GENETIC TESTING FOR PLN-ASSOCIATED VARIANT GENES

USING THE PENNGEN SITE FOR SUBMITTING SAMPLES

October 2015
Introduction

- After years of research supported by hundreds of Wheatens, their owners and breeders, Drs. Meryl Littman and Paula Henthorn at Penn Vet have identified mutations associated with PLN in two genes.
- As a result, there is now a test using a non-invasive cheek swab (also referred to as a “cheek brush” or a “cytology brush”), which an owner can use and submit to the University of Pennsylvania School of Veterinary Medicine for interpretation.
- All specimens submitted will support ongoing research into PLN as well as PLE.
- This guide will walk you through submitting information on the PennGen site.

For How To guides on swabbing, testing litters or frozen semen testing, visit the SCWTCA website at http://www.scwtca.org/health/dnatest.htm
Using the PennGen site

- “Paperwork” is done through the “PennGen” website at http://research.vet.upenn.edu/penngen

- Here’s the process
  - First, create an account for yourself. Once you’ve done this, you can go to your account in the future and add dogs.
  - Then, add information on the dog(s) you are going to submit.
  - Print out the submission form and send it with samples to Penn.
  - When the results are ready, you’ll get an email with a link to the site.
  - Log in, pay for the test and then your results will be immediately available.

- **NOTE**: Before starting, be sure you have swabs in hand and/or the appointment with your veterinarian for blood samples or obtaining frozen semen
Create your PennGen account

On the PennGen site, select Create an Account... if you have an account, log in and skip to page 6 to see how to add a dog.

On the next screen, select Animal Owner.
Create your PennGen Account

The screen expands so you can fill in your information...be sure to keep your Account Name and Password!!

1. You’ll get an email confirming you created your account.

2. You’ll see your account screen and you can add a dog by clicking at the bottom..

Click “Create Account” and two things will happen.
Create an Animal Record

Fill in all the information on the dog you’re testing...be sure to have his/her registration information available as well as that of his sire/dam.

Note that since PennGen also offers tests for cats, you’ll see references to “Tom” and “Queen” in addition to “Sire” and “Dam”.

When you click on Species, the Breed box will expand and when you click on “Yes” for “Do you know the exact date of birth” an option to type in the date or use a calendar appears.
Create an Animal Record

When you have completed filling in the information, scroll to the bottom.

You’ll see you have two options.

1. You can just create this dog’s record.

2. If you will be testing littermates, you can click on “Create Animal Record and Create more from the same litter.” This will give you the opportunity to add littermates without having to retype some of this info.

If you are testing a litter, be sure to see our Testing Litters Guide on the SCWTCA Website.
Edit an Animal Record

The dog is created but OOOOPS! We made a mistake...Nola isn’t spayed! Click the “Edit” link to fix it.

Correct the information and click the Update button.
Now it’s time to create a submission form but first, make sure you’ll have swabs on hand and know when you’ll be swabbing your Wheaten or if you are submitting blood or semen, when that will happen. You’ll be asked that question in the next steps.

Click “Create A Submission” to start.
Click the drop down arrow next to Test Type for a list of PennGen offered tests.

Select “PLN Associated Variant Genes”

Note: the $125 price will be used at this stage even if you are submitting a litter. Before charging you, PennGen will adjust for the litter discount. If you are testing a litter, be sure to see our Testing Litters Guide on the SCWTCA Website.
Create a Submission

Answer the questions and note the intended Collection and Shipping Dates. There is a “Comments” box for additional information, such as health status or littermate information.

When ready, select Create Test Request at the bottom of the page.
Completing your Submission

- At this point, you have everything ready. If you want to complete the submission now and print the form, you can. *Once you Complete the submission, you cannot change any information.*
- If you want to wait for later or if you think you may need to correct information, you can do that instead.
- Check the right option and proceed:

  ![Test Request Created](image)

  **Test Request Created**

  *You have successfully created a submission.*

  The submission has a status of NEW. In order for the submission to be recognized by the PennGen staff, you must mark it as COMPLETE. You should then print the submission form and send it in with your sample(s). Once you mark the submission as complete, you will not be able to add any more samples or test requests to this submission (you can always create additional submissions later).

  **What would you like to do next?**
  - [ ] Mark the submission as Complete and proceed to the sending instructions.
  - [ ] Add another test request to this submission.
  - [ ] Go to the submission details without completing this submission.
  - [ ] Go to the animal owner/agent details without completing this submission.

  [Continue]
Completing Your Submission

Your submission is now complete and PennGen will be waiting for your samples.

Print the form, sign it and enclose it and the other information with your samples. Send them to the address on the top of the form.

DO NOT SEND PAYMENT NOW!
Status of your Submission

- You’ll receive an email from PennGen when they receive your samples.
  
  ![Email Example]

- You can also see the information when you log in.
  - If you see “Received”, expect 1-4 weeks for results.
  - If you see “Waiting to be Received”, you can follow up with the delivery service to find out what happened.
Getting Results

- When the results are complete, you’ll receive an email with a link.

   ![Email 1]

   Dear Pam Mandeville,

   A PennGen Laboratories test request has been recently completed. Your account has a balance and is awaiting payment. You are required to pay before you can view the test results. Use the link below to pay your balance. Thank you.

   ![Link]

   Regards,
   PennGen Staff

- Log in and provide your credit card information When your credit card clears, within minutes you’ll receive an email with a copy of the report attached.

   ![Email 2]

   Dear Pam Mandeville,

   Attached is an official report of the test results for the sample you submitted. These results can also be viewed online at ![Link]

   Thank you.

   Regards,
   PennGen Staff
Note that the Genotype will be reported as follows:

- **1-1** – Homozygous Normal/Negative – a dog carrying **NO** copies of the variant alleles
- **1-2** – Heterozygous – a dog carrying **ONE** copy of the variant alleles
- **2-2** – Homozygous Positive – a dog carrying **TWO** copies of the variant alleles

You’ll always be able to see your results online in your dog’s record.
For more information

Check the SCWTCA website for:

- Audio of Dr. Meryl Littman’s presentation on May 5, 2012
- “How To” Guides
- FAQs for owners and breeders
- Research abstract presented at Tufts in 2011
- Contact information for more assistance
- ...and watch for more resources

http://www.scwtca.org/health/dnatest.htm