

# Genetic Summary Report

**Animal Name:** Aspen

**Owner:**

Luke Erb

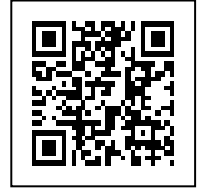
**Membership Number :** Not assigned

**Member Body/Breed Club :** Not assigned

**Approved Collection Method :** NO



## Genetic Summary Report



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### Owner's details

Name : Luke Erb

### Animal's Details

Registered Name : Arrowhead Aspen

Pet Name : Aspen

Registration Number : 5528227F

Breed : Australian Cobberdog

Microchip Number : 952000001266999

Sex : Female

Date of Birth : 16th Sep 2021

Colour : Chocolate

### Sample Collection Details

Case Number : 22G01757

Collected By :

Approved Collection : NO

Sample Type : SWAB

### Test Details

Test Requested : Australian Cobberdog – Full Breed Profile

Pet Name : Aspen

Date of Test : 29th Sep 2022

### Authorisation

Sample with Lab ID Number 22G01757 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

George Sofronidis BSc (Hons)

Dr Noam Pik BVSc, MAVS





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## Animal's Details

Registered Name :	Arrowhead Aspen
Pet Name :	Aspen
Registration Number :	5528227F
Breed :	Australian Cobberdog
Microchip Number :	952000001266999
Sex :	Female
Date of Birth :	16th Sep 2021
Colour :	Chocolate

## Tests Reported

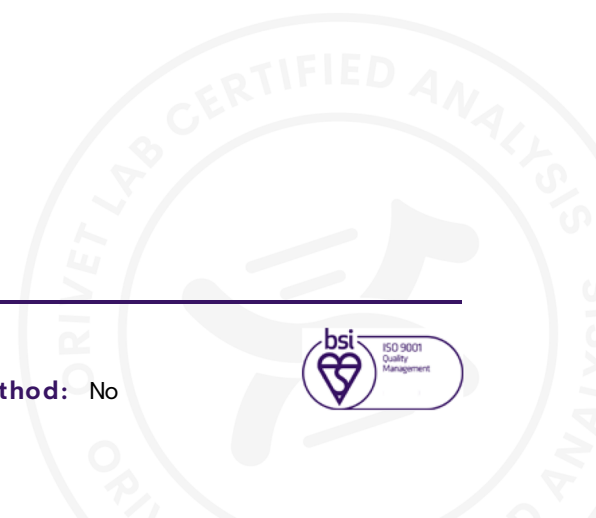
Diseases	Result
Achromatopsia (Labrador Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Autosomal Hereditary Recessive Nephropathy	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Centronuclear Myopathy (Labrador Retriever Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Collie Eye Anomaly/Choroidal Hypoplasia	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Cone-Rod Dystrophy I - PRA (crd -4/crd I)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Congenital Macrothrombocytopenia	NEGATIVE / CLEAR [NO VARIANT DETECTED]

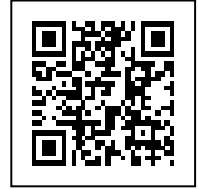
Owner's Name : Luke Erb

Pet Name : Aspen

Microchip Number : 952000001266999

Approved Collection Method: No





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## Tests Reported

Diseases	Result
Congenital Myasthenic Syndrome (Labrador Retriever Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Curly Coat Dry Eye Syndrome (Cavalier Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Cystinuria (SLC3A1) Labrador Retriever Type	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Degenerative Myelopathy	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Ehlers-Danlos Syndrome (Labrador Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Elliptocytosis B-spectrin (Labrador Retriever/Poodle Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Episodic Falling Syndrome (Cavalier Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Exercise Induced Collapse (Retriever Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Gangliosidosis GM2 (Poodle Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Generalised PRA 1 (Golden Retriever Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Globoid Cell Leukodystrophy/Krabbe's Disease	NEGATIVE / CLEAR [NO VARIANT DETECTED]

**Owner's Name :** Luke Erb

**Pet Name :** Aspen

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## Tests Reported

Diseases	Result
Glomerulopathy (PLN) KIRREL2	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Glomerulopathy (PLN) NPHS1	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Hereditary Nasal Parakeratosis/Dry Nose (Labrador Retriever Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Hyperuricosuria	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Ivermectin Sensitivity MDR1 (Multi Drug Resistance)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Macular Corneal Dystrophy (Labrador Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Malignant Hyperthermia	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Microphthalmia, Anophthalmia & Coloboma (Wheaten Terrier Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Mild Disproportionate Dwarfism (Labrador Type)*	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Mucopolysaccharidosis VI (Poodle Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Myotubular Myopathy X-linked*	NEGATIVE / CLEAR [NO VARIANT DETECTED]

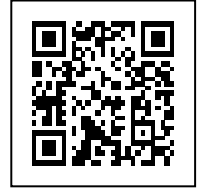
**Owner's Name :** Luke Erb

**Pet Name :** Aspen

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## Tests Reported

Diseases	Result
Narcolepsy (Labrador)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Neonatal Encephalopathy (Poodle Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Phosphofructokinase Deficiency (Spaniel Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Progressive Rod Cone Degeneration (prcd) - PRA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Pyruvate Kinase Deficiency (Canine)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Skeletal Dysplasia 2 (Mild Disproportionate Dwarfism)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Stargardt Disease (Retinal Degeneration)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Trapped Neutrophil Syndrome (Border Collie Type)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
von Willebrand's Disease Type I	NEGATIVE / CLEAR [NO VARIANT DETECTED]

Traits	Result
E Locus - (Cream/Red/Yellow)	E/e - BLACK CARRIES EXTENSION [YELLOW/WHITE/APRICOT/RUBY/RED]
EM (MC1R) Locus - Melanistic Mask	E <sup>m</sup> /E <sup>n</sup> - ONE COPY OF MASK ALLELE DETERMINED BY A SERIES

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## Tests Reported

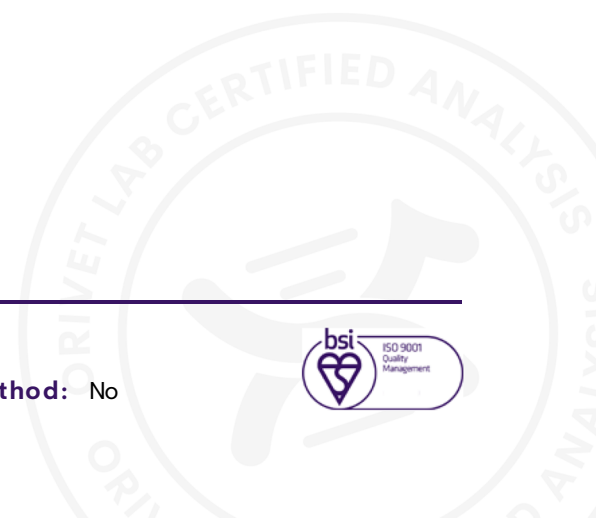
Traits	Result
Brown (345DELPRO) Deletion	B <sup>d</sup> /b <sup>d</sup> - CARRIER OF BROWN/LIVER/RED/CHOCOLATE [DELETION]
Brown (GLNT331STOP) Stop Codon	B <sup>s</sup> /B <sup>s</sup> - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [STOP CODON]
Brown (SER41CYS) Insertion Codon	B <sup>c</sup> /b <sup>c</sup> - CARRIER OF BROWN/LIVER/RED/CHOCOLATE [INSERTION]
Liver [TYRP1] (Lancashire Heeler Type)	B <sup>e</sup> /B <sup>e</sup> - 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/K - DOMINANT BLACK - SOLID [WILL NOT BE BRINDLED or EXPRESS AGOUTI]
A Locus (Fawn/Sable/Tri/Tan Points)	a <sup>t</sup> /a <sup>t</sup> - 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 [LOW SHEDDING] - NO COPIES OF THE SHEDDING (MC5R) VARIANT DETECTED [REFER TO R151W (IC) FOR LEVEL]

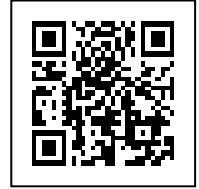
Owner's Name : Luke Erb

Pet Name : Aspen

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## Tests Reported

Traits	Result
Coat Composition CFA28 Gene (Double/Single Coat)	UDC/UDC - NO COPY OF THE DOUBLE COAT (DENSE UNDER COAT) PHENOTYPE DETECTED
Curly Coat/Hair Curl (KRT71 R151W)	ONE COPY OF THE KRT71 R151W (CU/Cu) VARIANT DETECTED - MOST LIKELY TO HAVE MODERATE 'WAVY' CURLY COAT PHENOTYPE
Curly Coat Phenotype (KRT71 - p.Ser422ArgfsTer)	NEGATIVE FOR THE KRT71 (p.Ser422ArgfsTer) VARIANT - NOT SHOWING THE CURLY COAT (C2) PHENOTYPE
Improper Coat (RSPO2)	IC2/IC2 - NO COPY OF THE IMPROPER COAT RSPO2 (DELETION) VARIANT DETECTED

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**Microchip Number :** 952000001266999

**Approved Collection Method:** No





# 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**

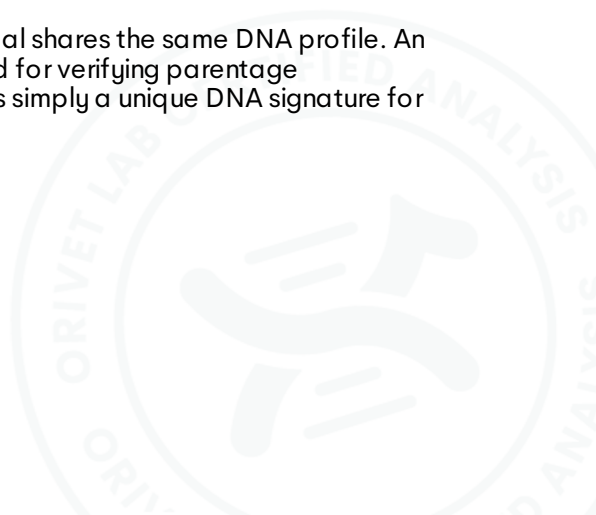
Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss.

## **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.



# 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.

## **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.

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.