

PPPA Health and Research Committee Report to the Club December 26, 2018

MyBreedData & Paw Print Genetics Updates

MyBreedData:

The appearance of both prcd/PRA and DM in the Smooths as reported in our June report, has caused an increase in testing especially in the Nordic countries. The overall percentage of "Carriers" is about 6% for prcd/PRA and 5.5% for DM. The new "Carrier" of crd4/PRA outside of the initial family where it was first found has caused great concern and increased testing of Smooths.

The Pequeno Wirehaired numbers are still basically the same as was reported in our last Report to the Board, 15% for prcd/PRA "Carriers" with 2.74% "At Risk" and 11.6% for DM "Carriers".

The MyBreedData is a statistical report from tests run by Optimal Selection and Genoscooper so unless results are made public we do not know what dogs are involved. This also means that none of the testing being done at PPG, VHL (Netherlands) or other US labs is included. Here are links to the MyBreedData information:

Smooth: (**Appendix "A"**)

http://www.mybreeddata.com/crm/index.html#breed/519248a83cd390a0520000d4/portuguese_podengosmoothhairedminiature/

Wire: (**Appendix "B"**)

http://www.mybreeddata.com/crm/index.html#breed/519248a83cd390a0520000d1/portuguese_podengowirehairedminiature/

Paw Print Genetics

Appendix "C" shows the results in testing conducted at Paw Print Genetics from 2017 to 2018. The percentage of DM has fallen with no new "Carriers" found here in the US while the percentage of "Carriers" has remained the same at 19%. Because we do not separate the Smooths from the Wirehaired, they appear to be running close to the European percentages

This reinforces the need to test ALL breeding partners for our previously defined panel at the very least!

Free DNA Test Results Listing

We are planning to update our Free Listing on the first of January so please, if you have tested your dogs for the three known mutations, we would encourage you to make those results public by listing them on the Free DNA Test Results List on the Health Page of the PPPA website. Currently there are 59 dogs listed. Remember you can now submit results from any lab. If you have any questions please let me know. Here is a link to the listing:

<http://nebula.wsimg.com/afb572289de5a16e83f158c7e6294fd7?AccessKeyId=36548DCF9EEE779CB56B&disposition=0&alloworigin=1>

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PLL Update

There still has been no formal results published for the testing in the UK where the first two cases of PLL (Primary Lens Luxation) mutation ADAMTS17 were certified by AHT (Animal Health Trust). Discussions I have had with breeder friends in the Northern Portuguese Podengo Association Breed Club, confirm they have heard nothing official yet; however, they have been told by other breeders that most of the dogs are clear. When I get an official response, I will post an update.

This is a Buyer Beware, if you are looking to import a breeding partner from the UK or any other country, you need to make sure they are tested at least for the three mutations we know about. Several are already testing and posting results.

OFA Update

The third quarter OFA Reports have been posted on the PPPA Website and are accessible from the Health Page. Here is a direct link to these:

<http://nebula.wsimg.com/e6969ecc4c9cddae6ffdfdd9ca55b2d9?AccessKeyId=36548DCF9EEE779CB56B&disposition=0&alloworigin=1>

The fourth quarter Reports should be available in January as well as the 2018 Final Report and will be published then.

Respectively Submitted,

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Appendix "A"

Portuguese Podengo - Smooth-haired Miniature

30-100 individuals tested

KNOWN DISORDERS IN THE BREED

The disorders listed below have been scientifically established in the selected breed. Genetic research continuously identifies new mutations underlying inherited disorders. Most of the currently known mutations can still be considered breed-specific, while others are more ancient and widespread across breeds.

Type	Disorder
Neurological Disorders	Degenerative Myelopathy, (DM)

Degenerative myelopathy (DM) is an inherited neurological disorder found in many dog breeds but most commonly seen in German Shepherd Dogs and Pembroke Welsh Corgis. It is not yet clear if all dogs carrying two copies of the mutation will develop clinical signs, especially considering the variable presentation noted amongst breeds found to carry it. Clinical signs develop at an old age and all affected dogs do not develop symptoms during their natural lifespan. It is unclear, whether the disease process is ongoing in these dogs or if the mutation does not cause disease in all affected dogs. DM is inherited in an autosomal recessive fashion.

See pdf documentation (/crm/disorders/010_SOD1/en/010_SOD1.pdf)

MODE OF INHERITANCE	SEVERITY	CARRIERS	GENETICALLY AT RISK
Autosomal Recessive (Incomplete Penetrance)	Considerable	5.56%	< 1%

Ocular Disorders	OptiGen® Progressive Rod Cone Degeneration, (prcd-PRA)
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Progressive retinal atrophy is the common name for a family of hereditary retinal dystrophies commonly leading to blindness. PRA is found in more than 100 dog breeds but the genetic background varies among breeds and populations. prcd-PRA is a very common form of PRA. This disease is caused by the degeneration of photoreceptor cells of the retina. The disorder is inherited in an autosomal recessive manner.

See pdf documentation (/crm/disorders/033_PRCd_new/en/033_PRCd_new.pdf)

MODE OF INHERITANCE	SEVERITY	CARRIERS	GENETICALLY AT RISK
Autosomal Recessive	Considerable	5.71%	< 1%

Appendix "B"

Portuguese Podengo - Wirehaired Miniature

30-100 individuals tested

KNOWN DISORDERS IN THE BREED

The disorders listed below have been scientifically established in the selected breed. Genetic research continuously identifies new mutations underlying inherited disorders. Most of the currently known mutations can still be considered breed-specific, while others are more ancient and widespread across breeds.

Type

Disorder

Neurological Disorders

Degenerative Myelopathy, (DM)

Degenerative myelopathy (DM) is an inherited neurological disorder found in many dog breeds but most commonly seen in German Shepherd Dogs and Pembroke Welsh Corgis. It is not yet clear if all dogs carrying two copies of the mutation will develop clinical signs, especially considering the variable presentation noted amongst breeds found to carry it. Clinical signs develop at an old age and all affected dogs do not develop symptoms during their natural lifespan. It is unclear, whether the disease process is ongoing in these dogs or if the mutation does not cause disease in all affected dogs. DM is inherited in an autosomal recessive fashion.

See pdf documentation (/crm/disorders/010_SOD1/en/010_SOD1.pdf)

MODE OF INHERITANCE

Autosomal Recessive (Incomplete Penetrance)

SEVERITY

Considerable

CARRIERS

11.54%

GENETICALLY AT RISK

< 1%

Ocular Disorders

OptiGen® Progressive Rod Cone Degeneration, (prcd-PRA)

Progressive retinal atrophy is the common name for a family of hereditary retinal dystrophies commonly leading to blindness. PRA is found in more than 100 dog breeds but the genetic background varies among breeds and populations. prcd-PRA is a very common form of PRA. This disease is caused by the degeneration of photoreceptor cells of the retina. The disorder is inherited in an autosomal recessive manner.

See pdf documentation (/crm/disorders/033_PRCd_new/en/033_PRCd_new.pdf)

MODE OF INHERITANCE

Autosomal Recessive

SEVERITY

Considerable

CARRIERS

14.86%

GENETICALLY AT RISK

2.70%

Appendix "C"

Bud Hidlay

From: Lisa Shaffer
Sent: Thursday, December 13, 2018 10:23 AM
To: budhidlay@comcast.net
Subject: Current Stats for your breed

	2018	2017	
DM			
33 dogs tested		18	
32 normal/clear		17	
1 carrier	3%	1	5%
0 at risk		0	
PRA-crd4/cord1		19	
35 dogs tested		19	
35 normal/clear	0%	0	0%
0 carrier		0	
0 at risk		0	
PRA-prcd			
63 dog tested		37	
51 clear/normal	19%	30	19%
12 carrier		7	
0 at risk		0	

Thanks!
Lisa

Note: Numbers are cumulative.

Lisa G Shaffer, PhD, FACMG
Founder & CEO, Paw Print Genetics



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Facebook: <https://www.facebook.com/PawPrintGenetics>

Note Received via Messenger from Lisa :

"There are still a lot of people that ordered testing but have not sent it in to us."

12/13/2018

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