

## Laboratory Report

<b>Laboratory #:</b>	294946	<b>Call Name:</b>	Josie
<b>Order #:</b>	179658	<b>Registered Name:</b>	GILMAN'S LITTLE RED JOSIE
<b>Ordered By:</b>	Taylor Flynn	<b>Breed:</b>	Miniature Australian Shepherd
<b>Ordered:</b>	March 13, 2023	<b>Sex:</b>	Female
<b>Received:</b>	March 31, 2023	<b>DOB:</b>	March 2021
<b>Reported:</b>	April 11, 2023	<b>Registration #:</b>	ASDM-IA-2216627

### Results:

Disease	Gene	Genotype	Interpretation
Collie Eye Anomaly	<i>NHEJ1</i>	WT/WT	Normal (clear)
Degenerative Myelopathy	<i>SOD1</i>	WT/WT	Normal (clear)
Hereditary Cataracts (Australian Shepherd Type)	<i>HSF4</i>	WT/WT	Normal (clear)
Hyperuricosuria	<i>SLC2A9</i>	WT/WT	Normal (clear)
Intestinal Cobalamin Malabsorption (Australian Shepherd Type)	<i>AMN</i>	WT/WT	Normal (clear)
Intestinal Cobalamin Malabsorption (Border Collie Type)	<i>CUBN</i>	WT/WT	Normal (clear)
Multidrug Resistance 1	<i>ABCB1</i>	WT/M	Carrier (At-Risk)
Multifocal Retinopathy 1	<i>BEST1</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	<i>PRCD</i>	WT/M	Carrier

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

### Interpretation:

Molecular genetic analysis was performed for nine specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in seven of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these seven mutations. However, we identified one normal copy and one mutant copy of the DNA sequences for *ABCB1*. Thus, this dog is a carrier of (and may be at risk for) Multidrug Resistance 1. In addition, we identified one normal copy and one mutant copy of the DNA sequences for *PRCD*. Thus, this dog is a carrier of Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration.

### Recommendations:

Multidrug Resistance 1 is inherited in an autosomal incomplete dominant manner in dogs. Based on this, and the fact that this dog showed a mutation in one copy of the *ABCB1* gene, this dog is a carrier of Multidrug Resistance 1 and at a low risk of developing neurological disease when given certain medications. Dogs affected with this disease lack the ability to remove certain drugs and toxins from the central nervous system putting them at risk for developing neurologic symptoms that could range from tremors, excess salivation, anorexia, and blindness to coma and even death. Though adverse reactions to certain drugs are most commonly seen in dogs having two copies of the mutated gene, carrier dogs can also experience drug sensitivities and dosages need to be adjusted accordingly. Thus, dogs that have one or two mutant copies of the gene are considered at risk for adverse drug reactions. Your veterinarian should be notified that this dog is a carrier for Multidrug Resistance 1 prior to

administration of any medications. When carriers of this mutation are bred with another dog that also is a carrier of the same mutation, there is risk of having affected pups. For each pup that is born to this pairing, there is a 25% chance that the puppy will inherit two copies of the mutation and a 50% chance that the puppy will inherit one copy of the mutation and, in either case, may be susceptible to having adverse drug reactions. Dogs related to this dog have an increased risk to be affected by or carry the mutated gene. Additional testing for this mutation is indicated for related dogs.

Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *PRCD* gene, this dog is a carrier of this disease. Although dogs that carry only one copy of this mutation will not be clinically affected, if bred with another carrier, the pairing could produce affected offspring. To avoid producing affected offspring, this dog should be bred with dogs that are normal (WT/WT) for this gene. Dogs related to this dog have an increased risk to be affected by or carry the mutated gene. Additional testing for this mutation is indicated for related dogs.

Paw Print Genetics® has genetic counseling available to you at no additional charge to answer any questions about these test results, their implications and potential outcomes in breeding this dog.



**Blake C Ballif, PhD**  
Laboratory & Scientific Director



**Christina J Ramirez, PhD, DVM, DACVP**  
Medical Director

Paw Print Genetics® performed the tests listed on this dog. The genes/diseases reported here were selected by the client. Normal results do not exclude inherited mutations not tested in these or other genes that may cause medical problems or may be passed on to offspring. The results included in this report relate only to the items tested using the sample provided. These tests were developed and their performance determined by Paw Print Genetics. This laboratory has established and verified the test(s)' accuracy and precision with >99.9% sensitivity and specificity. The presence of mosaicism may not be detected by this test. Non-paternity may lead to unexpected results. This is not a breed identification test. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think any results are in error, please contact the laboratory immediately for further evaluation. In the event of a valid dispute of results claim, Paw Print Genetics will do its best to resolve such a claim to the customer's satisfaction. If no resolution is possible after investigation by Paw Print Genetics with the cooperation of the customer, the extent of the customer's sole remedy is a refund of the fee paid. In no event shall Paw Print Genetics be liable for indirect, consequential or incidental damages of any kind. Any claim must be asserted within 60 days of the report of the test results.