



Orivet

Genetic Summary Report

Animal Name: Pearl

Owner:

Ashley Edens

Membership Number : Not assigned

Member Body/Breed Club: Not assigned

Approved Collection Method: No



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Accredited and Compliant with



Members of



Harmonization of Genetic Testing for Dogs

Genetic Summary Report



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Owner's details

Name: Ashley Edens

Animal's Details

Registered Name : SS Sapphire's Black Pearl

Pet Name : Pearl

Registration Number : ASDT-IA-2103510

Breed : Miniature Australian Shepherd

Microchip Number : 900235000140997

Sex : Female

Date of Birth : 2nd Dec 2020

Colour : Black Tri

Sample Collection Details

Case Number : 22G00483

Collected By :

Approved Collection : No

Sample Type : SWAB

Test Details

Test Requested : Australian Shepherd - Full Breed Profile

Pet Name : Pearl

Date of Test : 12th Sep 2022

Authorisation

Sample with Lab ID Number 22G00483 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:



George Sofronidis BSc (Hons)



Dr Noam Pik BVSc, MAVS



Genetic Summary Report



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Animal's Details

Registered Name :	SS Sapphire's Black Pearl
Pet Name :	Pearl
Registration Number :	ASDT-IA-2103510
Breed :	Miniature Australian Shepherd
Microchip Number :	900235000140997
Sex :	Female
Date of Birth :	2nd Dec 2020
Colour :	Black Tri

Tests Reported

Diseases	Result
Adult Onset Deafness Border Collie (Linkage Association Test)	NEGATIVE FOR THE EAOD RISK VARIANT [RESEARCH ONLY] - NO COPY DETECTED
Collie Eye Anomaly/Choroidal Hypoplasia	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Cone Degeneration (Shepherd/Arctic Breed Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Degenerative Myelopathy	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Dilated Cardiomyopathy - 2nd VARIANT (Doberman Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Hereditary Cataract (Dominant)	NEGATIVE / CLEAR [NO VARIANT DETECTED]

Owner's Name : Ashley Edens

Pet Name : Pearl

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Tests Reported

Diseases	Result
Hyperuricosuria	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Ivermectin Sensitivity MDR1 (Multi Drug Resistance)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Juvenile Dermatomyositis [MAP3K7CL RISK ALLELE]	bb - NO MAP3K7CL VARIANT DETECTED [THIS IS ONE OF THE 3 LOCI ASSOCIATED WITH DERMATOMYOSITIS]
Multifocal Retinopathy CMR1 (Mastiff/Bull Breeds Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Neuronal Ceroid Lipofuscinosis 6 (Australian Shepherd Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Progressive Rod Cone Degeneration (prcd) - PRA	NEGATIVE / CLEAR [NO VARIANT DETECTED]

Traits	Result
E Locus - (Cream/Red/Yellow)	E/E - DOMINANT BLACK DOES NOT CARRY YELLOW/RED/WHITE
E Locus (Cattle Dog Cream Variant) e2	E ² /E ² - DOMINANT BLACK DOES NOT CARRY "AUSTRALIAN CATTLE DOG" TYPE CREAM
Brown (345DELPRO) Deletion	B ^d /B ^d - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [DELETION]
Brown (GLNT331STOP) Stop Codon	B ^s /b ^s - CARRIER OF BROWN/LIVER/RED/CHOCOLATE [STOP CODON]
Brown (SER41CYS) Insertion Codon	B ^c /B ^c - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [INSERTION]

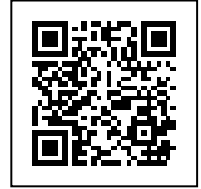
Owner's Name : Ashley Edens

Pet Name : Pearl

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Tests Reported

Traits	Result
B Locus [TYRP1] (Australian Shepherd Type)	B ^a /B ^a - NO COPY OF THE BROWN/RED c.555T>G VARIANT [AUSTRALIAN SHEPHERD TYPE] DETECTED
Liver [TYRP1] (Lancashire Heeler Type)	B ^e /B ^e - DOES NOT CARRY BROWN/LIVER [TYRP1]
D (Dilute) Locus	D/D - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL
K Locus (Dominant Black)	k ^u /k ^u - RECESSIVE NON- BLACK [COLOUR PATTERN DETERMINED BY A LOCUS]
A Locus (Fawn/Sable;Tri/Tan Points)	a ^t /a ^t - TAN POINTS/BLACK & TAN or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]
Pied (BOTH SINE and REPEAT VARIANTS)	S/sp - CARRIER OF PIEBALD [LIMITED WHITE SPOTTING, FLASH OR PARTI]
Merle	m [171bp] / m [171bp] - NON MERLE SOLID COAT (NO CHANGE TO COAT or EYE COLOUR)
Long Hair Gene (Canine C95F)	POSITIVE - SHOWING THE PHENOTYPE
Shedding (MC5R)	shd/shd [HIGH SHEDDING] - TWO COPIES OF THE shd (MC5R) VARIANT DETECTED REFER TO R151W (IC) FOR LEVEL OF SHEDDING
Coat Composition CFA28 Gene (Double/Single Coat)	udc/udc - TWO COPIES OF THE DOUBLE COAT (DENSE UNDERCOAT) PHENOTYPE DETECTED
Curly Coat/Hair Curl (KRT71 R151W)	NEGATIVE FOR THE KRT71 R151W (CU/CU) VARIANT - NOT SHOWING THE CURLY COAT PHENOTYPE

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Traits	Result
Natural Bob Tail (Short Tail Phenotype)	NEGATIVE / CLEAR [NO VARIANT DETECTED]

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Glossary of Genetic Terms (Results)



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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.

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

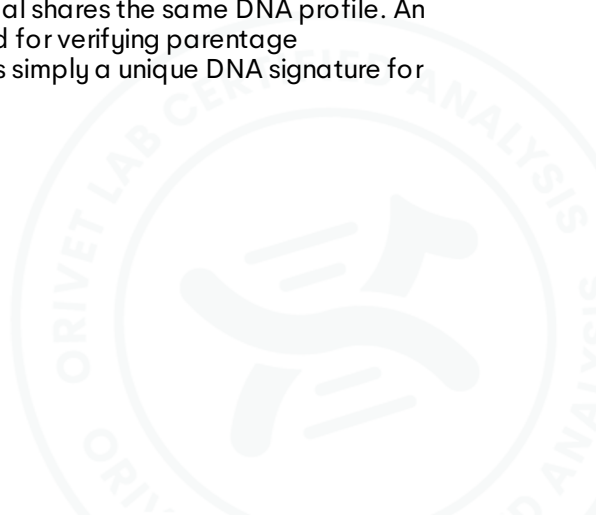
Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss.

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.



Glossary of Genetic Terms (Results)



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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

PENDING

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.

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.

