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Brianna Yi is a senior majoring in Animal Science, and she aspires to become a veterinary surgeon. Her passion for animal welfare grew while working as a Professorial Assistant for Dr. Jacquelyn Jacobs, where she contributed to research on shelter dog behavior. This experience, coupled with her interests in genetics, inspired Brianna to write literature reviews on canine and feline genetic diseases. Through her work, she aims to raise awareness of these conditions and promote responsible pet ownership and thoughtful consideration when choosing specific breeds.

Feline Polycystic Kidney Disease in the Domestic Cat

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Abstract

Polycystic kidney disease (PKD) is a hereditary condition that affects a variety of mammals, including humans and felines. Feline polycystic kidney disease (FPKD) has become one of the most common feline genetic diseases, with higher rates of diagnoses being seen among certain breeds, such as the Persian cat. The significant sign of this disease is the formation and growth of fluid-filled cysts on the kidney and can include other organs, such as the liver or pancreas. Without awareness and rigorous tracking of genetic carriers, cats will continue to be born with and succumb to FPKD, eventually resulting in kidney failure. Additionally, cats with FPKD are theorized to be ideal models for human PKD. Therefore, by understanding the feline condition, it is hoped that human PKD research can also progress (Lyons, L., et al. 2004.). There is currently no cure for this disease; however, dietary and lifestyle changes are used to increase quality of life and comfort. Understanding this disease and its mechanisms is crucial to decreasing the number of affected felines (Schirrer, L., et al. 2021.).

Introduction

Feline polycystic kidney disease (FPKD) is a genetic disorder characterized by the development and progressive growth of cysts on the kidneys. These cysts eventually lead to renal dysfunction and kidney failure. This condition is most commonly seen in Persian cats, making it a significant concern for breeders and owners especially due to the autosomal dominance. To manage the disease and improve quality of life, early detection is crucial. This paper explores the genetic causes, progression, diagnosis, and management of FPKD, as well as its implications on cat breeding.

Genetic Causes

The cause of feline polycystic kidney disease has been linked to the autosomal dominant transmission of a mutated PKD1 gene (Lyons, L., et al. 2004.). The normal PKD1 gene encodes protein polycystin-1, which covers the membrane of the kidney and aids in molecular interactions with proteins, fats, and other cellular components. Mutated PKD1 originates from insertions or deletions of base pairs, resulting in incomplete and nonfunctional polycystin-1. The cyst-causing mechanisms of the mutation are currently unknown. However, the prevalent hypothesis is that nonfunctional polycystin-1 disrupts normal cellular signaling. Disrupted signals subsequently cause abnormal growth and division of renal tubule cells, creating cysts on the kidney's membrane surface (National Library of Medicine, n.d.).

The autosomal dominant nature of this mutation means that only one copy of the defective gene is needed for the manifestation of PKD. A kitten born from a litter with a copy of mutated

PKD1 is guaranteed to be diagnosed with polycystic kidney disease in the future due to FPKD's complete penetrance (Schirrer, L., et al. 2021.). Because of its dominance, specific breeds such as Persians and Exotic Shorthairs have higher prevalence of polycystic kidney disease due to the frequency of mutated PKD1 within these populations as a result of breeding practices. In a French study, 41.8% of Persians and 39.1% of Exotic Shorthairs carried mutated PKD1 (Barthez, P., Begon, P. 2003.).

Progression of Feline PKD

Feline polycystic disease is a progressive condition. At birth, affected kittens may already display cysts on their kidney but appear to be asymptomatic. Cases differ, however, and a confirmed diagnosis can be given as early as 6 months of age. Most clinical signs and symptoms of FPKD are recognized in adulthood at around 7 years old (Cornell Feline Health Center. n.d.). This is a challenge of FPKD, as late diagnosis reduces available treatment options. As the cat ages, the cysts on the kidney begin to grow and disturb normal biological functions due to healthy tissue being replaced by cystic growth. Signs are synonymous to normal kidney disease; specifically, an increase in thirst and urination, loss of appetite, weight loss, vomiting, and lethargy are common indicators of decreased kidney function. During a physical exam, the veterinarian may also be able to see the kidney's outline while the cat is lying on its back (Cornell Feline Health Center. n.d.).

Unfortunately, most symptoms appear when the kidney has already suffered severe damage. Approximately 60-70% of kidney function is lost by the time FPKD is diagnosed in a cat (Barthez, P., Begon, P. 2003.). In the final stages of PKD, the cysts continue to grow and renal failure – as well as uremia – are inevitable. Chronic kidney disease (CKD) is often diagnosed alongside the final stages of PKD. Severe CKD is characterized by mouth ulcers, poor fur, bad breath, and lethargy (Weir, M., et al. n.d.).

Methods of Diagnosis

Testing for FPKD can be done through ultrasound or genetic testing. Genetic testing is conducted with either blood or saliva samples to identify mutated PKD1 in the genome. When tested against the definitive diagnostic method of ultrasounds, genetic testing showed accurate and similar results. Undergoing genetic screening for a cat may also reveal the cat as a carrier of mutated FPKD (Bonazzi, M., et al. 2008.). Kittens that have not yet been weaned are tested through a blood sample as the milk of the queen can give inaccurate results in a saliva test. Genetic screening is highly recommended for cats prone to FPKD, such as Persians (International Cat Care. 2018.). However, genetic testing still remains as a work in progress, and is a costly and novel technology in many veterinary clinics (Lyons, L. 2010.).

An ultrasound is an alternative diagnostic test for cats who have not undergone genetic screening to search for the presence of kidney cysts before more severe clinical signs appear. It is also used to definitively diagnose FPKD. Specifically, B-mode ultrasonography yields high quality images of kidney size and shape, as well as the size of the cysts present on the kidney's surface. Cysts at least 2 mm in size can be detected through this method (Debruyn, K., et al. 2012.). Ultrasound also serves as a reliable method of tracking cyst growth and formation after a FPKD diagnosis.

Management and Prevention

FPKD currently remains as an incurable condition. However, proper management can preserve kidney function and provide as much comfort as possible for the affected cat. One such

management strategy is dietary changes. Veterinarians recommend a low protein and phosphorus diet since it reduces the load the kidney must undertake to process the nutrients in food. Diets such as these minimize the kidney's waste production, ultimately increasing longevity. High blood pressure may also result from decreased kidney function, so it may be necessary for a cat to be prescribed blood pressure medications (Quimby, M. 2015.). Fluid therapies are also effective in the management of FPKD (Cornell Feline Health Center. n.d.). Additional fluids aid in preventing dehydration, especially if chronic kidney disease is diagnosed in addition to FPKD. Subcutaneous fluids may also improve the cat's appetite and minimize constipation (Polzin, D. 2013.).

Prevention heavily relies on breeders genetically testing their cats for the PKD1 mutation if the cat breed is susceptible to FPKD. As the PKD1 mutation is autosomal dominant, cats should be tested before being considered for breeding purposes. This ensures that the PKD1 mutation will not be passed down over generations with the goal of eventually drastically decreasing the number of PKD1 cats in the general population (Cornell Feline Health Center. n.d.).

Conclusion

Feline polycystic disease is an incurable, progressive disease that results from the dominant genetic mutation of the PKD1 gene. Certain breeds are more susceptible than others, such as Persians. Testing a susceptible cat early through a genetic test is highly recommended so that management of the disease can begin as soon as possible. Additionally, it is crucial that breeders genetically test their cats prior to breeding to prevent litters that carry the mutation. Despite the disease not having a cure, being aware of early signs and following a variety of management strategies can aid in providing an FPKD cat with comfort and aid in extending its lifespan.

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