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## **Laboratory Report**

**Laboratory #:** 245037 **Call Name:** Rascal

Order #: 111291 Registered Name: Regen's Runs With Scissors

Ordered By: Denise Misaras Breed: Labrador Retriever

**Ordered:** June 25, 2021 **Sex:** Male

 Received:
 July 13, 2021
 DOB:
 March 2020

 Reported:
 July 21, 2021
 Registration #:
 SS18177605

**Microchip #:** 941000021495168

## **Results:**

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	PTPLA	WT/WT	Normal (clear)
Cone Degeneration (Labrador Retriever Type)	CNGA3	WT/WT	Normal (clear)
Congenital Myasthenic Syndrome (Labrador Retriever Type)	COLQ	WT/WT	Normal (clear)
Cystinuria (Labrador Retriever Type)	SLC3A1	WT/WT	Normal (clear)
Degenerative Myelopathy	SOD1	WT/WT	Normal (clear)
Elliptocytosis	SPTB	WT/WT	Normal (clear)
Exercise-Induced Collapse	DNM1	WT/WT	Normal (clear)
Hereditary Nasal Parakeratosis	SUV39H2	WT/WT	Normal (clear)
Hyperuricosuria	SLC2A9	WT/WT	Normal (clear)
Ichthyosis (Golden Retriever Type)	PNPLA1	WT/WT	Normal (clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	CHST6	WT/WT	Normal (clear)
Myotubular Myopathy 1	MTM1	WT/Y	Normal/Clear Male
Narcolepsy (Labrador Retriever Type)	HCRTR2	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4	RPGRIP1	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Golden Retriever 2	TTC8	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	PRCD	WT/WT	Normal (clear)
Pyruvate Kinase Deficiency (Labrador Retriever Type)	PKLR	WT/WT	Normal (clear)
Retinal Dysplasia/Oculoskeletal Dysplasia 1	COL9A3	WT/WT	Normal (clear)
Skeletal Dysplasia 2	COL11A2	WT/WT	Normal (clear)
Stargardt Disease	ABCA4	WT/M	Carrier

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

## Interpretation:

Molecular genetic analysis was performed for 20 specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in 19 of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 19 mutations. However, we identified one normal copy

and one mutant copy of the DNA sequences for ABCA4. Thus, this dog is a carrier of Stargardt Disease.

## **Recommendations:**

Stargardt Disease is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *ABCA4* 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<sup>®</sup> 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

Casey R Carl, DVM
Associate 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.