weakness and compensatory excessive muscling. Dysphagia, as a result of oesophageal dysfunction and glossal enlargement, may also be seen. Treatment is symptomatic.

Myotonia

Hereditary myotonia has been seen in a small number of cats, with no particular breed predisposition. An autosomal recessive mode of inheritance is suspected. It is characterised by the continuing contraction of a muscle after the cessation of voluntary effort, which results in muscle spasm, stiffness and an awkward gait. Limbs are often abducted because the proximal limb joints have limited flexion. The stiffness is worse after rest and improves with exercise. There is widespread hypertrophy of muscle groups.

Affected kittens may fall into lateral recumbency with their legs extended and exhibit spasms of the facial muscles when startled. Occasionally, they are dysphagic and diagnosis is by electromyogram or muscle biopsy.

Myopathies

Other forms of muscular disease have been reported occasionally in the cat, including a merosindeficient myopathy in Maine coons and Siamese. All affected cats had progressive hindlimb weakness, from two to five months of age. The mode of inheritance is unknown.

A nemaline myopathy has been reported in DSH cats. It has been suggested that this is an autosomal recessive condition that results in a progressive weakness, a staccato gait and tremors. The clinical signs are first noted from six months of age. Laminin α 2-deficient muscular dystrophy has been seen in DSH and Siamese cats from six to 12 months of age. Clinical signs include paraparesis, progressing to tetraparesis, trismus, severe extensor contracture and muscle atrophy. The mode of inheritance is unknown. All these conditions carry a guarded prognosis.

Devon rex myopathy has been identified in the breed between three and 23 weeks of age. Clinical signs include passive ventroflexion of the head and neck, generalised muscle weakness and dysphagia. Clinical signs progress until the cat matures, at which point they stabilise and may improve. The disease is thought to be inherited in an autosomal recessive fashion.

Neuromuscular disease

This condition has been recognised in four to eight-month-old snowshoe kittens, which develop intermittent hindlimb weakness and muscle atrophy. The condition becomes progressively more debilitating and the prognosis is guarded. Inheritability of the condition is, as yet, unknown.

Patellar luxation

Patellar luxation is usually medial and may be present as a congenital defect. In these cases, it is usually bilateral. It is often seen in association with hip dysplasia.

There are several stages of patellar luxation and surgery may be required in more severe cases. Patellar luxation has been recognised in many breeds, notably the Abyssinian, Devon rex, Bengal and Maine coon.

Spinal muscular atrophy

This neurodegenerative disorder has been identified in a family of Maine coon cats. Clinical signs occur between 15 and 17 weeks of age and include fine muscle tremors and fasciculations, with progressive muscular weakness. Cranial nerves, mentation and spinal reflexes are normal. Although signs are progressive, with help cats can enjoy an acceptable quality of life. The mode of inheritance is autosomal recessive.

As can be seen from the (nonexhaustive) list in [Table 1](#), the Burmese and oriental breeds are over-represented with regard to musculoskeletal and neurological disorders having a suspected or

proven genetic basis. Other breed descriptions, such as the Scottish folds, are based almost entirely on the presence of genetic disease.

Few DNA tests are currently available in cats and the heritability of many of the conditions described in the table is still poorly understood.

However, as mapping of the feline genome progresses, our ability to identify the presence of genetic disease in young animals should improve, allowing the reduction of disease incidence in specific breeds by the employment of careful and selective breeding.



Left: Persians are predisposed to musculoskeletal disorders, including prognathism and hip dysplasia.



Right: Birmans suffer from neurological diseases that are suspected or confirmed as having a genetic basis. These include spongiform degeneration and trembling kitten syndrome.

Breed	Disorder
Abyssinian and Somali	Myasthenia gravis; hip dysplasia
Bengal	Distal neuropathy; flat-chested syndrome; hip dysplasia
Birman	Spongiform degeneration; axonopathy; shaking or trembling kittens
British shorthair	Femoral neck fracture
Burmese and Asians	Gangliosidosis GM2; feline orofacial pain; congenital peripheral vestibular disease; kinky tail; elbow osteoarthritis; flat-chested syndrome; hypokalaemia; Burmese head defect
Cornish rex	Umbilical hernia
Devon rex	Devon rex myopathy; hip dysplasia
Himalayan	Hip dysplasia
Korat	Lysosomal storage disease (GM1 and GM2)
Maine coon	Spinal muscular atrophy; hip dysplasia; patellar luxation
Manx	Manx syndrome
Munchkin	Dwarfism and chondrodystrophic disorders
Norwegian forest cat	Glycogenosis; deafness
Persian and chinchilla	Lysosomal storage disease (mannosidosis); prognathism; umbilical hernia; hip dysplasia
Ragdoll	None recognised
Scottish fold	Osteochondrodysplasia
Siamese and Balinese	Lysosomal storage diseases (GM1; α -mucopolysaccharidosis type six); Niemann Pick disease type A; ceroid lipofuscinosis; ataxia; congenital peripheral vestibular disease; hydrocephalus; hyperaesthesia syndrome; cleft palate; kinky tail; femoral neck fracture (avascular femoral neck necrosis)
Siberian	None recognised
Snowshoe	Neuromuscular disease
Sphynx	None recognised
Turkish Van	None recognised

TABLE 1. Common breeds and their predispositions to musculoskeletal and neurological disorders

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