Paw Print Genetics
A division of Genetic Veterinary Sciences, Inc.

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Laboratory Report

Laboratory #:	100620	**Call Name:**	Pearl
Order #:	44137	**Registered Name:**	Sapphire's Pearl of Planet K9
Ordered By:	Roberta Lilienthal	**Breed:**	Rough Collie
Ordered:	July 25, 2018	**Sex:**	Female
Received:	Aug. 23, 2018	**DOB:**	May 2017
Reported:	Aug. 31, 2018	**Registration #:**	DN50140206

Results:

Disease	Gene	Genotype	Interpretation
Collie Eye Anomaly	*NHEJ1*	WT/M	Carrier
Cyclic Neutropenia	*AP3B1*	WT/WT	Normal (clear)
Degenerative Myelopathy	*SOD1*	WT/WT	Normal (clear)
Hyperuricosuria	*SLC2A9*	WT/WT	Normal (clear)
Multidrug Resistance 1	*ABCB1*	WT/M	Carrier (At-Risk)
Progressive Retinal Atrophy, Rod-Cone Dysplasia 2	*RD3*	WT/WT	Normal (clear)

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

Interpretation:

Molecular genetic analysis was performed for six specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in four of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these four mutations. However, we identified one normal copy and one mutant copy of the DNA sequences for *NHEJ1*. Thus, this dog is a carrier of Collie Eye Anomaly. In addition, 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.

Recommendations:

Collie Eye Anomaly is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *NHEJ1* 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.

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.

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.

**Note: Final results for Collie Eye Anomaly, Degenerative Myelopathy, and Multidrug Resistance 1 were reported to the client via phone on Aug. 15, 2018.*

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