GENETIC ANALYSIS REPORT

OWNER'S DETAILS

Katalin Bodo
Jozsef Attila u 48 HUNGARY!!!!!!
Siofok
8600 Hungary

COLLECTION DETAILS

Case Number	:	19220145
Date of Test	:	22nd Jan 2020
Collected By	:	
Approved Collection	:	**NO**

ANIMAL'S DETAILS

Registered Name	: IT Albafeles Darcy
Pet Name	: Darcy
Registration Number	: HU-0294-2020
Breed	: Birman
Microchip Number	: 380260004161418
Sex	: Intact Male
Date of Birth	: 14th Oct 2018
Colour	: SBI n=seal point

Sample with Lab ID Number 19220145 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]
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	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Metabolic - Associated with the enzymes and metabolic processes of cells	
HYPOKALAEMIA PERIODIC POLYMYOPATHY - BURMESE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Urogenital (Associated with the Urinary and Genital Tracts)	
POLYCYSTIC KIDNEY DISEASE	NEGATIVE / CLEAR [NO VARIANT DETECTED]

RESULTS REVIEWED & CONFIRMED BY:

Dr. Noam Pik BVSc, BMVS, MBA, MACVS

George Sofronidis BSc(Hons)

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Osaka-shi, Osaka 546-0002, Japan
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Authentication Code

Scan To Verify

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.

GENETIC COMPREHENSIVE REPORT

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CHRE1.69446066 G G CHRE1.7439176 A C CHRE2.13480422 A C CHRE2.33619241 G G CHRE2.72592582 A A CHRE3.15324152 A A CHRE3.35126702 A G CHRE3.68341106 A A
CHRF1.2018000 A A CHRF1.24753896 A T CHRF1.35115194 A A CHRF1.60720527 G G CHRF2.11898988 A G CHRF2.20222767 A G CHRF2.29161378 G G CHRF2.47823420 G G
CHRF2.67965848 A A CHRF2.73327273 G G CHRX.109143347 A A CHRX.157577155 A A CHRX.49536490 A A CHRX.5996958 G G

RESULTS REVIEWED & CONFIRMED BY:

Dr. Noam Pik BVSc, BMVS, MBA, MACVS

George Sofronidis BSc(Hons)

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www.orivet.jp

Authentication Code

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GENETIC ANALYSIS

Sample with Lab ID Number 19220145 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported:	Hereditary Retinal Degeneration PRA (CEP290)
Result:	**NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
Gene :	**CEP290 on B4**
Variant Detected :	**IVS50 + 9T>G**

[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 19220145 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported:	Polycystic Kidney Disease
Result:	**NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
Gene :	**PKD1**
Variant Detected :	**Base Substitution C>A**

[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 19220145 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported:	Hypertrophic Cardiomyopathy - Maine Coon
Result:	**NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
Gene :	**MYBPC**
Variant Detected :	**Base Substitution G>C**

[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 19220145 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported:	Hypertrophic Cardiomyopathy - Ragdoll
Result:	**NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
Gene :	**MYBPC**
Variant Detected :	**Base Substitition 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 COLLECTION DETAILS

Authentication Code

Case Number: 19220145

Date of Test: 22nd Jan 2020

Approved Collection Method: NO

Collected By:

Scan To Verify

Sample with Lab ID Number 19220145 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported:	Hypokalaemia Periodic Polymyopathy - Burmese
Result:	**NEGATIVE / CLEAR [NO VARIANT DETECTED][1]**
Gene :	**WNK4**
Variant Detected :	**Base Substitution C>T**

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SAMPLE COLLECTION DETAILS

Authentication Code

Case Number: 19220145

Date of Test: 22nd Jan 2020

Approved Collection Method: NO

Collected By:

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EXPLANATION of RESULT TERMINOLOGY

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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.

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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.

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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.

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INDETERMINABLE
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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
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PENDING
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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.

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GENETIC ANALYSIS DNA PROFILE REPORT

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CHRF1.2018000 A A CHRF1.24753896 A T CHRF1.35115194 A A CHRF1.60720527 G G CHRF2.11898988 A G CHRF2.20222767 A G CHRF2.29161378 G G CHRF2.47823420 G G
CHRF2.67965848 A A CHRF2.73327273 G G CHRX.109143347 A A CHRX.157577155 A A CHRX.49536490 A A CHRX.5996958 G G

RESULTS REVIEWED & CONFIRMED BY:

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Authentication Code

Scan To Verify

II. DESCRIZIONE DELL'ANIMALE
DESCRIPTION OF ANIMAL

FOTO DELL'ANIMALE
(facoltativa)

Picture of the animal
(optional)

1. **Nome**/*Name** ALBAFELES DARCY
2. **Specie**/*Species* FELINA
3. **Razza**/*Breed** BIRMAN
4. **Sesso**/*Sex* M
5. **Data di nascita**/*Date of birth** 14.10.2018
6. **Colore**/*Colour* SEAL POINT
7. **Eventuali tratti o caratteristiche visibili o distintive**/
   Any notable or discernable features or characteristics:

* ***Secondo quanto dichiarato dal proprietario***/*As stated by owner*

IT 030 000281129

XII. VARIE
OTHERS

13.11.2020. Ultrasound test: PKD, HCM: negativ

IT 030 000281129