

# GENETIC ANALYSIS REPORT



## OWNER'S DETAILS

Allison Britton  
53 Scott Thomas Drive  
Sexton  
Queensland 4570 Australia

## COLLECTION DETAILS

Case Number : 18178183  
Date of Test : 27th Mar 2018  
Collected By :  
**Approved Collection : NO**

## ANIMAL'S DETAILS

Registered Name : Bonus  
Pet Name : Bonus  
Registration Number : 4100268720  
Breed : Border Collie  
Microchip Number : 953010001001253  
Sex : Intact Male  
Date of Birth : 18th Dec 2015  
Colour :

Sample with Lab ID Number 18178183 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

## GENETIC ANALYSIS SUMMARY

**<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.

## TESTS REPORTED

## RESULT <sup>1</sup>

### *Metabolic - Associated with the enzymes and metabolic processes of cells*

COBALAMIN MALABSORPTION: CUBILIN DEFICIENCY (BORDER COLLIE TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Ophthalmologic - Associated with the eyes and associated structures*

COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA NEGATIVE / CLEAR [NO VARIANT DETECTED]  
PRIMARY LENS LUXATION NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Nervous system / Neurologic - Associated with the brain, spinal cord and nerves*

DEGENERATIVE MYELOPATHY NEGATIVE / CLEAR [NO VARIANT DETECTED]  
IVERMECTIN SENSITIVITY MDR1 (MULTI DRUG RESISTANCE) NEGATIVE / CLEAR [NO VARIANT DETECTED]  
NEURONAL CEROID LIPOFUSCINOSIS 5 (BORDER COLLIE TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Musculoskeletal - Associated with muscles, bones and associated structures*

MYOTONIA CONGENITA CLCN1 (CATTLE DOG TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Immunologic - Associated with the organs and cells of the immune system*

TRAPPED NEUTROPHIL SYNDROME (BORDER COLLIE TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Haemolympathic - Associated with the blood and lymph*

VON WILLEBRAND'S DISEASE TYPE II NEGATIVE / CLEAR [NO VARIANT DETECTED]

### *Trait (Associated with Phenotype)*

A LOCUS (FAWN/SABLE; TRI/TAN POINTS)  
BROWN (345DELPRO) DELETION  
BROWN (GLNT331STOP) STOP CODON  
BROWN (SER41CYS) INSERTION CODON

a<sup>t</sup>/a<sup>t</sup> - TAN POINTS - TAN POINTS or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]  
BB<sup>d</sup> - DOES NOT CARRY BROWN or CHOCOLATE (DELETION)  
BB<sup>s</sup> - DOES NOT CARRY BROWN or CHOCOLATE (STOP CODON)  
bb<sup>c</sup> - BROWN/CHOCOLATE, LIVER OR RED (INSERTION)

E LOCUS - (CREAM/RED/YELLOW)  
EM (MC1R) LOCUS - MELANISTIC MASK  
K LOCUS (DOMINANT BLACK)  
LONG HAIR GENE (CANINE)

EE - DOMINANT BLACK DOES NOT CARRY YELLOW/RED/WHITE  
E<sup>m</sup>/E<sup>n</sup> ONE COPY OF MASK ALLELE DETERMINED BY A SERIES  
K / k<sup>y</sup> or k<sup>br</sup>- ONE COPY DOMINANT BLACK (K) and ONE COPY OF  
NON-BLACK (k<sup>y</sup>) dog MAY be brindled  
POSITIVE - SHOWING THE PHENOTYPE

**RESULTS REVIEWED & CONFIRMED BY:**

Dr. Noam Pik BVSc, BMVS, MBA, MACVS



George Sofronidis BSc(Hons)

**ORIVET GENETIC PET CARE**

Suite 102A/ 163 - 169 Inkerman Street,  
St Kilda 3182, Australia  
t +61 3 9534 1544 | f +61 3 9525 3550  
e admin@orivet.com  
www.orivet.com

**ORIVET INTERNATIONAL - USA**

20 Church Street,  
Hartford, CT 06103  
t +844-4 ORIVET (Ext. 105)  
e usa@orivet.com  
www.orivet.com

**ORIVET INTERNATIONAL - JAPAN**

3-6-2, Kumata, Higashisumiyoshi-ku,  
Osaka-shi, Osaka 546-0002, Japan  
t 080 8312 41187 (Japan)  
e japan@orivet.com.au  
www.orivet.jp

Authentication Code



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## **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. A recollection is required at no charge.

### **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 (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.