Studies of Renal Dysplasia in the Cairn Terrier

Progress Report: September 23, 2024

Renal dysplasia (RD) in Cairn Terriers has been the subject of our studies, primarily focusing on its genetic basis, clinical presentation, and management strategies. Here are some summary points from our research:

1. **Genetic Studies**: Our research has aimed to identify specific genetic markers associated with renal dysplasia in Cairn Terriers. We utilized pedigree analysis to trace hereditary patterns, suggesting a potential autosomal recessive inheritance, meaning that both parents contribute to the disease in affected puppies. We also have some evidence suggesting that there is a main (primary) gene that causes RD and some modifier (minor) genes that make the disease milder or more severe.

2. **Clinical Manifestations**: Our studies have documented the clinical signs of renal dysplasia in affected Cairn Terriers, which can include growth retardation, polydipsia, and recurrent urinary infections. Histopathological (microscopic) examinations often reveal abnormal renal architecture, such as dysplastic glomeruli and interstitial fibrosis.

3. **Diagnostic Approaches**: Imaging studies, like ultrasound, are frequently used to evaluate kidney structure in Cairn Terriers. In our publication, we have emphasized the importance of early diagnosis to manage the disease effectively, often recommending routine screening in breeds predisposed to renal dysplasia.

4. **Long-term Prognosis**: Some studies have tracked the long-term health outcomes of Cairn Terriers diagnosed with renal dysplasia. These findings indicate a variable prognosis, with some dogs managing well with appropriate dietary and medical interventions, while others may progress to renal failure. The outcome is also very dependent on the severity of the RD in each individual.

5. **Breeder Awareness and Education**: Our studies have highlighted the need for increased breeder awareness about the signs of RD and the importance of genetic testing in breeding practices to reduce the prevalence of the condition.

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These studies collectively emphasize the need for further research into effective treatment options and breeding strategies to mitigate RD in Cairn Terriers. The aim of this research project has been to investigate the heritability of renal dysplasia in the Cairn terrier breed. We have also sought out the degree to which genetics influence the severity of disease - whether it is mild, moderate, or severe – and the degree of relatedness between renal dysplasia and aplasia. Upon examination of several pedigrees, we have assessed the familial ties of over 2000 individuals, 1572 of which could be represented on a single pedigree. Within the population of dogs involved in the study, there are 34 Cairns that have ultrasonographic changes of the kidney that are consistent with renal dysplasia. 23 of the 31 affected animals have been directly linked to a single common ancestor. Parentage information has not yet been provided for 7 of the 8 remaining affected individuals; therefore, their relation to the aforementioned common ancestor has not yet been evaluated.

Scans have been performed at the University of Pennsylvania, Ontario (Canada), Texas, Virginia, Maryland, at the Montgomery County Dog Show and at many others. A review of the data shows clearly that the number of even mildly affected dogs has decreased to the point at which we only see one or two a year. This is largely due to the fact that the breeders affiliated with the CTCA take testing seriously. This not only helps the individual breeding programs but also our research to discover the genes causing renal dysplasia.

Lastly, our research has shown that it is important to wait until the puppies are at least 12 weeks old for their kidney scans, as younger dogs have less developed kidneys, which may lead to erroneous results. The research has also shown that mild speckling seen in some of the kidneys is not correlated with future clinical signs, but we know that it is linked to renal dysplasia. However, if there is moderate speckling or more, we do recommend placing such dogs on renal diets to avoid any unneeded stress on the kidneys. Yearly blood rechecks to assess kidney values are recommended.

What do we need to move forward? We need more DNA samples, - that means we need more blood from dogs with abnormal scan results. This would be for genome wide association study (GWAS), which costs $100 per sample but we need to submit in multiples of 12 ($1,200). Another affected 12 dogs (no matter the severity) would be ideal, but we realize that may not be possible. Another option would be to whole genome sequence (WGS) 2 affected dogs ($2,400). While this is more costly, it will

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probably lead us the answer quicker. The difference between GWAS and WGS is that the GWAS gets you to the area of where the gene may be found but then you still have to do sequencing. The WGS allows you to directly find the mutation that is causing disease. A combination of both is most ideal. We anticipate that $5,000 (this includes materials to make DNA) is sufficient to finally find the causative gene(s).

Margret Casel, Dr med vet, PhD
Professor of Medical Genetics
University of Pennsylvania School of Veterinary Medicine