Orivet

Single Report

Animal Name: Ginny

Owner:
Kimberly Horn
Membership Number : 9089380176
Member Body/Breed Club: Rocky Creek Labradors

Approved Collection Method: ☑Yes

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Single Report

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Owner's details

Name:	Kimberly Horn

Animal's Details

Registered Name :	Rockycreek & WillOMoor Custom Made
Pet Name :	Ginny
Registration Number :	SS31035107
Breed :	Labrador Retriever
Microchip Number :	956000014250660
Sex :	Female
Date of Birth :	9th Dec 2021
Colour :	Black

Sample Collection Details

Case Number :	22M000070
Collected By :	MC28570US
Approved Collection :	Yes
Sample Type :	SWAB

Test Details

Test Requested :	Copper Toxicosis (ATP7B & ATP7A) (Labrador Retriever Type)
Pet Name :	Ginny
Date of Test :	19th Aug 2022

Authorisation

Sample with Lab ID Number 22M000070 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

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George Sofronidis BSc (Hons)

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Dr Noam Pik BVSc, MAVS

ISO 9001 Quality Management

ORIVET LAB CERTIFIED ANALYSIS

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Animal's Details

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Sample with Lab ID Number 22M000070 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : COPPER TOXICOSIS (ATP7B & ATP7A) (LABRADOR RETRIEVER TYPE)

Result : CARRIER OF THE ATP7B VARIANT / NEGATIVE FOR THE ATP7A VARIANT [1]

Gene : ATPase copper transporting beta (ATP7B) on chromosome 22ATPase copper transporting alpha (ATP7A) on chromosome X

Variant Detected : Base SubstitutionATP7A: c.980C>TATP7A: p.Thy327IleATP7B: c.4358G>AATP7B: p.Arg1453Gln

There are two mutations, ATP7A:c.980C>T and ATP7B:c.4358G>A are run and reported. The two mutations work differently, ATP7B:c.4358G>A is associated with an increase in hepatic copper levels and ATP7A:c.980C>T is associated with a decrease in hepatic copper levels. The scientific literature suggests that if both mutations are present, the ATP7A attenuates some of the effect of the causative mutation. You can think of this as ATP7B:c.4358G>A being the variant for "at risk" and ATP7A:c.980C>T being the "protective" variant.The effect of the phenotype is that ATP7B is associated with hepatic copper accumulation which induces hepatic cirrhosis usually in middle-aged dogs. The mode of inheritance is complex disease whereby the ATP7B variant leads to increase hepatic copper accumulation over a long period of time which may lead to copper toxicosis.

Clarification of Genetic Testing

Genetic inheritance is not a simple process, and may be complicated by several factors. Below is some information to help clarify these factors.

1) Some diseases may demonstrate signs of what Geneticists call "genetic heterogeneity". This is a term to describe an apparently single condition that may be caused by more than one mutation and/or gene

2) It is possible that there exists more than one disease that presents in a similar fashion and segregates in a single breed. These conditions – although phenotypically similar – may be caused by separate mutations and/or genes.

3) It is possible that the disease affecting your breed may be what Geneticists call an "oligogenic disease". This is a term to describe the existence of additional genes that may modify the action of a dominant gene associated with a disease. These modifier genes may for example give rise to a variable age of onset for a particular condition, or affect the penetrance of a particular mutation such that some animals may never develop the condition.

The range of hereditary diseases continues to increase and we see some that are relatively benign and others that can cause severe and/or fatal disease. Diagnosis of any disease should be based on pedigree history, clinical signs, history (incidence) of the disease and the specific genetic test for the disease. Penetrance of a disease will always vary not only from breed to breed but within a breed, and will vary with different diseases. Factors that influence penetrance are genetics, nutrition and environment. Although genetic testing should be a priority for breeders, we strongly recommend that temperament and phenotype also be considered when breeding.