

## Cystinuria and DNA Research

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### *CYSTINURIA IN IRISH TERRIERS*

Cystinuria (CU) in Irish Terriers appears to be on the rise in North America. It is an inherited disease that impairs cystine reabsorption in the kidney which can lead to the formation of cystine stones in the bladder, blockage of urine flow, kidney failure and death. Because it is a genetic recessive trait, it remains mostly hidden in the population and although only a few dogs may exhibit symptoms, far more are carriers, passing the gene mutation on to future generations. At present, there is no genetic test for the disease, so there is no way to identify carriers.

New technology has been developed which makes the development of a genetic test quicker and less expensive. It does depend, however, on having DNA samples from a sufficient number of affected dogs and close relatives. The DNA can be analyzed to identify the mutation responsible so a genetic test can be developed. Once we have a DNA test, we can breed *any* dog to an appropriate mate with no risk of producing diseased puppies. This way we can maintain good dogs in the gene pool and eventually eliminate the disease entirely.

Test development is done in the lab of Dr. Gary Johnson, a lifetime member of the ITCA, and he has offered to cover the cost (\$7,500) of testing samples from three selected dogs. What we have to do is provide DNA samples from as many affected dogs and close relatives as possible. We can do this by submitting blood and urine samples to the Canine Health Information Center.

DNA samples from all Irish terriers are encouraged but those with confirmed cystinuria are particularly important, along with their close relatives. For those dogs, pedigrees to show the relationships and full clinical/diagnostic information should be submitted along with the sample. For individual submission instructions, go directly to the CHIC DNA repository website (<http://www.caninehealthinfo.org/dnabank.html>). Fill out the application form to get a blood collection kit. The sample will go directly to Gary Johnson's lab.

From the submissions, Dr. Johnson will select three dogs most likely to provide candidate genes for developing a DNA test for cystinuria. The availability of dogs from different family trees will help reduce the number of common variants to eliminate. The more DNA samples we can provide, the easier it will be to develop a DNA test for cystinuria. Once the test is developed, we will be able to breed with confidence so we can maintain the size of our gene pool and keep desirable traits while avoiding the risk of producing affected dogs and eventually eliminating the disease.