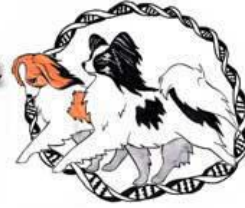


# *Papillon Club of America* *Health & Genetics*



## ” As Far as the Eyes Can See “

I have learned so much during this last year being Chair of PCA Genetic Research Committee (GRC) and I find myself wanting to share this information for all Papillon Breeders.

When it comes to eyes, clearly there are different situations that arise and initially cause alarm until further examination. Being a nurse for the last 42 years has helped me take better care of our dogs. There is still something new to learn everyday both in the human and canine world.

This article is coauthored with Dr. Simon Petersen-Jones in hope that we can educate and provide some knowledge to help with breeding when it comes to eye issues. At the end of this article there will be a link to the current statistical data published from OFA on their eye exam results of 2014.

There will be information regarding the two genetic research projects that the PCA Genetic Research Committee is working in collaboration with Dr. Simon Petersen-Jones, PRA type 2 and Juvenile Cataracts. Both of these diseases are considered recessive traits meaning both parents must carry the gene for any of their offspring to have the condition. Those that carry only one copy of the defective gene (known as carriers) do not develop the condition, but those with two copies of the defective gene do.

I think first we need to state the suggested recommendations of eye exams and why they should be done on a regular basis and the reasoning why Dr. Simon Petersen-Jones feels these ages are the best to have your dogs examined throughout their life.

The first eye exam is based on what you expect to find in your breed. Some have developmental eye problems that can be diagnosed at 6-7 weeks of age. People often have the entire litter screened for these. For Papillons, it has been suggested that the first routine eye exam could be performed at 6-9 months of age. Most eye testing should start somewhere around 12-14 months of age. Each breeding dog should be checked prior to breeding and an annual re-examination is recommended while they are used for breeding. After that time less frequent examinations should be considered to catch later onset diseases.

If an eye exam is done between 8-12 weeks of age, your puppy may be diagnosed with a temporary form of cataracts. These temporary cataracts are opacities at the tips of the Y suture lines within the lens. As any opacity in the lens is by definition a cataract they will be diagnosed as having cataracts. The interpretation and significance of cataracts at that age is important. The suture tip cataracts are temporary, seen as the lens develops and will disappear as the puppy matures. However, other forms of cataracts may also be present at that age. I personally wait until after 12 months of age, so that I can do all the CHIC certified requirements for eyes, heart and patellas. It is at that time I have our dogs microchipped. If there are any health concerns at that time you can

make an educated decision whether or not to keep this dog in your breeding program. It is still important to get regular OFA eye examinations performed on your breeding dogs, even if they have been cleared by a DNA test PRA1, as there is more than one type of PRA in Papillons. Health clearances are only one of many factors for consideration in choosing your breeding dogs.

Papillons are relatively healthy dogs with regards to eye diseases. That being said in the last year and a half I have been called to help with some very unusual occurrences: PRA diagnosed in one eye, unusual cataract formation not determined juvenile cataracts, and corneal dystrophy-stromal. I would like to address PRA first.

Progressive Retinal Atrophy (PRA) is a group of genetic diseases seen in dogs, including our breed and is recessively inherited in all breeds of dogs studied, with the following exceptions: PRA is dominantly inherited in Old English Mastiffs and Bullmastiffs, and is sex-linked and found primarily in male dogs in Siberian Husky and Samoyed breeds. PRA is an inherited disease of the retina that causes the photoreceptor cells in the retina to die. PRA occurs in both eyes simultaneously and is nonpainful. Because PRA typically causes death of the rod photoreceptors, and rods are responsible for vision in dim light, or “night vision”, the first clinical signs that the dog owner may notice is night blindness. Owners may also notice increased “eye shine” or dilated pupils. PRA is similar to retinitis pigmentosa in humans and is characterized by the bilateral degeneration of the retina, causing progressive vision loss culminating in blindness.

We have been so very blessed to have Dr. Simon Petersen-Jones work on finding the gene mutation that causes PRA in Papillons. During his research in 2012, Dr. Simon Petersen-Jones and his team discovered a gene mutation for the form of PRA Type 1 and developed a DNA-based test to detect this form of PRA. DNA tests for PRA 1 are now available worldwide at several labs. Dr. Simon Petersen-Jones wants us to be aware that there is more than one type of PRA in Papillons. Research is continuing to isolate all additional gene mutations that cause PRA in Papillons. Currently he needs dogs that have been diagnosed as affected with PRA and have tested clear for the DNA test for PRA 1.

The PRA diagnosed in one eye, in a Papillon as occurred last year is very unlikely to be true PRA. There are rare genetic anomalies that could be present in a dog that could result in the PRA causing gene defect being present in both copies of the gene in one eye and not in the other (for example a condition known as mosaicism). Other potential causes for retinal degeneration that could mimic PRA include inflammatory change in the eye, parasites and infection.

Corneal dystrophy is a non inflammatory corneal opacity (white to gray). The commonest form presents as a central round to oval shaped opacity that is present in the layer of the cornea known as the stroma. The term dystrophy implies an inherited condition. In several breeds corneal dystrophy has been suggested to be an autosomal recessive trait. In some breeds inheritance appears sex-linked. In Papillons it has been implied to be an inherited condition. It is usually bilateral although not necessarily symmetrical and the onset in one eye may precede the other.

You may notice this white to grey opacity in one or both of your dog’s eyes. Other forms of corneal dystrophy include endothelia dystrophy. This affects the function of the corneal endothelial cells that line the inside of the cornea and act to pump fluid out of the cornea. When these cells die the result is a build-up of fluid in the cornea (corneal edema) which clouds the normally transparent cornea and may decrease vision. Edema may cause the eye to appear blue. Recurring non-healing shallow corneal ulcers occur as well. It has been suggested that a form of recurrent

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superficial corneal ulceration that occurs in middle-aged dogs and is commoner in certain breeds may also be another form of corneal dystrophy.

Affected dogs diagnosed with stromal dystrophy should not be used for breeding. Affected dogs and their close relatives diagnosed with endothelial dystrophy should not be used for breeding.

I have had a report of one puppy born with only one eye. The other eyeball was present but very small and abnormal. This is a form of microphthalmia. Microphthalmia is a genetic disorder of the eyes in many breeds of dogs.

Persistent pupillary membranes (PPM) are persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by three months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. Iris to iris is the least severe form and if it only involves a few strands of remaining tissue it is considered safe to breed a dog diagnosed with this disorder. A prudent breeding approach is to breed a dog with a clear eye examination to a dog diagnosed with PPM.

The rest of this article is going to talk about Cataracts. Cataracts are one of the most common problems affecting the eyes of dogs. There are many different forms and causes that contribute to the formation of cataracts. They affect all breeds and ages of dogs, and Juvenile Cataracts have been disclosed in our breed since 1997.

Cataract is defined as any opacity of the lens and/or its capsule, showing clinical features seen from pinhead marks to total lens opacity. There are several forms and causes of cataracts, and they all develop in a similar fashion. Cataracts occur when there is a disruption of the normal arrangements of the lens fibers or its capsule. When this disruption occurs there is a loss of transparency and if extensive enough a reduction in vision.

Cataracts can be caused by trauma to the eye or metabolic disorders, like diabetes mellitus. In diabetic dogs, the glucose concentration in the lens increases. Cataracts in diabetic dogs can develop extremely rapidly, particularly if the dog's glucose levels are not regulated. It generally affects both eyes. Puppies being hand raised by bottle feeding can develop cataracts due to nutritional deficiencies from inappropriate milk supplement.

The age at which a dog develops cataracts is very important in determining the type of cataract. It is especially important for determining if the cataracts are the result of a heredity trait within our breed.

Congenital cataracts are cataracts that are present at birth. These cataracts usually occur in both eyes. Despite the fact that they are born with them, they are not necessarily inherited. Infection, toxins, or nutritional deficiencies during pregnancy can contribute to congenital cataracts. In some dog breeds, congenital cataracts are inherited. In Papillons, Juvenile cataracts are thought to be inherited as a recessive trait, meaning both parents (if unaffected by cataract themselves) must carry the one copy of the gene defect for their offspring to develop the condition.

Juvenile cataracts may occur in a puppy under one year of age, but may also occur in dogs up to five years of age. That is important to note. There has been one puppy recently reported to me

that was diagnosed at 8-9 months of age, and a Papillon diagnosed at 3 years of age, and the prior CERF eye exams were normal.

Currently the PCA Genetic Research Committee is working with Dr. Simon Petersen-Jones with collection of blood samples of affected Papillons diagnosed with Juvenile Cataracts and blood from the first and second generations of dogs. We need your help to meet the goal of collecting 20 affected blood samples out of 100 total samples of related dogs blood. It all comes down to statistics when it comes to finding a mutation gene. Please go to our website at [www.pcagenetics.com](http://www.pcagenetics.com) and read about how to help. Information including submittal form, and collections and shipping instructions are now available online. You can remain anonymous and send blood directly to Dr. Simon Petersen-Jones with the current eye examination results and pedigree of the affected Papillon. He will keep all information confidential. It is very important that the diagnosis of the affected dogs is accurate. The success of the study depends on the accuracy of the diagnosis and also that there is only one form of Juvenile Cataract in the breed. If there are more than one genetic forms of Juvenile Cataract occurring in the breed then it makes it much harder to find the gene mutations responsible.

With the ongoing support of the Papillon Club of America and breeders and owners around the world, the MSU Research Team has continued their work to locate the genetic mutation that causes the second type of PRA in Papillons and now in the genetic mutation that causes Juvenile Cataracts. This research has been possible only with the cooperation and support of papillon/phalene breeders and owners who, with the help of their veterinary professionals, have submitted DNA blood samples from their dogs to our MSU Research Team. We still need more blood.

As breeders we work hard to find the perfect companion homes, see that our puppies are socialized, and raised with love. The control of inherited disease is our responsibility as a breeder. DNA based tests play an increasingly significant role to allow breeding dogs that are carriers to non carriers (clear), and allow breeders to preserve the breed gene pool for future generations. With DNA based tests, we will eventually eliminate the disease while preserving those desirable attributes that make up our Papillons.

For further information please go to our website [www.pcagenetics.com](http://www.pcagenetics.com) and check out the new information on Juvenile Cataract Research. The instructions for blood collection for the PRA 2 and Juvenile Cataract research projects are the same. You will be able to download the forms and work directly with Dr. Simon Petersen-Jones.

Here is the link for OFA at [www.offa.org](http://www.offa.org). For further information you can go to ocular disorders presumed to be inherited in purebred dogs and check out the Bluebook 2014 Seventh Edition for each breed.

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