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A Study of APKD in Cats

Autosomal polycystic kidney disease is not an uncommon occurrence in felines; in fact, it has become one of the most common feline diseases to occur. This disease typically occurs in cats who are considered senior age (7 years old and older). Many of these felines who suffer from autosomal polycystic kidney disease are Persians, or breeds that are genetically similar to Persians. Because of the prevalence of autosomal polycystic kidney disease in Persians, studies from around the world narrowed down research specifically to Persian breeds to better understand how autosomal polycystic kidney disease (APKD) forms. A particular group of researchers (A. Ghavihelm, S. Jamshidi, and others) noticed that there was not a study of this disease in Persians in Iran, so they took this opportunity to identify the formation of APKD in Persian breed cats residing in that country.

For context, kidney disease has been known to be resultant from a mutation in a gene known as PKD1. This particular gene was studied to confirm if there were any mutations in it, and if so, where the mutation was occuring. An important thing to know about the PKD1 gene is that it has a very important protein called PC1 (short for polycystin-1). PC1 plays a vital role in the maintenance of the overall structure and function of the kidneys, as well as other vital organs. Without the PC1, cell apoptosis can occur and cysts can form in the kidneys. The more apoptosis and cyst formation, the greater the presence of kidney disease. This article mentions that lack of PC1 function can also result in cyst formation in the liver, but the main focus of this study was on the kidneys.

The lack of PC1 function results from a point mutation in the PKD1 gene. This point mutation causes a stop codon to occur in the PC1 protein, therefore causing the PC1 protein to stop forming.

To start this study, researchers examined 47 healthy Persian cats, all who were under 7 years old. These researchers took buccal swab samples from each cat, and the DNA from those swabs were extracted and used to perform a PCR test. The point of the PCR test was to determine if and where the mutation in the PKD1 gene resides. A control group was used to better identify the PKD1 gene mutation; Ghavihelm, Jamshidi, and others used DNA samples from two cats who suffered from polycystic kidney disease, and used those findings from the PCR to compare with PCR findings from the healthy Persian cats. The testing showed that 46.8% (or 22 out of the 47 Persian cats) tested heterozygous for the PKD1 gene.

Findings prior to this study show that there is a similarity between APKD in cats and APKD in humans. The difference with humans is that there are many other genes that play a role in kidney disease; one of these genes, however, is PKD1, which is the gene that causes kidney disease in cats. In a sense, identifying the point mutation in the PKD1 gene in cats can help researchers better understand autosomal polycystic kidney disease in humans as well.

In conclusion, autosomal polycystic kidney disease is sadly a common disease in many cats. Using this study specifically from Persian and breed similar feline in Iran, researchers were better able to understand how and why APKD forms. Although there is no current cure for kidney disease in felines or humans, being able to better understand the role of the PKD1 gene and the PC1 protein can potentially be a good first step towards better understanding of various treatment options for APKD.

Cited Source

Ghavihelm, A., Shahram, J., Tamai, I., Zangisheh, M. Molecular detection of polycystic kidney disease in Persian and Persian-related breeds in Iran. *Journal of Feline Medicine and Surgery Open Reports.* **8**, https://doi.org/10.1177/20551169211070991. (2022).