



Feline Genetic Diseases: A Review

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ABSTRACT

Feline genetic disorders pose a considerable challenge in veterinary medicine, influencing multiple organ systems and frequently displaying breed-specific tendencies. Recent developments in genetic screening have uncovered a variety of hereditary conditions in cats, including hypertrophic cardiomyopathy, polycystic kidney disease, and progressive retinal atrophy, all of which can affect cardiac, renal, and visual functions. Many of these conditions arise from single-gene mutations or intricate genetic interactions, with both autosomal dominant and recessive inheritance patterns being prevalent. The implementation of genetic testing and preventive breeding methods has become crucial in diminishing the occurrence of these diseases, thereby enhancing feline health and welfare. Continued research is broadening our comprehension of feline genetics, facilitating the creation of targeted diagnostics, treatments, and responsible breeding methodologies.

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INTRODUCTION

Inherited disorders are medical conditions that result from abnormal genes transmitted from one generation to the next. While some genetically determined disorders may be evident at birth, others might not manifest or become apparent until later in life. Similar to other animals, cats can experience inherited disorders; however, these conditions are more prevalent among pedigree cats. This increased occurrence is attributed to selective breeding and in-breeding practices aimed at enhancing specific breed characteristics, which may inadvertently elevate the risk of inherited disorders. In fact, certain breeds may even be defined by an inherited disorder that could pose health risks.

A genetic disease refers to a condition arising from one or more abnormalities within the genome. In felines, these diseases can either be congenital, appearing at birth, or develop as the cat ages. Although mixed-breed cats can also inherit genetic disorders, the likelihood is considerably greater in purebred populations due to several factors:

- **Founder Effect:** Breeds are frequently established from a limited number of 'founder' cats, whose genetic traits, including harmful mutations, become widespread within the breed.
- **Popular Sire Effect:** A single, highly regarded male cat may be extensively utilized for breeding, leading to the rapid dissemination of his genetic traits—both advantageous and detrimental—throughout the breed.
- **Genetic Bottlenecks:** Events that significantly diminish a population's size, further constraining genetic diversity.

Comprehending these diseases is essential for veterinarians, breeders, and pet owners, enabling them to make informed choices that enhance the health and welfare of future generations of cats [1].

Genetic testing of cats

Recent advances in genetic investigation and testing have meant that it has been possible to identify the gene defects associated with a number of inherited conditions, confirm their genetic basis, and also in many cases develop diagnostic tests to identify affected cats (and occasionally 'carrier' cats). Although some diseases have a simply genetic basis with the disorder being determined by a single pair of genes, in other disorders that may have an hereditary component, the inheritance may be much more complicated. Multiple genes can be involved in some disorders (polygenic) and there may be a combination of genetic

and environmental effects (multifactorial) in others. Single gene disorders are much easier to characterize, investigate and develop diagnostic tests for. With different populations of cats in different parts of the world, some genetic disorders may be seen more commonly, or even exclusively, in some geographical locations. However, with international travel of both owners and cats, many disorders are seen worldwide (although the frequency of disease may vary in different regions). Many different veterinary diagnostic laboratories offer genetic (DNA) tests for different animal diseases. Many laboratories offer DNA testing for cat coat colours and cat parentage, as well as tests for inherited disorders.

Precautions

When undertaking DNA testing of cats (to determine whether they are suitable for a breeding programme) a veterinarian should always be present to supervise, and a reputable and reliable testing laboratory should be used. *International Cat Care* believe that whenever genetic tests are run on cats for the selection of breeding stock, the gene test result should be linked to a method of permanently identifying the cat that has been tested (e.g., a standard, internationally recognized microchip number), and that a vet should collect the sample (blood sample or cheek swab) so that the identification (microchip number) can be verified and recorded on the submission form and result.

Types of Feline Genetic Diseases

Here are some of the most common genetic diseases in cats:

Polycystic Kidney Disease

Polycystic kidney disease (PKD) is an inherited disorder that can lead to feline kidney failure. In this disease, small fluid-filled cysts are formed in the feline kidneys. *PKD is the most common single-gene feline disorder identified so far. Affecting 6% of cats worldwide, this condition is present in 38% of Persian cats.* It is also very frequent among Persian-derived breeds such as Himalayans. Affected cats are always heterozygous for the gene causing for PKD, as the gene in homozygous form is lethal

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common feline heart disease. HCM causes the left ventricle of a cat's heart to thicken, which decreases the space available for blood to flow. This means that a smaller amount of blood is pumped through the cat's body with each heartbeat compared to a healthy cat. In order to compensate, HCM cats have an increased heart rate (tachycardia). Tachycardia and heart rhythm abnormalities can be a clue that your veterinarian should test your cat for HCM. HCM can be caused by several genetic mutations. In Maine coon cats a mutation in the myosin-binding protein C gene has been identified and it occurs in 33% of cats of this breed. This mutation is autosomal dominant. In 20% of Ragdolls different mutations have been identified to cause the condition. A genetic test is available for breed-specific mutations for HCM in both of these breeds

Diabetes Mellitus

Diabetes mellitus (DM) is a metabolic disorder in which blood sugar levels remain high for long periods of time. This occurs when there isn't enough insulin being produced in the body or the body isn't responding to it. Without insulin sugar from the blood can't be transmitted to the target cells and tissues. As a consequence, despite high blood sugar levels, the body cells and tissues are starving. Luckily, diabetes can be successfully treated. This condition is commonly diagnosed among felines, interestingly, primarily random-bred cats. However, according to some studies, some breeds such as Burmese, Siamese, Norwegian forest, Russian blue and Abyssinian seem to be at a higher risk from diabetes.

Feline Lower Urinary Tract Disease (FLUTD)

Feline lower urinary tract disease or FLUTD is a term used to address multiple conditions associated with the lower urinary tracts in cats. It includes bladder inflammation (cystitis), urethra inflammation, a formation of urinary stones in the bladder (urolithiasis) and urethral obstructions. *Feline idiopathic cystitis* and *>feline urologic syndrome* have been observed as the most common feline hereditary predisposition in practice. According to studies (Dorsch R et al, 2014), (Defauw PA et al, 2011) and (Stella J et al, 2013), FLUTD affects about 1-2% of domestic cats. Persians seem to be at a higher and Siamese at a lower risk from FLUTD

Progressive Retinal Atrophy

Progressive retinal atrophy (PRA) is a genetically determined condition that causes blindness in mid to late life in cats. Two genes have been identified to be associated with the condition. The identified genes are associated with Abyssinian, Somali and Ocicat breeds. The genes are recessive, which means only homozygous carriers are affected. Unfortunately, the condition isn't treatable, but there are many ways in which we can help blind cats have a happy and satisfying life.

Deafness

Deafness is a relatively common condition among white cats, specifically white cats with one or both eyes blue in color. The dominant allele *W* of *the white masking gene* causes disrupted replication and migration of melanocytes into the skin. All the cats carrying this allele (*W/W* and *W/w*) will appear white, even if they

carry other color genes. Only recessive homozygotes (w/w) will express the normal pigmentation. Carriers of this W allele are at a much higher risk from deafness in comparison to non-carriers. This is because this gene doesn't only interfere with the coat (and eye) color but also with the development of structures within the ears and can occasionally cause the degradation of the cochlea (a part of the inner ear involved in hearing).

Glycogen Storage Disease (GSD)

This group of inherited disorders primarily affects Norwegian Forest Cats. GSD hampers the cat's ability to break down and utilize glycogen, stored energy. Affected cats may show muscle weakness, exercise intolerance, and tremors. As the disease advances, it could lead to severe heart and liver conditions, reducing the cat's lifespan.

Mucopolysaccharidosis (MPS)

MPS is a group of inherited metabolic disorders that occur most frequently in Siamese cats and related breeds. Affected cats are deficient in certain enzymes that help break down and recycle molecules called glycosaminoglycans. This results in abnormal accumulation of these molecules in the cat's body, leading to various symptoms, including facial deformities, vision problems, dwarfism, and a reduced lifespan.

Spinal Muscular Atrophy (SMA)

SMA is a genetic disorder specific to Maine Coon cats. This disease damages the neurons in the spinal cord that communicate with muscles, leading to progressive muscle weakness and wasting, especially noticeable in the hind limbs. Despite these physical limitations, cats with SMA can have a normal lifespan and good quality of life with the right care and attention. They typically remain playful and active, though they may develop a distinctive sway or hop in their gait due to muscle weakness.

Causes of Genetic Diseases in Cats

Here are some examples of the common reasons why certain breeds of cats have genetic diseases

Inheritance of Abnormal Genes: Direct Inheritance: One of the most common ways genetic diseases are passed in cats is through direct inheritance, where a disease-causing gene is transmitted from one or both parents to their offspring. Some genetic disorders are recessive, meaning a cat must inherit the disease-causing gene from both parents to develop the disease. Other disorders are dominant, meaning that only one copy of the abnormal gene is needed for the cat to be affected. Still, other conditions are sex-linked, which means the abnormal gene is carried on one of the sex chromosomes (X or Y).

Mutations: Mutations, or changes in DNA, can occur spontaneously and lead to genetic disorders. These mutations can occur in any cell of the body, but those that happen in the reproductive cells (sperm or egg) are the ones that can be passed on to offspring. Some genetic disorders are caused by such spontaneous mutations, which may occur for reasons that are not well understood. For instance, Glycogen Storage Disease (GSD) in Norwegian Forest Cats is likely a result of spontaneous mutation. Although it is a recessive disorder, meaning that the cat needs to inherit the gene from both parents to develop the disease, the gene mutation that causes this disease could have initially occurred spontaneously in a cat's DNA.

Environmental Triggers

Epigenetics Epigenetics refer to gene expression or cellular phenotype changes caused by mechanisms other than changes in the underlying DNA sequence. In other words, it's the study of how your behaviors and environment can cause changes that affect the way your genes work. These changes might be temporary or could last and be passed on to offspring. For example, it's hypothesized that environmental factors such as exposure to certain chemicals or stress could trigger epigenetic changes, leading to diseases such as Feline Lower Urinary Tract Disease (FLUTD) in susceptible breeds like Persians.

Exposure to Risk Factors In some cases, exposure to certain environmental risk factors can interact with a cat's genetic predisposition to trigger the disease. For instance, a cat may be genetically susceptible to a condition like Hypertrophic Cardiomyopathy (HCM). Still, the disease may only manifest if the cat is exposed to certain stressors or environmental conditions.

Breeding Practices

Inbreeding Inbreeding, or mating between close relatives, can increase the risk of genetic disorders in offspring. This is because close relatives are more likely to carry the same harmful genes, and mating between such individuals can increase the chances of these genes being passed on. Certain purebred cats are particularly prone to specific genetic diseases due to extensive inbreeding.

For instance, inbreeding in breeds like Maine Coon and Ragdoll is one of the primary reasons for the higher incidence of diseases like Hypertrophic Cardiomyopathy (HCM) and Spinal Muscular Atrophy (SMA).

Selective Breeding Selective breeding for certain traits can inadvertently promote the passing on of harmful genes. Breeders might select a particular physical trait without realizing that the gene responsible for that trait is linked to a disease-causing gene.

For instance, the gene causing the unique coat color pattern in Siamese cats is linked to the gene causing Mucopolysaccharidosis (MPS). Therefore, breeding for the coat color inadvertently increases the prevalence of MPS in this breed.

DIAGNOSTIC TESTS FOR COMMON GENETIC DISEASES IN CATS [2]

Physical Examination

Diagnosing genetic conditions in cats typically begins with an exhaustive physical evaluation. By assessing the cat's physical condition and behavioral patterns, veterinarians may identify symptoms indicative of certain inherited diseases, especially if it's a pedigree cat. These symptoms could include cat size or shape abnormalities, unique physical traits, or apparent signs of discomfort. Nevertheless, relying solely on physical examination is only partially definitive since various health issues, not just genetic ones, can present similar symptoms.

Genetic Testing

The most accurate method to diagnose genetic diseases and traits is through a DNA test. In this process, DNA sourced from the cat's blood or other tissues is scrutinized in a laboratory setting to detect mutations causing the disease. Such genetic tests available today can identify many genetic diseases in cats, including Polycystic Kidney Disease (PKD), Hypertrophic Cardiomyopathy (HCM), and Glycogen Storage Disease (GSD). The findings from these tests can help confirm a diagnosis, establish the cat's carrier status, or even forecast the potential of the cat developing a specific disease later in its life.

Biopsy

Occasionally, performing a biopsy may be necessary to diagnose a genetic condition. This procedure involves extracting a small tissue sample from the impacted organ or body part. This sample is then microscopically examined for abnormal cells or structures. Biopsies prove particularly beneficial when diagnosing genetic diseases that trigger tumors or skin afflictions.

Blood Tests and Other Laboratory Tests

Blood tests, including those determining the blood type and other laboratory assessments, contribute to the diagnosis of inherited diseases. For instance, blood tests can expose blood cell or chemistry irregularities, suggesting certain genetic defects. Urine examinations, fecal analysis, and other lab tests offer supplementary data about the cat's holistic health and the performance of its various organs.

Medical History

Collecting a comprehensive health history forms another pivotal step in diagnosing inherited diseases in the cat population. The vet will inquire about your cat's medical background, encompassing past illnesses and treatments. Information about the health histories of the cat's parents and siblings can also prove useful, providing insights into the probability of inherited diseases.

Imaging Tests

Imaging tests, such as X-rays, ultrasounds, and CT scans, are commonly employed to diagnose genetic diseases that impact the cat's physical structures. For instance, X-rays can highlight skeletal defects, whereas ultrasounds can uncover issues with organs like the heart or kidneys. These tests can often pinpoint physical indicators of a disease even before the cat starts showing symptoms and are identified in cats across the feline population.

Electrodiagnostic Testing

Electrodiagnostic tests, including electrocardiograms (ECG) or electromyography (EMG), can diagnose genetic diseases affecting the nerves or muscles. These tests gauge electrical activity to establish whether these tissues are functioning optimally. For instance, an ECG can identify heart conditions such as Hypertrophic Cardiomyopathy (HCM).

Treatment for Common Genetic Diseases in Cats

The treatment options for genetic diseases in cats largely depend on the specific disease. However, most treatments aim to manage symptoms, slow the progression of the disease, and improve the cat's quality of life. Let's look at some general categories of treatment options:

MEDICATIONS [3]

Heart Medications: Cats with heart diseases such as Hypertrophic Cardiomyopathy (HCM) might be given ACE inhibitors that reduce the heart's workload and lower blood pressure. Beta-blockers may also be used to control the heart rate and reduce the heart muscle's thickening. Anticoagulants like aspirin or clopidogrel might be prescribed to prevent blood clot formation, a common and dangerous complication of HCM.

Kidney Medications: Cats with Polycystic Kidney Disease (PKD) might receive ACE inhibitors or angiotensin receptor blockers (ARBs) to control high blood pressure. Drugs that decrease phosphorus levels, increase calcium levels, or help with anemia might also be used to manage symptoms of kidney disease.

Dietary Management

Kidney-Friendly Diet: This diet for cats with PKD is typically low in phosphorus and protein to reduce the kidneys' workload. It also usually contains increased levels of B vitamins and antioxidants to help the body deal with the toxins that the kidneys can't filter out as effectively.

Diabetic Diet: A diet for cats with diabetes usually involves high protein and low carbohydrate content to minimize blood sugar spikes after meals. Consistent feeding schedules and portion control are essential to managing feline diabetes.

Regular Monitoring and Check-ups

Regular vet visits allow for ongoing evaluation of the cat's health and adjustments to the treatment plan as necessary. This could include:

Blood Tests: Blood tests can provide information on organ function and detect complications early.

Ultrasound or X-rays: These imaging tests can help assess the disease's progression. For example, echocardiograms (ultrasound) are useful for monitoring HCM.

Genetic Counseling

Genetic counseling can help cat owners and breeders understand the potential risks of various genetic diseases. This might include:

DNA Testing: DNA testing can identify cats that carry genes for specific diseases. This is particularly important for breeders to prevent breeding cats with harmful genetic traits.

Physical Therapy Physical therapy can help improve strength and mobility, particularly for diseases that impact the cat's musculoskeletal system. This could involve:

Stretching and Massage: Gentle stretching and massage can help maintain muscle flexibility and reduce stiffness in cats with Glycogen Storage Disease (GSD).

Hydrotherapy: Some practices may offer hydrotherapy, which can be beneficial for maintaining muscle mass without putting stress on the joints.

Supportive Care Supportive care aims to improve the cat's quality of life. For example:

Sensory Enrichment: For cats with progressive conditions like Progressive Retinal Atrophy (PRA), toys that stimulate other senses, like sound or smell, can keep a cat engaged and happy.

Safe Environment: For cats with PRA, removing potential hazards, keeping furniture in the same place, and using night lights can help them navigate their environment safely.

Heart Surgery: In rare cases, surgery might be recommended for cats with certain types of heart disease. This could involve procedures to widen a narrowed artery or to implant a pacemaker.

Preventive Measures for Common Genetic Diseases in Felines Prevention of hereditary diseases in cats is primarily focused on prudent breeding practices. Numerous genetic abnormalities are inherited and transmitted from parent to offspring. Here are some steps that can be taken:

Genomic Screening and Accountable Breeding: Many inherited disorders can be identified through genetic testing. Breeders ought to have their breeding cats screened for known hereditary diseases. If an affected cat is identified as a genetic disease carrier, that cat should be excluded from breeding. Some breeders may elect only to mate cats with two healthy copies of each gene, thus guaranteeing that their kittens cannot inherit these congenital conditions.

Frequent Veterinary Visits: Not all genetic disorders can be completely averted despite responsible breeding. Regular veterinary medical care is critical to identify any potential health problems early. Early diagnosis typically results in more effective management of the clinical disease. Some genetic disorders don't manifest symptoms until the cat is several months or even years old, making regular check-ups critical even for seemingly healthy domestic shorthair cats or other breeds.

Nutritionally Balanced Diet and Regular Physical Activity: A balanced diet and ensuring your cat exercises regularly can help maintain overall health and strengthen the immune system. This can make them less susceptible to complications from genetic diseases.

Avoidance of Inbreeding: Inbreeding can heighten the likelihood of genetic disorders. Mating closely related cats can amplify the chances of passing on harmful genetic markers to kittens.

Lastly, it's vital to remember that not all cat health issues, including irritation caused by various factors, are preventable. As a cat owner, offering your feline companion a secure, caring environment and high-quality veterinary care is best.

CONCLUSION AND FUTURE DIRECTIONS

Feline genetic diseases represent a significant challenge to animal welfare, particularly within the purebred community. However, increased awareness, widespread genetic testing, and a commitment to responsible breeding practices offer a powerful solution. The collaboration between veterinarians, geneticists, and dedicated breeders is essential to reduce the prevalence of these disorders.

Future directions in the field hold even more promise. Gene therapy, where a functional copy of a gene is delivered to replace a defective one, is being explored. Technologies like CRISPR-Cas9 offer the potential for precise gene editing, although their application in clinical veterinary medicine is still in its infancy and carries significant ethical considerations.

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