

# GENETIC ANALYSIS REPORT

## OWNER'S DETAILS

Lauren Sikkema  
8055 Concession 6  
Moorefield  
N0G2K0 CA



## ANIMAL'S DETAILS

Registered Name:		Pet Name:	Mizpah
Registration Number:		Breed:	Australian Cobberdog
Microchip Number:		Sex:	
Date of Birth:	//	Colour:	

## COLLECTION DETAILS

Case Number:	17089344	Date of Test:	29/09/2017
Approved Collection Method:	NO (Collected by Owner)	Collected By:	

*Sample with Lab ID Number 17089344 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:*

## TESTS REPORTED

## RESULT <sup>1</sup>

<sup>1</sup>**Please Note:** This is a summary disease and trait report. To view more details on each test, including a DNA profile, please log in to your account and view the detailed single DNA report.

### *Urogenital (Associated with the Urinary and Genital Tracts)*

AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CANINE HYPERURICOSURIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CYSTINURIA (NEWFOUNDLAND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Dermatologic (Associated with Skin)*

BLACK HAIR FOLLICULAR DYSPLASIA	NEGATIVE - NOT SHOWING THE PHENOTYPE
COAT COLOUR DILUTION ALOPECIA	INDETERMINABLE [INCONCLUSIVE RESULT]
DRY NOSE (HEREDITARY NASAL PARAKERATOSIS)	NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Neurologic (Associated with the Brain, Spinal and Nerves)*

CENTRONUCLEAR MYOPATHY (LABRADOR RETRIEVER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CONGENITAL MYASTHENIC SYNDROME (LABRADOR RETRIEVER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
DEGENERATIVE MYELOPATHY	<b>CARRIER [ONE COPY OF THE VARIANT DETECTED]</b>
EPISODIC FALLING SYNDROME	NEGATIVE / CLEAR [NO VARIANT DETECTED]
EXERCISE INDUCED COLLAPSE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
IVERMECTIN SENSITIVITY MDR1 (MULTI DRUG RESISTANCE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NARCOLEPSY (LABRADOR)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEONATAL ENCEPHALOPATHY	NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Ophthalmologic (Associated with the Eyes)*

COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CONE-ROD DYSTROPHY I - PRA (CORD I)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
GENERALISED PRA 1	NEGATIVE / CLEAR [NO VARIANT DETECTED]
OCULO-SKELETAL DYSPLASIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
PROGRESSIVE RETINAL ATROPHY - RCD4	INDETERMINABLE [INCONCLUSIVE RESULT]
PROGRESSIVE ROD CONE DEGENERATION (PRCD) - PRA	NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Metabolic (Associated with the Body's Enzymes and Cell Metabolism)*

COPPER TOXICOSIS (ATP7B & ATP7A) LABRADOR RETRIEVER TYPE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
GANGLIOSIDOSIS GM2 (HEX B)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
MALIGNANT HYPERTHERMIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
PHOSPHOFRUCTOKINASE DEFICIENCY	NEGATIVE / CLEAR [NO VARIANT DETECTED]

PYRUVATE KINASE DEFICIENCY (CANINE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

**Haemolympathic (Associated with the Circulatory System)**

ELLIPTOCYTOSIS (B-SPECTRIN)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GLOBOID CELL LEUKODYSTROPHY/KRABBE'S DISEASE

NEGATIVE / CLEAR [NO VARIANT DETECTED]

VON WILLEBRAND'S DISEASE TYPE I

NEGATIVE / CLEAR [NO VARIANT DETECTED]

**Musculoskeletal (Associated with Bones and Muscles)**

MUCOPOLYSACCHARIDOSIS VI

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTUBULAR MYOPATHY X-LINKED

NEGATIVE / CLEAR [NO VARIANT DETECTED]

SKELETAL DYSPLASIA 2 (DWARFISM SD2)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

**Immunological (Associated with the Immune System)**

TRAPPED NEUTROPHIL SYNDROME

NEGATIVE / CLEAR [NO VARIANT DETECTED]

**Trait (Associated with Phenotype)**

A LOCUS (FAWN/SABLE; TRI/TAN POINTS)

a<sup>t</sup>/a - TRI COLOUR / TAN POINTS [CARRYING BICOLOUR GENE]

BLACK AND TAN/SADDLE COAT COLOUR

CARRIER - CARRYING ONE COPY OF THE PHENOTYPE

BROWN (345DELPRO) DELETION

Bb<sup>d</sup> - CARRIER OF BROWN/CHOCOLATE/LIVER (DELETION)

BROWN (GLNT331STOP) STOP CODON

BB<sup>s</sup> - DOES NOT CARRY BROWN or CHOCOLATE (STOP CODON)

BROWN (SER41CYS) INSERTION CODON

Bb<sup>c</sup> - CARRIER OF BROWN/LIVER/CHOCOLATE [INSERTION]

D (DILUTE) LOCUS

DD - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL

E LOCUS - (CREAM/RED/YELLOW)

EE - DOMINANT BLACK DOES NOT CARRY YELLOW/RED/WHITE

EM (MC1R) LOCUS - MELANISTIC MASK

E<sup>m</sup>/E<sup>m</sup> - TWO MASK ALLELES DEPENDS ON A and K SERIES

K LOCUS (DOMINANT BLACK)

K<sup>B</sup>/k<sup>y</sup> - ONE COPY DOMINANT BLACK (K<sup>B</sup>) and ONE COPY NON BLACK (k<sup>y</sup>) or BRINDLE (k<sup>br</sup>)

LONG HAIR GENE (CANINE)

POSITIVE - SHOWING THE PHENOTYPE

**RESULTS REVIEWED AND CONFIRMED BY:**



Dr. Noam Pik BVSc, BMVS, MBA, MACVSc

George Sofronidis BSc (Hons)



## **EXPLANATION of RESULT TERMINOLOGY**

The terms below are provided to help clarify certain results phrases on your genetic report. The phrases below are those as reported by Orivet and may vary from one laboratory to the other.

### **NEGATIVE / CLEAR [NO VARIANT DETECTED]**

No presence of the variant (mutation) has been detected. The animal is clear of the disease and will not pass on any disease-causing mutation.

### **CARRIER [ONE COPY OF THE VARIANT DETECTED]**

This is also referred to as HETEROZYGOUS. One copy of the normal gene and copy of the affected (mutant) gene has been detected. The animal will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal - if breeding with another carrier or affected or unknown then it may produce an affected offspring.

### **POSITIVE / AT RISK [TWO COPIES OF THE VARIANT DETECTED]**

Two copies of the disease gene variant (mutation) have been detected also referred to as HOMOZYGOUS for the variant. The animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a Veterinarian.

### **POSITIVE HETEROZYGOUS [ONE COPY OF THE DOMINANT VARIANT DETECTED]**

Also referred to as POSITIVE ONE COPY or POSITIVE HETEROZYGOUS. This result is associated with a disease that has a dominant mode of inheritance. One copy of the normal gene (wild type) and affected (mutant) gene is present. Appropriate treatment should be pursued by consulting a Veterinarian. This result can still be used to produce a clear offspring.

### **POSITIVE HOMOZYGOUS [TWO COPIES OF THE DOMINANT VARIANT DETECTED]**

Also referred to as POSITIVE HOMOZYGOUS. Two copies of the disease gene variant (mutant) have been detected and the animal may show symptoms associated with the disease. Please Note: This disease has dominant mode of inheritance so if mated to a clear animal ALL offspring with be AFFECTED – HETEROZYGOUS ONE COPY.

### **NORMAL BY PARENTAGE HISTORY**

The sample submitted has had its parentage verified by DNA. By interrogating the DNA profiles of the Dam, Sire and Offspring this information together with the history submitted for the parents excludes this animal from having this disease. The controls run confirm that the dog is NORMAL for the disease requested.

### **NORMAL BY PEDIGREE**

The sample submitted has had its parentage verified by Pedigree. The pedigree has been provided and details (genetic testing reports) of the parents have been included. Parentage could not be determined via DNA profile as no sample was submitted.

### **NO RESULTS AVAILABLE**

Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss.

### **INDETERMINABLE**

The sample submitted has failed to give a conclusive result. This result is mainly due to the sample failing to "cluster" or result in the current grouping. This will be repeated and looked at manually; if a result cannot be determined, a recollection may be requested.

### **DNA PROFILE**

Also known as a DNA fingerprint. This is unique for the animal. No animal shares the same DNA profile. An individual's DNA profile is inherited from both parents and can be used for verifying parentage (pedigrees). This profile contains no disease or trait information and is simply a unique DNA signature for that animal.

### **PARENTAGE VERIFICATION**

#### **QUALIFIES/CONFIRMED or DOES NOT QUALIFY/EXCLUDED**

Parentage is determined by examining the markers on the DNA profile. A result is generated and stated for all DNA parentage requests. Parentage confirmation reports can only be generated if a DNA profile has been carried out for Dam, Offspring and possible Sire/s.

### **PENDING**

Results for this test are still being processed. Some tests are run independently and are reported at a later date. When completed, the result will be emailed.

### **APPROVED COLLECTION METHOD (YES)**

The sample submitted for testing HAS met the requirements recommended by member bodies for the DNA collection process. The animal has been identified via its microchip number (Positive ID) and collected by a Veterinarian or Approved Collection Agent. APPROVED COLLECTION METHOD (NO)

The sample submitted for testing HAS NOT met the requirements recommended by member bodies for the DNA collection process.

### **TRAIT (PHENOTYPE)**

A feature that an animal is born with (a genetically determined characteristic). Traits are a visual phenotype that range from colour to hair length, and also includes certain features such as tail length. If an individual is AFFECTED for a trait then it will show that characteristic eg. AFFECTED for the B (Brown) Locus or bb will be brown/chocolate.

### **POSITIVE – SHOWING THE PHENOTYPE**

The animal is showing the trait or phenotype tested.

## **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.

**Orivet Genetic Pet Care** aims to frequently update breeders with the latest research from the scientific literature. If breeders have any questions regarding a particular condition, please contact us on **(03) 9534 1544** or **admin@orivet.com** and we will be happy to work with you to answer any relevant questions.