

## Laboratory Report

<b>Laboratory #:</b>	130415	<b>Call Name:</b>	Cookie
<b>Order #:</b>	57324	<b>Registered Name:</b>	Old-Time One Tuff Cookie
<b>Ordered By:</b>	Dianne Armbrust	<b>Breed:</b>	Australian Shepherd
<b>Ordered:</b>	March 26, 2019	<b>Sex:</b>	Female
<b>Received:</b>	April 5, 2019	<b>DOB:</b>	Dec. 2017
<b>Reported:</b>	April 19, 2019	<b>Registration #:</b>	-

### Results:

Disease	Gene	Genotype	Interpretation
Collie Eye Anomaly	<i>NHEJ1</i>	WT/WT	Normal (clear)
Cone Degeneration	<i>CNGB3</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/M	Carrier
Intestinal Cobalamin Malabsorption (Australian Shepherd Type)	<i>AMN</i>	WT/WT	Normal (clear)
Multidrug Resistance 1	<i>ABCB1</i>	WT/WT	Normal (clear)
Multifocal Retinopathy 1	<i>BEST1</i>	WT/M	Carrier
Neuronal Ceroid Lipofuscinosis 6	<i>CLN6</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	<i>PRCD</i>	WT/WT	Normal (clear)

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

### Interpretation:

Molecular genetic analysis was performed for 10 specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in eight of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these eight mutations. However, we identified one normal copy and one mutant copy of the DNA sequences for *SLC2A9*. Thus, this dog is a carrier of Hyperuricosuria. In addition, we identified one normal copy and one mutant copy of the DNA sequences for *BEST1*. Thus, this dog is a carrier of Multifocal Retinopathy 1.


### Recommendations:

Hyperuricosuria is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *SLC2A9* 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.

Multifocal Retinopathy 1 is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *BEST1* 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.



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**Helen F Smith, PhD**  
Assistant Laboratory Director



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

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. These tests were developed and their performance determined by Paw Print Genetics®. This laboratory has established and verified the tests' accuracy and precision. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think these 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.