

GENETIC COMPREHENSIVE REPORT



OWNER'S DETAILS

Ming Babington
6 Evans Street
West Pymble
New South Wales 2073 AU

COLLECTION DETAILS

Case Number : 18199243
Date of Test : 7th Nov 2018
Collected By :
Approved Collection : NO

ANIMAL'S DETAILS

Registered Name : Superior First Influential
Servilia
Pet Name : Servilia
Registration Number : 2100505623
Breed : Toy Poodle
Microchip Number : 900006000234865
Sex : Intact Female
Date of Birth : Not Provided
Colour : Red

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

DNA PROFILE

P1_2 GG P3_2 AA P3_3 AG P11_3 CC P12_1 GG P24_2 AG P12_3 GG P30_3 AA P13_1 CC
P24_3 AA P31_1 AA P28_3 TT P31_3 GG P25_1 GG P32_2 CC P13_2 AA P13_3 AC P25_2 GG
P25_3 CC P32_3 AG P33_1 AG P14_1 AA P10_1 GG P26_1 AG P33_3 GG P26_2 AC P14_2 GG
P26_3 AA P14_3 AA P15_1 AG P34_1 AA P34_2 AA P34_3 AA P10_3 CC P15_2 GG P15_3 CC
P16_3 GG P35_1 GG P35_2 AG P36_1 AA P17_1 AA P36_2 CC P37_2 GG P17_2 AA P29_1 CG
P37_3 GG P38_1 CC P38_2 GG P27_1 CG P17_3 GG P27_2 CC P4_3 AG P18_2 CC P18_3 AC
P5_1 AG P11_1 GG P19_1 TT P19_2 AG P5_2 GG P19_3 AG P2_1 CG P2_3 AC P27_3 TT
P20_1 AG P20_3 AA P5_3 GG P11_2 CC P6_2 GG P6_3 CC P21_1 AG P21_3 AG P22_2 AA
P28_1 AG P7_1 AC P7_2 AG P28_2 CC P7_3 AA P29_2 GG P8_1 AA P22_3 GG P8_2 GG
P8_3 AG P23_1 GG P9_3 AT P23_2 CC P23_3 GG P24_1 GG P3_1 GG

RESULTS REVIEWED & CONFIRMED BY:

Dr. Noam Pik BVSc, BMVS, MBA, MACVS



George Sofronidis BSc(Hons)

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GENETIC ANALYSIS

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Degenerative Myelopathy
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : Superoxide dismutase 1 (SOD1) on chromosome 31
Variant Detected : Base Substitution c.118G>A p.Glu40Lys

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.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Progressive Rod Cone Degeneration (prcd) - PRA
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : Photoreceptor disc component (PRCD) on Chromosome 9
Variant Detected : Base Substitution c.5 G>A p.Cys2Tyr

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.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: von Willebrand's Disease Type I
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : VWF1 on Chromosome 27
Variant Detected : Base Substitution of C>T

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.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Mucopolysaccharidosis VI (Poodle Type)
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene :
Variant Detected :

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.

SAMPLE COLLECTION DETAILS

Case Number: 18199243

Approved Collection Method: YES

Date of Test: 7th Nov 2018

Collected By: Andrew Morgan

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This report has been generated by Orivet Genetic Pet Care - (Case Number : 18199243)

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Gangliosidosis GM2 (Poodle Type)
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : Hexosaminidase subunit beta (HEXB) on Chromosome 2
Variant Detected : Nucleotide Deletion c.391delG p.Val95fsX

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.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: E Locus - (Cream/Red/Yellow)
Result: ee - DOG IS HOMOZYGOUS FOR NON-EXTENSION (WHITE/YELLOW/APRICOT)¹
Gene : MC1R
Variant Detected : Em (point mutation) > E (wild type) > e (point mutation)

¹ 2 copies of red/yellow are present referred to as "non-extension". Dog's coat is entirely phaeomelanin based ie. red/yellow/cream/apricot/white. Please note in some breeds an "ee" phenotype can often Colours can be cream to white rather than yellow to red. Shades can vary between littermates.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: EM (MC1R) Locus - Melanistic Mask
Result: Eⁿ/Eⁿ - NO MASKING (Eⁿ) EXTENSION ALLELE¹
Gene : MC1R
Variant Detected : Base Substitution G>A

¹ Dog does not have a mask and has 2 recessive alleles.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: A Locus (Fawn/Sable;Tri/Tan Points)
Result: a^t/a^t - TAN POINTS - TAN POINTS or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]¹
Gene : ASIP
Variant Detected : Base Substitution 246 G>T(A82S); G>A (R83H): C>T (p.R96C)

¹ Homozygous for black and tan/tricolour (no hidden colours) allele. Tri factored/white factored in dogs that have white points. No Bi Factoring (Black White & Tan). Animals are primarily black and have areas of pheomelanin (tan) which tends to be seen on the leg and stomach areas, the side of the head and spots above the eyes. Please note the colour and distribution of pheomelanin "tan" will be dependent on the breed and other colour genes. Please note that any genes on the "A" series will only be expressed if the K locus is kk, kkbr or kbrkbr.

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Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: K Locus (Dominant Black)
Result: K / k^y or k^{br}- ONE COPY DOMINANT BLACK (K) and ONE COPY OF NON-BLACK (k^y) dog MAY be brindled¹
Gene : CBD103
Variant Detected : Deletion of GGG

¹ One copy of non black and one copy of brindle is present. Dog will express the alleles on the A locus but any and all pheomelanin (red) in the coat will be brindled. This allele overrides the ASIP (A) locus. The agouti phenotype may be altered for some breeds and therefore be brindle. There are three alleles at the K Locus with the following dominance hierarchy KB > Kbr > k. The first KB represents dominant black, the second allele Kbr represents brindling and may display A locus gene. Brindle in most breeds appears as black stripes on a red base. Please Note: At this stage no commercial genetic testing can distinguish brindle so breeders should rely on their pedigree or breed standard to exclude or include brindle phenotype.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: D (Dilute) Locus
Result: Dd - CARRIER OF DILUTE (WILL HAVE NORMAL PIGMENT)¹
Gene : MLPH
Variant Detected : Base Substitution

¹ Full colour, carries 1 copy of the dilute gene. May be produce dilute (dd) offspring if mated with another dilute carrier (Dd).

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (GLNT331STOP) Stop Codon
Result: Bb^s - CARRIER of BROWN/CHOCOLATE/LIVER (STOP CODON) ¹
Gene : TYRP1
Variant Detected : Point Mutation

¹ One copy of brown stop codon SNP present – carrier. Can produce brown/chocolate/liver pups if mated with another carrier. Please note this could be a "compound heterozygote" and thus be brown/chocolate. Refer to the other 2 chocolate SNPs to confirm.

Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (345DELPRO) Deletion
Result: BB^d - DOES NOT CARRY BROWN or CHOCOLATE (DELETION) ¹
Gene : TYRP1
Variant Detected : Base Substitution (Point Mutation)

¹ Does not carry brown – cannot have brown offspring.

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Sample with Lab ID Number 18199243 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (SER41CYS) Insertion Codon
Result: BB^c - DOES NOT CARRY BROWN or CHOCOLATE (INSERTION)¹
Gene : TYRP1
Variant Detected : Base Substitution (Point Mutation)

¹ Does not carry brown – cannot have brown offspring.

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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 will 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.

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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.

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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.

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