



TOPIC TITLE: Lafora, MPS₁, Thrombopathia & POAG Genetic Testing

BACKGROUND

Genetic tests are available for six mutations associated with disease in Basset Hounds. All are passed through simple recessive inheritance.

The BHCA Health Policy strongly recommends testing for four of them:

1. Thrombopathia,
2. Primary Open Angle Glaucoma (POAG),
3. Mucopolysaccharidosis (MPS₁), and
4. Lafora Disease.

As more dogs are tested, carriers of these mutations can be identified. In general, carrier dogs do not have features of the disease but when bred with another carrier of the same mutation, there is a risk of having affected pups. By breeding to dogs that test clear, the presence of these diseases can be reduced over successive generations and eventually eliminated, without sacrificing genetic diversity and vigor in the breed.

This approach was successful in greatly reducing the incidence of thrombopathia in Basset Hounds. As of June 2022, 4% of 545 dogs tested by Neogen/Paw Print Genetics were carriers, and none was affected. The incidence of POAG is similar: 4% of 514 dogs tested were carriers and one dog was affected.

MPS₁ was discovered in the Basset Hound in 2019. The disease typically results in early puppy mortality. The University of Missouri genetics lab identified and conducts the testing for the disease-causing mutation for MPS₁. Their test can be ordered through the Orthopedic Foundation for Animals (OFA – see below). As of June 2022, 532 dogs have been tested and, of these, 7% were carriers and another 2% were affected.

Lafora Disease has been recognized in Basset Hounds for years, but prior to last year most of us had never heard of it. Signs usually appear between 5-7 years of age and progress to epileptic seizures. Two laboratories in Europe offer tests for it. This past year, several BHCA members had roughly 100 U.S. dogs tested at these labs and a high percentage of dogs were found to be carriers or genetically affected. Some have developed clinical signs.

Neogen/Paw Print Genetics has been working on a test for Lafora for several years. With samples from carriers and affected dogs sent by BHCA members, they were able to complete test development. **Starting October 1, 2022, we will have a validated and accurate cheek swab test in the U.S.!**

The BHCA Health Committee urges members to test all of their dogs for Lafora, not just those they intend to breed. Here's why:.

- 1. Current testing data indicate the mutation is widespread. All breeding pairs should be tested before breeding. Those found with the mutation should only be bred to dogs that are found clear.**
- 2. Anecdotal reports suggest most veterinarians are not aware of Lafora Disease and look elsewhere for the diagnosis in dogs presenting with clinical signs. Informing your veterinarian of test results can save time and money diagnosing signs should they occur. And although the science is not yet clear, there may be progress in the development of treatment options that lengthen the time dogs are free of clinical signs and/or reduce the severity of signs once they appear.**
- 3. There is much we do not yet know about Lafora. The relationship between the mutation and the clinical presentation of the disease is confusing. On the one hand, we know the disease-causing mutation for Lafora has been identified in the breed for some time. Yet very few people report seeing seizures in older dogs. Tests of U.S. dogs done in European labs suggest the mutation is widespread, yet few dogs show clinical signs. Perhaps there is a recent upsurge in the number of dogs with the mutation or perhaps not all dogs that are genetically "affected" actually develop disease.**

For the sake of the breed, we need to know:

- What percentage of Basset Hounds carry the mutation and how many are genetically affected, and**

- Of those with two copies of the mutation, how many go on to develop clinical signs.

The more dogs that are tested and the more owners participate in research studies, the better able we will be to address these questions. The Paw Print Test Order Form has a box to check to indicate your willingness to participate in Lafora research. Please check the box if you are interested. Participant identities will remain confidential and never be released.

On August 25, 2022 BHCA members including the Health Committee and others with a strong interest in Lafora participated in a ZOOM session with Neogen/Paw Print Genetics Directors who developed the test. The session lasted 90 minutes and was recorded. For more information about the science behind Neogen/Paw Print Genetics' Lafora test and what we know and do not know about the disease, check out the video at <https://youtu.be/Pjo8Ed9Cc8k> .

TESTING OPPORTUNITIES

NEOGEN/PAW PRINT GENETICS

Neogen/Paw Print Genetics is giving the BHCA a 50% discount on all their genetic tests from **October 1st to November 30th, 2022**. They offer 5 tests for Basset Hounds - all from the one oral swab sample.

Strongly recommended by the BHCA Health Policy

1. Lafora
2. Thrombopathia
3. Primary Open-Angle Glaucoma

Additional tests offered by Paw Print Genetics

- Chondrodystrophy and Intravertebral Disk Disease Risk (CDDY-IVDD).
- Severe Combined Immunodeficiency Disease, X-Linked (Not currently seen in Basset Hounds).

These tests are available ala carte - you pick only the ones you want. Individual results will be shared **ONLY** with the owner. **Please see the next section (OFA) for MPS1 testing.**

WHAT DOES IT COST?

Per dog testing: 1 test \$40; 2 tests \$75; 3 Tests \$105; 4 tests \$130; all 5 tests \$137.50

In addition to Paw Print's discount, the BHCA Foundation is providing a \$20 subsidy for Lafora testing. If Lafora is one of the tests you order, you can take another \$20 off the cost for each of the dogs you test. There will be a \$6 to \$8 charge for the test kit (and shipping if you order on-line).

Neogen/Paw Print Genetics has a genetic diversity score and puppy predictor test in development and is offering the test for Basset Hounds at no charge to persons ordering other tests during October and November. Similar to a coefficient of inbreeding (COI), the test analyzes the genomes of any individual dog or potential breeding pairs to predict the potential genetic diversity in a litter of puppies. Comparing the Diversity Scores from different potential breeding pairs allows breeders to find a breeding to increase the genetic diversity of the offspring. If you are ordering other tests and would like to participate in test development and receive a complimentary copy of your dog's diversity score, check the box on the order form.

Go to the Paw Print webpage - www.pawprintgenetics.com and follow these steps:

Click on "Find Tests" and select Basset Hound

- 1. Select the tests you wish to run**
- 2. Create a profile for your dog if it is not already registered with Paw Print**
- 3. Create an account if you do not already have one**
- 4. At check-out, enter the discount code BHCAHC22**

If you prefer to order the tests by phone, call 509-483-5950 Monday – Friday 8 am - 5 pm Pacific Time.

These same tests will be offered at the same price at the Nationals:

- Fun Day, Saturday, October 1**
- Field Trials, Saturday and Sunday, October 1-2**
- After Dog Judging and the Health Committee presentation, Wednesday, October 5**
- By appointment, in your room or RV (only for persons with 3 or more dogs). [Call/text Rosemary at 206.687.0891.](tel:2066870891)**

We are expecting a large number of people to want to test at the Nationals. This may translate to long lines and long waits. There are some things you can do to make the process go more smoothly:

- Test at home instead. The cost will be the same. If you are unsure about how to swab, you can come by the swab clinic and observe how it is done.
- We will have order forms at the Nationals, but if possible, print off the order form ([CLICK HERE](#)) and complete your paperwork in advance.
- Have the AKC registration number for each of the dogs you want to test. The microchip # is optional.
- If you do none of these things, still come and test your dog. We'll figure it out.

OFA – MPS1 TESTING

Paw Print Genetics does not offer testing for Mucopolysaccharidosis 1 (MPS1). At this time, the test is available only on-line through the Orthopedic Foundation for Animals (OFA) and can be ordered at <https://ofa.org/mucopolysaccharidosis-i/>

The cost is \$65. It is also an oral swab test. Use the discount code BHCAHC22 for a \$20 discount also funded by the BHCA Foundation, reducing the cost to \$45.00. The discount is available from now until November 30th, 2022.

If you need assistance placing the order, let us know and we'll help.