# PPPA Health and Research Committee Report to the Club April 1, 2017

We have had a very busy last 8 months with the discovery of several Genetic Markers in the breed. This was an unexpected benefit of the using the Optimal Selection test panel and will ultimately help us keep the Portuguese Podengo Pequeno the healthy breed it is.

# **UPDATE ON DNA TESTING:**

We currently have identified 3 Genetic Tests that are applicable to the Portuguese Podengo Pequenos. There are two forms of PRA, crd4 (formally cord-1) and prcd. The third Genetic test is for DM (SOD1).

Most of the testing has occurred overseas in Sweden, Finland, Norway, Germany, and Portugal. It is impossible at the moment to tell exact numbers as there are many results not yet known but I will list what I have currently in my database as of Monday, March 20, 2017.

There are only three terms that can be used to identify the results of the DNA test. The first is "AFFECTED" and means there are two copies of the mutated genes and the individual dog will likely develop the condition at some point in their life. The second result is "CARRIER" which means the dog has one (1) copy of the mutated gene and may pass it to offspring if bred. A "CARRIER" will not develop the condition. Lastly, the result we always like to see is "NORMAL" or "CLEAR" which means the dog has no copy of the mutated gene to pass along and of course will not develop the disease. In the last case, I will refer to this as "NORMAL" in this report .

#### crd4 PRA

There have only been four (4) "CARRIERS" found and no (0) "AFFECTED" found. The first discovered PRA was crd4 reported on July 21, 2016 in the USA from an Optimal Selection panel. The mother was also found to be a "CARRIER" so there are two in the USA. The grandfather in Portugal (now deceased) was also a "CARRIER". There were two sons of the Portuguese father were tested in Sweden with one being a "CARRIER" and the other was "NORMAL". There have been many other non-related dogs tested here in the USA with Optimal Selection" as well as standalone testing in the overseas countries mentioned above and all known results are "NORMAL". All of these dogs are smooth; however, the grandfather did have one wire line in his pedigree.

### prcd PRA

The first evidence of "AFFECTED" dogs was reported on November 17, 2016 when two diagnosed dogs were tested in Finland. In addition to these two, there were two other siblings from the same litter of one also diagnosed with PRA. There have been over 48 other dogs tested now with NO additional "AFFECTED" dogs and fourteen (14) dogs found to be "CARRIERS". All of the reported "CARRIERS" are wires. The smooths that have been tested, including one in the USA, have all been "NORMAL". It needs to be noted that many of the dogs tested under Optimal Selection here in the USA have had DNA samples set to Paw Print Genetics for testing for prcd PRA.

#### DM

The first "CARRIERS" of DM were reported on October 30, 2016 and were all related. There had been one additional reported "CARRIER" in the initial study of **DM (SOD1)** published in 2014. So far, there have been only "NORMAL" results on all of the Optimal Selection test as well as 9 independent tests in the overseas countries above. All of the "CARRIERS" have been wires.

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# Where do we go from here?

It is now very clear that the **prcd PRA** is the major concern when it comes to PRA in the breed; however, we need to proceed with caution so we don't ignore **crd4 PRA** or **DM** for low volume occurance. Those who tested with Optimal Selection will only have to run the **pcrd PRA** test. If you have not run the Optimal Selection panel they should run all three, especially on your breeding partners. Paw Print Genetics has developed a special discount program which is outlined below.

We need to make sure that everyone understands that this does not eliminate the need for yearly eye test as there are many other conditions that occur. See the Attachment "A" from the ACVO Blue Book.

You can see the entire 2015 Blue Book at:

http://www.acvo.org/new/diplomates/resources/Blue%20Book%202015%208th%20Edition%20CORRECTED.pdf

## Here is the discount program Paw Print Genetics has created for PPPs for 2017

Discount code: PPPA17

25% off 1-2 disease tests in a dog 50% off 3 disease tests in a dog

60% off 4 or more tests in a dog that include PRA-prcd, PRA-crd4 and DM, and then one or more coat colors and traits such as furnishings

### **Examples of how this will help with testing for the above referenced DNA?**

Any single test: List price \$80. 25% off member's discount. **Final Price \$60** PRA-prcd

3 disease tests: List price \$210. 50% off member's discount. **Final price \$105** PRA-prcd DM PRA-crd4/cord1

3 disease tests plus 1 or more color or trait tests in the same dog: 60% off member's discount. For example, the 3 disease tests, plus 5 colors and traits, list price is \$340. **60% off will be \$136**.

This price includes the cheek swab kit, shipping, prepaid mailer for returning the samples to the laboratory, free access to posting on Paw Print Pedigrees and our concierge level of service.

Note: The above discount code can be used by anyone who owns a Portuguese Podengo Pequeno whether they are members of the club or not. Paw Print Genetics will be providing the PPPA Health and Research Committee periodic Statistical Data on the results which will not include specific dog information.

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A full list of all disorders and traits available for testing can be found at the Paw Print Genetics website:

https://www.pawprintgenetics.com/

# **Health Disorder Reporting**

Many AKC Parent Clubs have developed forms for reporting diagnosed conditions that are not part of the known disorders or diagnosed conditions which do not have a reliable reporting path ie. ACVO, OFA, etc.

We are in the process of developing the **PPPA Report of Health Disorder** form with instructions for providing details of diagnosed Health Issues. When the form is released you will be able to submit it to the committee so we can track any new conditions.

If you have any questions please let me know.

Respectively Submitted,

Robert "Bud" Hidlay Chairman, PPPA Health and Research Committee budhidlay@comcast.net

Attachment "A": Blue Book 2015 Pages 783-785

## **OCULAR DISORDERS REPORT**

PORTUGUESE PODENGO PEQUENO - 1

# PORTUGUESE PODENGO PEQUENO

	DISORDER	INHERITANCE	REFERENCE	BREEDING ADVICE
A.	Distichiasis	Not defined	1	Breeder option
B.	Persistent pupillary membranes - iris to iris - all other forms	Not defined Not defined	2 2	Breeder option NO
C.	Vitreous degeneration	Not defined	1	Breeder option

# **Description and Comments**

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 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. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## C. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

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## **OCULAR DISORDERS REPORT**

PORTUGUESE PODENGO PEQUENO - 2

### References

There are no references providing detailed descriptions of hereditary ocular conditions of the Portuguese Podengo Pequeno breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

- 1. ACVO Genetics Committee, 2015 and Data from OFA All-Breeds Report, 2014-2015.
- 2. ACVO Genetics Committee, 2013-2014 and Data from OFA All-Breeds Report, 2013-2014.

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# OCULAR DISORDERS REPORT PORTUGUESE PODENGO PEQUENO

TOTAL DOGS EXAMINED	1	1991-1999 0		2000-2009		2010-2014 66		2015 99	
Diagnostic Name	#	%	#	%	#	%	#	%	
EYELIDS									
20.140 ectopic cilia	0		0		1	1.5%	0		
25.110 distichiasis	0		0		4	6.1%	5	5.1%	
UVEA									
93.710 persistent pupillary membranes, iris to iris	0		0		5	7.6%	5	5.1%	
93.730 persistent pupillary membranes, iris to cornea	0		0		0		1	1.0%	
LENS									
100.210 cataract, significance unknown	0		0		3	4.5%	4	4.0%	
100.301 punctate cataract, anterior cortex	0		0		1	1.5%	1	1.0%	
100.311 incipient cataract, anterior cortex	0		0		0		1	1.0%	
100.312 incipient cataract, posterior cortex	0		0		1	1.5%	1	1.0%	
100.317 incipient cataract, capsular	0		0		1	1.5%	0		
100.330 generalized/complete cataract	0		0		0		1	1.0%	
100.340 resorbing/hypermature cataract	0		0		0		1	1.0%	
100.375 subluxation/luxation, unspecified	0		0		0		2	2.0%	
VITREOUS									
110.120 persistent hyaloid artery/remnant	0		0		0		1	1.0%	
110.200 vitritis	0		0		1	1.5%	1	1.0%	
110.320 vitreous degeneration syneresis	0		0		2	3.0%	6	6.1%	
110.330 vitreous degeneration anterior chamber	0		0		1	1.5%	0		
RETINA									
120.170 retinal dysplasia, folds	0		0		1	1.5%	0		
120.310 generalized progressive retinal atrophy (PRA)	0		0		1	1.5%	1	1.0%	
120.960 retinopathy	0		0		3	4.5%	0		
OTHER									
900.000 other, unspecified	0		1		0		0		
900.100 other, not inherited	0		0		1	1.5%	3	3.0%	
NORMAL									
0.000 normal globe	0		14		83	125.8%	81	81.8%	