GENETIC ANALYSIS SINGLE REPORT

OWNER'S DETAILS

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COLLECTION DETAILS

Case Number	:	19143700
Date of Test	:	6th Mar 2019
Collected By	:	
Approved Collection	:	**NO**

ANIMAL'S DETAILS

Registered Name	:	Hillmeadow Beatitude
Pet Name	:	Beatrix
Registration Number	:	215000092
Breed	:	Australian Cobberdog
Microchip Number	:	952000001048862
Sex	:	Intact Female
Date of Birth	:	24th Jan 2017
Colour	:	Parchment

Sample with Lab ID Number 19143700 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

TEST REPORTED	:	**COPPER TOXICOSIS (ATP7B & ATP7A) LABRADOR RETRIEVER TYPE - RESEARCH ONLY**
RESULT	:	**NEGATIVE FOR ATP7B / CARRIER OF THE ATP7A VARIANT[1]**
GENE	:	**ATPASE COPPER TRANSPORTING BETA (ATP7B) ON CHROMOSOME 22 ATPASE COPPER TRANSPORTING ALPHA (ATP7A) ON CHROMOSOME X**
VARIANT DETECTED	:	**BASE SUBSTITUTION ATP7A: C.980C>T ATP7A: P.THY327ILE ATP7B: C.4358G>A ATP7B: P.ARG1453GLN**

[1] 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.

RESULTS REVIEWED & CONFIRMED BY:

Dr. Noam Pik BVSc, BMVS, MBA, MACVS

George Sofronidis BSc(Hons)

CLARIFICATION OF GENETIC TESTING

The goal of genetic testing is to provide breeders with relevant information to improve breeding practices in the interest of animal health. However, 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.

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