GENETIC ANALYSIS SINGLE REPORT

OWNER'S DETAILS

GRAUFFEL Véronique
2 rues des Près GRIESBACH LE BASTBERG
BOUXWILLER
67330 France

COLLECTION DETAILS

Case Number	:	19221043
Date of Test	:	20th Nov 2019
Collected By	:	DR ROZET VINCENT
Approved Collection	:	**YES**

ANIMAL'S DETAILS

Registered Name	: Grauffel
Pet Name	: Sparkling Stardust Paige
Registration Number	: 18030
Breed	: Border Collie
Microchip Number	: 250268732651060
Sex	: Intact Female
Date of Birth	: 3rd Aug 2019
Colour	: Tricolore

Sample with Lab ID Number 19221043 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

TEST REPORTED	**: COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA**
RESULT	**: NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
GENE	**: NON-HOMOLOGOUS END JOINING FACTOR 1 (NHEJ1) ON CHROMOSOME 37**
VARIANT DETECTED	**: NUCLEOTIDE DELETION 7799 BASE PAIR DELETION IN INTRON 4 OF THE NHEJ1 GENE**

[1] We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

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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