CEA/CH DNA Testing – Opportunity Awaits

Collie eye anomaly or choroid hypoplasia is a recessively inherited eye disorder that causes abnormal development of the choroid, a layer in the eye beneath the retina. An eye exam detects CEA/CH only in the first few months; after that, developing pigmentation of the retina obscures the choroid.

Clinical Signs in CEA/CH. These vary from minimal visual problem to malformations of the optic nerve or eye (e.g., coloboma), retinal detachment, intraocular bleeding, and subsequent blindness. **Both mild and severe forms are associated with a mutation in the same gene, NHEJ1.** Predicting severity of the disease in an affected puppy is difficult. Luckily there is a gene test available to breeders and the mutation is identical in affected breeds (e.g., Australian Shepherds, Rough and Smooth Collies, Shetland Sheepdogs, Bearded Collies).

Autosomal recessive inheritance. An autosomal recessive disorder means two copies of an abnormal gene must be present for the disease or trait to develop. Progeny outcome is highlighted below.

	Parent A – normal	Parent A – carrier	Parent A - affected
Parent B – normal	All normal	Normal 50% Carrier 50%	Carrier 100%
Parent B – carrier	Normal 50% Carrier 50%	Normal 25% Carrier 50% Affected 25%	Carrier 50% Affected 50%
Parent B – affected	Carrier 100%	Carrier 50% Affected 50%	All affected

The goals of using a pre-breeding genetic test for a recessive disease are to

1. Eliminate disease
2. Gradually reduce frequency of carriers over several generations

Suggestions. One CEA affected Beardie (in the UK and Kennel Club registered) and a number of carriers have been identified with the CEA test. Three carriers have American Bearded Collies as ancestors. The BCCA Health Committee and BeaCon suggest that US breeders consider including a CEA test in their pre-breeding health screening test panel.

CEA/CH Testing Options. After review of the companies that offer CEA testing, it was decided to go with Paw Print Genetics for Bearded Collies in North America. Paw Print is one of 4 companies listed by OFA and it has a sublicense for the CEA test from Optigen. Paw Print tests every sample with two different methods (and a third if the first 2 don't agree) and uses procedures for quality control that match those applied in human genetic testing.

Other labs offer CEA testing, too. If you choose a different lab for reasons of cost or convenience, you should ask:

1. What test is done? For the gene mutation or a marker gene?
2. If the test is for the gene mutation, does the lab have a license or sublicense? That assures that quality control measures are used in testing as the original licensed company's procedures are followed.
3. Does OFA accept results from the lab? This is important if you want the test results displayed on a dog's online OFA record.

CEA/CH DNA Test at Paw Print Genetics.

For a limited time tests will be available to owners of North American Beardies at a cost of only $20! Your benefit – $60 per test due to negotiations and support/subsidies from Paw Print Genetics, BCCA, Beardies of the World Calendar, and BeaCon.

Canadian owners who order from home will receive a suggestion for swab materials that help you save mailing the kit from Paw Print Genetics; you will be responsible for the return mailing cost to Paw Print.

How to Obtain DNA Kits. Up to 50 available at 2017 specialty (early October) and up to 25 available initially at Paw Print Genetics (Starting July 1).

2017 Specialty

- Reserve DNA kit - contact Andrea Hobbe (barkley_puppy@hotmail.com), BCCA Health Committee Co-Chair.
- Obtain DNA kit at specialty, swab cheek, complete Paw Print form (you will need dog's registered name and registration #), bring check for $20 made out to BCCA, give to Andrea for return mailing
- If you are from Canada and will be at the specialty, obtaining the kit there will save you return mailing costs

From Home

- Call Paw Print Genetics. M-F 8-5 Pacific time, 509-483-5950 or toll free (US & Canada) 1-855-202-4889.
- They will set up your online account, provide your log in information, take your payment of $20, and mail a DNA kit
- Upon receipt of kit, do cheek swab following directions, complete paper work, and return promptly in prepaid mailer (US) or complete the customs form if you are mailing from Canada (this is not prepaid)
- If you are in Canada, there are swabs you can purchase over the counter for use to eliminate cost of receiving the kit – ask the Paw Print representative.
    - https://www.pawprintgenetics.com/samples/cheek_swab/#purchasing
    - Proaxa G.U.M. brush

Results. Results are available in your online account about 2 weeks after receipt of the DNA sample. You are notified by email when results are posted to your account. Log in to review the information which you can print. You will need a copy of the Canine Health Certificate for OFA.

OFA. To have the results posted with your dog's OFA online record

- Go to OFA. www.ofa.org, forms (nav menu at very top of screen), application forms, DNA based genetic disease. Print and complete the form, choose payment method, return to OFA with the Paw Print Canine Health Certificate. There is no charge for affected dogs.

Genetic Guidance is available free from Paw Print. They have veterinarians and geneticists on staff to assist with your questions.

Collective result reporting. Paw Print Genetics will provide an anonymous report for the BCCA Health Committee and BeaCon. No dog or owner will be identified in that report.