

# GENETIC ANALYSIS SINGLE REPORT



## OWNER'S DETAILS

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

## COLLECTION DETAILS

Case Number : 19220076  
Date of Test : 20th Nov 2019  
Collected By : DR ROZET VINCENT  
**Approved Collection : YES**

## ANIMAL'S DETAILS

Registered Name : Grauffel  
Pet Name : Pockets du Domaine du Baschberri  
Registration Number : 18030  
Breed : Border Collie  
Microchip Number : 250268600239218  
Sex : Intact Male  
Date of Birth : 13th May 2019  
Colour : Red merle

Sample with Lab ID Number 19220076 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

**TEST REPORTED** : COLLIE EYE ANOMALY/CHOROIDDAL HYPOPLASIA  
**RESULT** : **CARRIER [ONE COPY OF THE VARIANT DETECTED]<sup>1</sup>**  
**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

<sup>1</sup> We have scanned your animal's DNA and one copy of the normal gene and copy of the positive (mutant) gene has been detected. The genotype of the animal tested is CARRIER this result may also be referred to as HETEROZYGOUS or A/N or "+/-". Being an autosomal recessive disease the animal will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal as it may produce affected offspring if mated to another carrier. It is recommended to have any breeding partner tested before breeding.

## 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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