

## Laboratory Report

<b>Laboratory #:</b>	294659	<b>Call Name:</b>	Jayla
<b>Order #:</b>	133749	<b>Registered Name:</b>	I. Bar Jayla
<b>Ordered By:</b>	Ivan Fisher	<b>Breed:</b>	Standard Smooth Dachshund
<b>Ordered:</b>	March 3, 2022	<b>Sex:</b>	Female
<b>Received:</b>	March 25, 2022	<b>DOB:</b>	July 2020
<b>Reported:</b>	April 4, 2022	<b>Registration #:</b>	HP60500405

### Results:

Disease	Gene	Genotype	Interpretation
Neuronal Ceroid Lipofuscinosis 1	<i>PPT1</i>	WT/WT	Normal (clear)
Neuronal Ceroid Lipofuscinosis 2	<i>TPP1</i>	WT/WT	Normal (clear)
Osteogenesis Imperfecta (Dachshund Type)	<i>SERPINH1</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy (Giant Schnauzer Type)	<i>NECAP1</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy	<i>NPHP4</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4	<i>RPGRIP1</i>	WT/M	Carrier

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 five of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these five mutations. However, we identified one normal copy and one mutant copy of the DNA sequences for *RPGRIP1*. Thus, this dog is a carrier of Progressive Retinal Atrophy, Cone-Rod Dystrophy 4.

### Recommendations:

Progressive Retinal Atrophy, Cone-Rod Dystrophy 4 is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *RPGRIP1* 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.