

## DNA Test For Fluffies

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They are adorable, round balls of fur, unmistakable with their jackets of soft cottony hair. They are homozygous recessive—fluffies. And that’s how breeders have traditionally found out that their perfectly normal-seeming parents were heterozygous recessive—fluffy gene carriers. One breeder describes her experience this way, “By six weeks of age, the fact that puppies are destined to be fluffy is indisputable. They just puff right out as if someone had pumped them full of air.” It’s a serious fault in our breed. But now there is a new tool to avoid the Michelin puppy scenario.

Donna Housley, a postdoctoral research associate with Dr. Patrick Venta, a geneticist at Michigan State University, has found the key. After the publication of the dog genome sequence in 2005, Dr. Housley was casting about for a project when she came across a shelved study in Dr. Venta’s lab. It had to do with the genetic factors governing hair length in dogs. She was intrigued.

“Many of the hair cycle genes have been identified and studied, just not in dogs.” And that’s how she made an important discovery. In examining the sequences, she found that the gene FGF5 (Fibroblast Growth Factor 5) is responsible for whether a dog has a long (rough or fluffy) coat or a short, smooth coat. “Elimination of FGF5 was known to cause long hair in mice (angoras), so it was a perfect candidate to test for its association with long hair in dogs.” It is a recessive allele, an ancient mutation “that’s probably been around for hundreds of years.” It appears in a number of breeds including Dachshunds, German Shepherds and most notably in Corgis, where her conclusive research was centered.

Last year Dr. Housley expanded the study to include both Norwich and Norfolk and has found the same genetic marker. “All the fluffs and none of the normals had two copies of the longhair mutation (or allele). Although a small number of dogs were genotyped, the results are statistically significant and consistent with the association of this allele with fluffy terriers.” Since Corgis have the same marker, this means that a test is now in place to determine if your Norwich or Norfolk is a carrier.

It’s a convenient non-invasive cheek swab test, sent by regular mail to the lab. And it can be conducted at any age. Along with other genetic testing labs, DDC Veterinary offers the test. According to the DDC website, the “DNA Fluffy Test,” yields a diagnosis of 3 possible genotypes:

N/N Clear (those having 2 copies of the normal short-hair allele (N))

N/F Carrier (those having 1 copy of the normal allele (N) and 1 copy of the long-hair mutation (F))

F/F Affected “Fluffy” (those having 2 copies of the long-hair mutation)

|                   |                | Parent 2 Genotype        |  |                             |
|-------------------|----------------|--------------------------|--|-----------------------------|
|                   |                | Clear (N/N)              | Carrier (N/F)                            | Affected (F/F)              |
| Parent 1 Genotype | Clear (N/N)    | 100% Clear               | 50% Clear<br>50% Carrier                 | 100% Carrier                |
|                   | Carrier (N/F)  | 50% Clear<br>50% Carrier | 25% Clear<br>50% Carrier<br>25% Affected | 50% Carrier<br>50% Affected |
|                   | Affected (F/F) | 100% Carrier             | 50% Carrier<br>50% Affected              | 100% Affected               |

**Legend:** N = Normal Short-Hair Allele F = Long-Hair Mutation

“Fluffy is an autosomal recessive trait; therefore dogs that are carriers of the long hair mutation will appear to be normal but will likely pass on the ... “fluffy” mutation 50% of the time.” Autosomal recessive means that two copies of an abnormal gene must be present in order for the trait (or disease) to develop. Genes come in pairs. Recessive inheritance means BOTH genes in a pair must carry the mutation in order for it to appear. Carriers have just one of the defective genes which they can pass to their offspring.

Now that breeders have a conclusive test for this trait, we can make informed decisions about future matings. However, is it really in the interest of the gene pool to only breed clear to clear? An otherwise terrific dog may be a carrier. This is not an inherited disease, but rather a charming anomaly. So I would caution breeders to test breeding stock, find out if there is a problem and make wise decisions accordingly. Don't throw the baby out with the bathwater however.

NB: Dr.Housley is looking to expand her data base to other breeds. Contact her at [housleyd@cvm.msu.edu](mailto:housleyd@cvm.msu.edu)

— Leandra Little, AKC Gazette Breed Columnist

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