GENETIC
COMPREHENSIVE REPORT

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TEST DETAILS

Test Requested :	-
Pet Name :	Rutlands Beau Rouge
Date of Test :	20th Mar 2013

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

RESULTS REVIEWED AND CONFIRMED BY

George Sofronidis BSc (Hons)

Dr Noam Pik BVSc, MAVS

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

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Owner's Name : Beverley Rutland Manners
Pet Name : Rutlands Beau Rouge
Microchip Number : Pending

bsi. ISO 9001 Quality Management

ORIVET GENETIC COMPREHENSIVE REPORT

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported :

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : -

Variant Detected : -

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.

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Test Reported :

Result : [1]

Gene : -

Variant Detected : -

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : Collagen type IV alpha 4 chain (COL4A4) on chromosome 25

Variant Detected : Base Substitutionc.115A>Tp.Lys39STOP

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : CANINE DNA PROFILE & PARENTAGE VERIFICATION

Result : [1]

Gene : -

Variant Detected : -

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : CENTRONUCLEAR MYOPATHY (LABRADOR RETRIEVER TYPE)

Result : [1]

Gene : 3-hydroxyacyl-CoA dehydratase 1 (HACD1) also known as PTPLA on chromosome 2

Variant Detected : 236 bp SINE repeat insertion in exon 2 of HACD1

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : CYSTINURIA (NEWFOUNDLAND TYPE)

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : Solute carrier family 3 member 1 (SLC3A1) on chromosome 10

Variant Detected : Base Substitutionc.586C>Tp.Arg221STOP

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 13055910 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 Substitutionc.118G>Ap.Glu40Lys

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : EXERCISE INDUCED COLLAPSE (RETRIEVER TYPE)

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : DNM1

Variant Detected : Base Substitution c.767 G>T

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : MYOTUBULAR MYOPATHY X-LINKED

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : MTM1 on Chromosome X

Variant Detected : Base Substitution c.465C>A

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : NARCOLEPSY (DOBERMANN TYPE)

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : HCRTR2 on Chromosome 12

Variant Detected : Nucleotide Insertion226 bp insertion intron 4

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : NEONATAL ENCEPHALOPATHY (POODLE TYPE)

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : Activating transcription factor 2 (ATF2) on Chromosome 36

Variant Detected : Base Substitutionc.152T>Gp.Met51Arg

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : PHOSPHOFRUCTOKINASE DEFICIENCY (SPANIEL TYPE)

Result : [1]

Gene : Phosphofructokinase muscle (PFKM) on Chromosome 27

Variant Detected : Base Substitutionc.2228G>Ap.Trp743STOP

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : PROGRESSIVE ROD CONE DEGENERATION (PRCD) - PRA

Result : [1]

Gene : Photoreceptor disc component (PRCD) on Chromosome 9

Variant Detected : Base Substitutionc.5 G>Ap.Cys2Tyr

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : GLOBOID CELL LEUKODYSTROPHY/KRABBE'S DISEASE

Result : NEGATIVE / CLEAR [NO VARIANT DETECTED][1]

Gene : Galactosylceramidase (GALC) on Chromosome 8

Variant Detected : Base Substitutionc.473A>Cp.Tyr158Ser

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 13055910 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 : VWF

Variant Detected : c.7437G>A

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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : E LOCUS - (CREAM/RED/YELLOW)

Result : e/e - HOMOZYGOUS FOR NON-EXTENSION [WHITE/YELLOW/APRICOT/WHEATEN][1]

Gene : MC1R

Variant Detected : Em (point mutation) > E (wild type) > e (point mutation) chr5:63694334-63694334: C>T

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/wheaten. 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 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : BROWN COAT COLOUR PROFILE

Result : B/B - DOES NOT CARRY BROWN/LIVER/RED/CHOCOLATE[1]

Gene :

Variant Detected :

Does not carry brown – cannot have brown offspring. Brown or Chocolate is a "modifier" of black. This dog cannot produce any brown/chocolate offspring.

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : D (DILUTE) LOCUS

Result : D/D - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL[1]

Gene : MLPH

Variant Detected : Base Substitution

Full colour, no dilute gene present. The D allele modifies the Melanophillin (MLPH) gene. This animal cannot produce "dilute" offspring. Please Note: There are other dilute variants d2 (Sloughi, Chow Chow & Thai Ridgeback) and rare d3 (Italian Greyhound & Chihuahua) so this test/result may not identify dilute in these breeds.

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : K LOCUS (DOMINANT BLACK)

Result : E^m/E^n - ONE COPY OF MASK ALLELE DETERMINED BY A SERIES[1]

Gene : CBD103

Variant Detected : Deletion of GGG

1 copy of mask and 1 copy of red/yellow – dog has mask and carries red/yellow/cream. Carries one dominant allele and one recessive allele.

Sample with Lab ID Number 13055910 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported : LONG HAIR GENE (CANINE C95F)

Result : POSITIVE / AT RISK [TWO COPIES OF THE VARIANT DETECTED][1]

Gene : FGF5

Variant Detected : p.Cys95Phe c284G>T (Point Mutation)

We have scanned your animal's DNA and two copies of the disease associated variant (mutation) has been detected. The genotype of the animal tested is POSITIVE this result may also be referred to as HOMOZYGOUS, AFFECTED, A/A or "+/+". The animal is "AT-RISK" and MAY show the symptoms (affected) associated with the disease. Penetrance can vary within breed. Appropriate treatment should be pursued by consulting a Veterinarian. Mating with a genetically CLEAR/NORMAL animal will produce 100% CARRIER offspring.

GLOSSARY OF GENETIC TERMS (RESULTS)

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.

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 hairlength, 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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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.

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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.