Papillon Club of America
Health & Genetics

Genetic Research Committee PRA/CHIC/OFFA Report
May 2016

PRA1 DNA Testing :

It has been four years now since Dr. Simon Petersen-Jones, Michigan State University, announced that his research team had identified the gene mutation for PRA1 in Papillons and Phalénes and developed a simple DNA test for people to use to test their breeding stock. As of this report, nearly 3400 Papillons and Phalénes have been DNA tested by eight genetic test labs: OptiGen (USA), Paw Print Genetics (USA), VetGen (USA), Laboratory of Veterinary Genetics (Canada), Laboklin (Germany), Biofocus, (Germany), EVG (Slovenia), Genomia (Czechoslovakia). Of those Papillons and Phalénes tested, about 18% are Carriers and 1% are PRA1 Affected. Over more time and with "smart breeding", we should see an overall decrease in both carriers and affected dogs. ***Eventually, with planned "smart" breeding, the PRA1 genetic mutation will be eliminated from our gene pool while maintaining genetic diversity!***

DNA testing our breeding stock is just the first, and most important step we all need to take. But sharing those results is another key step towards our goal to eradicate PRA1 from our Papillon and Phaléne gene pool! Our goal should be two-fold: 1) know the status of our own breeding stock and use that information wisely going forward and 2) promote responsible breeding for the improved health of our Papillons and Phalénes. First, contact those who are directly involved in the dog's lineage (i.e., owners/breeders of parents, progeny and siblings, if known). This personal contact is also an opportunity to discuss with that person which dogs they should test and what the significance is to their breeding program. There are as many people out there who do not understand as there are those who have a clear picture of the significance of DNA testing and "smart breeding". So we need to help educate each other whenever we have the chance. Secondly, share your dog's PRA1 status publicly. Our PCA Genetics site has both a cumulative listing of PRA1 Results by year (2012, 2013, 2014 and 2015 are posted) and a chart showing worldwide Statistics for PRA1 Test Results - based on data provided by the genetic testing laboratories.

One of the advantages of publicly disclosing the PRA1 status of your Papillon(s)/Phaléne(s) is that you don't have to figure out who all might have related dogs. Breeders can look it up for themselves. Believe me, word spreads like wildfire once the data is posted!! I've talked with several people who have reviewed the PRA1 Results reports on the PCA Genetics site - only to find that they have related dogs. They were then eager and motivated to test their own dogs! No responsible breeder *wants* to produce PRA1 carrier or affected dogs. The wider the dissemination of PRA1 status on our dogs, the sooner we will be able to eradicate PRA1 from our gene pool!

Important points to remember :

- "Don't look back!" Test your current breeding stock and go forward from there with "smart breeding" practices.
- It is not necessary nor is it productive to try to "place the blame" on dogs (or breeders) who might have contributed the genetic mutation. We did not have a DNA test back then, so what's important is what we do going forward.
- Breeders did not *intentionally* breed carriers. Prior to PRA1 DNA testing, they had no way to know whether their dog was a carrier or not.
- "Don't throw the baby out with the bath water." **Carriers *can* be bred!** Breed a Carrier dog to a genetically Normal/Clear dog and test the offspring. Statistically, such a breeding has a 50% chance of producing Normal/Clear dogs.
- *There is good news:* DNA testing assures you that you need *never* produce a PRA1-Affected dog!
- By DNA testing and sharing our results, we can eliminate the PRA1 genetic mutation from our Papillons in just *a few generations!*
-

REPORTS : Genetic Research Committee PRA/CHIC/OFFA Report - May 2016

PRA2 Research :

Dr. Simon Petersen-Jones and his MSU research team are continuing their scientific search for the second type of PRA in Papillons and Phalénes. With the help of Alla Jenkins from Germany, blood samples were collected from a group of closely-related Papillons/Phalénes in Europe. All of these dogs were DNA tested Clear/Normal of PRA1, but some of these dogs are *PRA2 Affected*. Using DNA samples from these dogs that have tested PRA1 Clear/Normal makes it easier for researchers to focus on the *second* PRA genetic mutation (PRA2). The DNA from this "family" of dogs will provide valuable data for our MSU researchers. MSU has obtained additional blood samples from OptiGen and other collaborative sources, which increased the statistical sample. However, researchers have not been able to locate the genetic mutation. More blood samples from PRA2 Affected dogs and their relatives are needed to further their
research.

Eye Exams by Veterinary Ophthalmologist :

It is still critically important that you continue to have your dogs' eyes examined, on a regular basis, by a veterinary ophthalmologist. Other eye diseases besides PRA still exist for Papillons and Phalénes (e.g., glaucoma and cataracts). We are seeing an increase in reported cases of Juvenile Cataracts, which show up at a very early age. In
the past, many breeders haven't done CERF or OFFA exams on their Papillons and Phalénes until a year or two of age - namely, prior to breeding. However, it is now become apparent that we must have our dogs' eyes checked *at a much younger age!* Elyse Vandermolen is our Genetic Research Committee member who is spearheading the Juvenile Cataract research project through MSU.

CHIC Papillons/OFFA Reports :

Each year, on a quarterly basis, Orthopedic Foundation for Animals (OFFA) provides PCA with a listing of all new and updated CHIC Papillons. This listing is added to our cumulative CHIC Papillon list on our Genetics website (www.pcagenetics.com) – 843 total CHIC Papillons as of 1st quarter 2016. The listing is linked to the main OFFA online
database, so you can click on the dog's registration number to see that dog's complete OFFA health information.

In early April, I received the attached announcement from Eddie Dziuk about OFFA's new Advanced Cardiac Database. This new program will require that cardiac exams be done by a board certified veterinary cardiologist. It looks like this new program will be very similar to the CAER (formerly CERF) eye exams. Here is an excerpt from that announcement:

"There will be a transition period (length to be determined) where the current OFA Congenital Cardiac Database will continue to be available with no changes to the forms, submission procedures or exam protocols. However, effective April 1, 2016, we will be offering the Advanced Cardiac Database (ACA), which will become the new standard for cardiac exams and will eventually replace the existing Congenital Cardiac Database.
Please note, exams for the new Advanced Cardiac Database are limited to boarded veterinary cardiologists with Diplomate status in either the ACVIM (American College of Veterinary Internal Medicine/Cardiology subspecialty) or the ECVIM (European College of Veterinary Internal Medicine/Cardiology subspecialty). General practitioners and other specialists may not perform Advanced Cardiac Database exams."

This requirement will have an effect on folks who are used to getting their OFFA cardiac exams done by their regular veterinarian - including additional expense for cardiac testing. PCA Genetic Research Committee is concerned about this new Advanced Cardiac Database program and is consulting with our PCA Board for further guidance.

OFFA also sends PCA a quarterly report of *all* papillon tests that have been done and submitted to OFFA for publication. To date, these tests include hips, elbows, cardiac, patella, thyroid, PRA1, neuroaxonal dystrophy (NAD), spinocerebellar ataxia, Legg-Calve-Perthes, primary lens luxation, CERF/OFA eye registry, and dentition databases.

The online OFFA database (http://www.offa.org) is another valuable tool for breeders to use to research dogs' health information when contemplating possible breedings.

Submitted by:
Stephanie Koeniger

PRA/CHIC/OFFA Liaison - PCA Genetic Research Committee