GENETIC ANALYSIS REPORT

OWNER'S DETAILS

Orivet vet clinic
Dr John Smith

ANIMAL'S DETAILS

Registered Name	:
Pet Name	: Misty
Registration Number	:
Breed	: Domestic Short Hair
Microchip Number	: -382-3084=
Sex	: Neutered Male
Date of Birth	: 31st Dec 2016
Colour	: grey and white

COLLECTION DETAILS

Case Number	: 17168475
Date of Test	: 20th Nov 2017
Collected By	: Noam Pik
Approved Collection	: **YES**

Sample with Lab ID Number 17168475 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

GENETIC ANALYSIS SUMMARY

[1] **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 [1]
Metabolic - Associated with the enzymes and metabolic processes of cells	
ACUTE INTERMITENT PORPHYRIA (DOMESTIC SHORT/LONG HAIR TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
ALPHA MANNISIDOSIS (PERSIAN/DOMESTIC TYPE)*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CHYLOMICRONEMIA - LIPOPROTEIN LIPASE DEFICIENCY (DOMESTIC TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
GLYCOGEN STORAGE DISEASE TYPE IV (NORWEGIAN FOREST CAT TYPE) - SINGLE ASSAY TEST*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HYPEROXALURIA GRHPR (DOMESTIC SHORT/LONG HAIR TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HYPOKALAEMIA PERIODIC POLYMYOPATHY - BURMESE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
LIPOPROTEIN DEFICIENCY (DOMESTIC SHORT/LONG HAIR TYPE)*	CARRIER [ONE COPY OF THE VARIANT DETECTED]
MUCOPOLYSACCHARIDOSIS TYPE I	NEGATIVE / CLEAR [NO VARIANT DETECTED]
PYRUVATE KINASE DEFICIENCY (FELINE)	CARRIER [ONE COPY OF THE VARIANT DETECTED]
VITAMIN D RICKETS	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Urogenital (Associated with the Urinary and Genital Tracts)	
CYSTINURIA SLC3A1 (DOMESTIC SHORT/LONG HAIR TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POLYCYSTIC KIDNEY DISEASE	POSITIVE HETEROZYGOUS [ONE COPY OF THE DOMINANT VARIANT DETECTED]
Nervous system / Neurologic - Associated with the brain, spinal cord and nerves	
GM1 - GANGLIOSIDOSIS*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURODEGENERATIVE LYSOSOMAL STORAGE DISEASE (BURMESE TYPE)*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NIEMANN-PICK DISEASE - SPHINGOMYELINOSIS	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Haemolymphatic - Associated with the blood and lymph	
HAEMOPHILIA B	NEGATIVE / CLEAR [NO VARIANT DETECTED]
MUCOPOLYSACCHARIDOSIS TYPE VI (D520N) NO ASSOCIATION (MILD FORM)	CARRIER for D520 VARIANT/NEGATIVE for L467P & E3511K VARIANT

MUCOPOLYSACCHARIDOSIS TYPE VI (L467P) - SEVERE FORM*	NEGATIVE / CLEAR [NO VARIANT DETECTED]

Ophthalmologic - Associated with the eyes and associated structures

HEREDITARY RETINAL DEGENERATION PRA (CEP290)	NEGATIVE / CLEAR [NO VARIANT DETECTED]

Cardiorespiratory (Associated with Heart and Lungs)

HYPERTROPHIC CARDIOMYOPATHY - MAINE COON	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HYPERTROPHIC CARDIOMYOPATHY - RAGDOLL	POSITIVE HETEROZYGOUS [ONE COPY OF THE DOMINANT VARIANT DETECTED]

Musculoskeletal - Associated with muscles, bones and associated structures

MYOPATHY (COLQ)*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
MYOTONIA CONGENITA (FELINE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SPINAL MUSCULAR ATROPHY	CARRIER [ONE COPY OF THE VARIANT DETECTED]

Trait (Associated with Phenotype)

AGOUTI	a/a - HOMOZYGOUS FOR NON-AGOUTI PATTERN / SOLID COLOURED
ALBINISM - SIAMESE	N/N - NO COPIES OF THE ALBINO VARIANT DETECTED
AMBER	E/E - NO COPIES OF THE MUTATION PRESENT FOR AMBER
BLOOD GROUPS	INDETERMINABLE BLOOD GROUP
CHOCOLATE & CINNAMON	b/b^{l} – BROWN [CARRIER OF CINNAMON]
COLOURPOINT RESTRICTION (SIAMESE/BURMESE)	C/C - FULL COLOUR, CAT DOES NOT CARRY BURMESE (SEPIA) or SIAMESE COLOURPOINT
CURLY COAT - CORNISH REX	N/N - NO COPIES OF THE CORNISH REX VARIANT DETECTED
CURLY COAT - SELKIRK REX	N/N - NO COPIES OF THE SELKIRK REX VARIANT DETECTED
DILUTE (MLPH)	D/d - CARRIER OF DILUTE [ONE COPY OF DILUTE ALLELE PRESENT]
LONG HAIR / SHORT HAIR	N/M1, N/M2, N/M3, N/M4 - CARRIES ONE COPY OF A LONG HAIR VARIANT [CAT HAS SHORT HAIR]
PREAXIAL POLYDACTYL*	NEGATIVE - NO COPY OF THE POLYDACTYL (Pd) VARIANT DETECTED
DEVON REX COAT*	INDETERMINABLE [INCONCLUSIVE RESULT - RECOLLECTION REQUIRED]
SPHYNX (KRT71 VARIANT)	NORMAL - NO SPHYNX VARIANT DETECTED
WHITE GLOVES (BIRMAN PATTERN)	N/N - DOES NOT CARRY THE GLOVING PATTERN

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.