A Pathway?
Or a Superhighway?

by Debby Rothman

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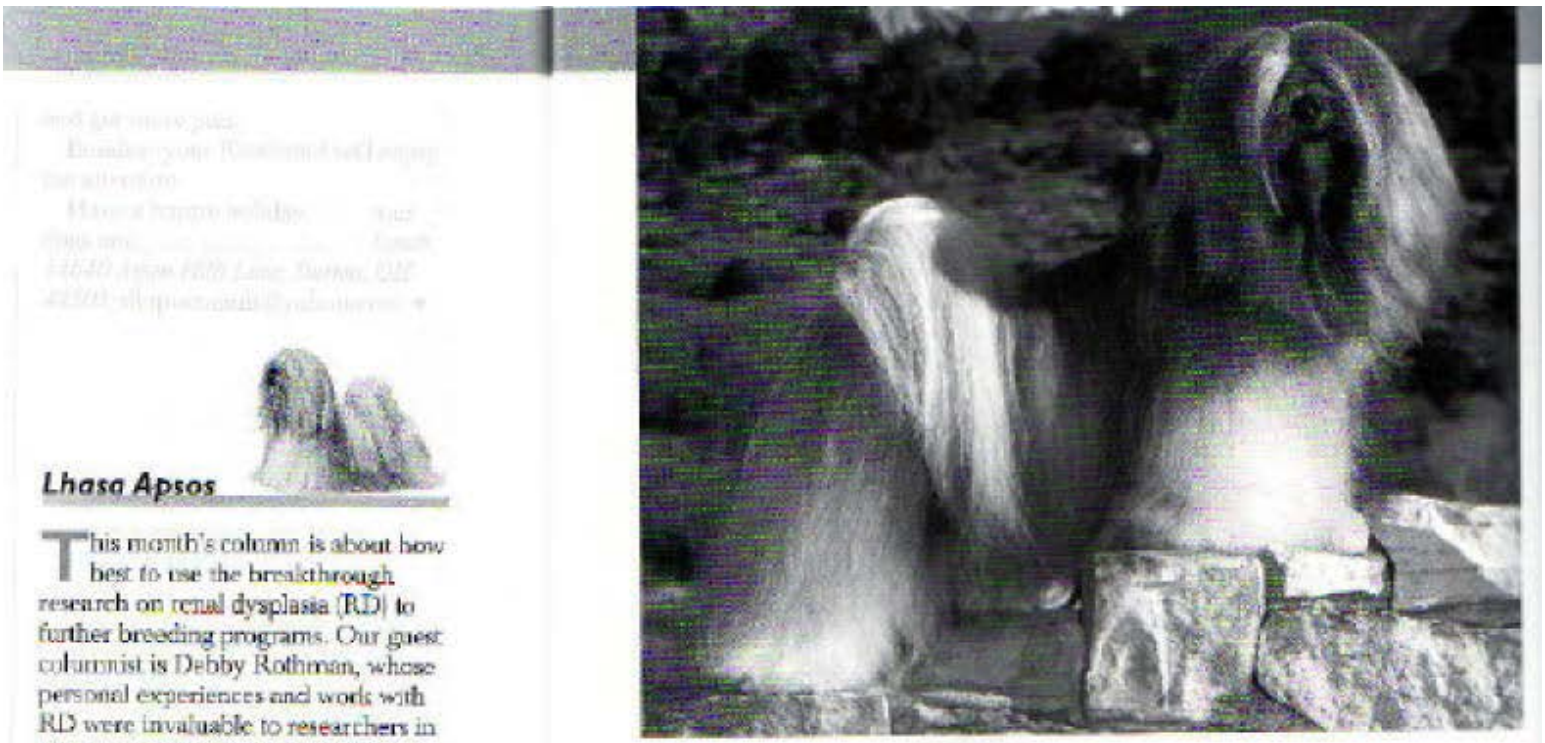
Author's note: Continued research led to the discovery of a stronger mutation relating to the development of fetal glomeruli. The Dogenes RD test for Lhasa Apsos is for this mutation.



Lhasa Apso

This month's column is about how best to use the breakthrough research on renal dysplasia (RD) to further breeding programs. Our guest columnist is Debby Rothman, whose personal experiences and work with RD were invaluable to researchers in identifying mutations responsible for the disease through DNA testing.

*A PATHWAY? OR A SUPER-HIGHWAY?

So, now what? We've got a valuable tool to help prevent future puppies from dying of renal dysplasia. Super! Shouldn't breeders simply test potential breeding dogs and eliminate those positive for mutation A? This would certainly seem like a responsible approach—but is it?

Misuse of DNA test results has placed at least two other breeds in further jeopardy, according to Dr. Jerold Bell, a leading researcher at the Tufts University School of Veterinary Medicine. Could the Lhasa Apso be the next?

Dr. Mary Whiteley, of Dogenes, Inc., discovered two mutations responsible for renal dysplasia in the Shih Tzu. Mutation A is dominant with incomplete penetrance. Mutation B is recessive. She continues to look for mutation B in the Lhasa Apso: Color mutation A does exist in the gene but can pass on the trait with varying degrees of clinical manifestations to their offspring. This can range from normal to a positive biopsy with no clinical signs of kidney dysfunction, to severely affected. These dogs are genetically affected, but not always phenotypically affected. In other words, many—if not most—of these dogs live long, normal lives.

Identifying these dogs was previously a problem because the only accurate method was a specialist-read wedge biopsy of the kidney. Now these dogs can be revealed with a simple DNA cheek swab test. However, new additional information presents different issues. What if breeding animals test positive? What if puppies test positive? Should breeders eliminate those dogs from breeding programs? What about placing puppies that carry mutation A? Should breeders place those puppies in pet homes?

What works best? Make sure one of the parents is genetically clear [biopsy normal]; select—if possible—genetically clear offspring; and as Dr. Bell recommends, breed and replace, breed and replace.

What about pet puppies who test genetically affected? Obviously, if they're showing symptoms, a responsible breeder won't place them in unsuspecting homes. The typical tests—BUN, creatinine, and specific gravity—are of little value in determining if the kidneys are compromised. An ultrasound of the kidneys will reveal if the kidneys are of proper size and shape. Most likely these Apsos will live a normal life span and can be placed in loving homes.

To learn more about renal dysplasia visit, *www.fleetfeetlabs.com*.

Thank you, Debby. *–Cassandra de la Rosa, 7372 71th Court SW, Olympia, WA 98502; dlrosa@msn.com* ✦

pool. Dogenes, Inc., now offers a DNA cheek swab test for RD mutation A.

What does dominant with *incomplete penetrance* mean? Mutations that show variable degrees of clinical symptoms (variable expression of the gene) are said to be inherited with incomplete penetrance. This means that individuals with one or two copies of mutation A can appear to be normal.

Having wedge-biopsied a large number of Apsos for over 10 years, having known this now-available additional information, having dealt with the above issues, I do not believe we need to be alarmed. Because mutation A proved to be dominant with incomplete penetrance, the biopsies did indeed give the information now available with a simple cheek swab. I am 10 years farther down the path.

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Debby!
Thank you for your contribution!
Sheila

260 Madison Avenue New York, NY 10016
Tel 212 696-8200 Fax 212 696-8299 www.akc.org