

# GENETIC ANALYSIS REPORT

## OWNER'S DETAILS

**Kaylene Steffens**

Po Box 1027

Kenmore, QL 4069



Add: P.O. Box 110  
St Kilda 3182 VIC

Ph: +61 3 9534 1544

Fax: +61 3 9525 3550

email: info@orivet.com.au

website: www.orivet.com.au

A.B.N. 8 722 516 58 99

## ANIMAL'S DETAILS

**Registered Name:** Thornton Anarone

**Pet Name:** True

**Breed:** Labradoodle

**Registration No:** Pending

**Microchip No:** 953010000246899

**Sex:** Female

## COLLECTION DETAILS

**Case Number:** 15-053403

**Date of Test:** 02/05/15

**Collected By:** Kaylene Steffens

**Approved Coll. Mthd.:**

*Sample with Lab ID Number 15-053403 was received at Orivet Genetics, DNA was extracted and analysed with the following results reported:*

**DNA PROFILE** The DNA Profile below represents the genetic identification of Thornton Anarone

SNP01 AG	SNP02 AT	SNP03 GA	SNP04 CC	SNP05 GC	SNP06 CC	SNP07 GC	SNP08 AA	SNP09 AG	SNP10 CA	SNP11
SNP12 GG	SNP13 GA	SNP14 AG	SNP15 CC	SNP16 TT	SNP17 CC	SNP18 GG	SNP19 TT	SNP20 CC	SNP21 CC	SNP22 CC
SNP23 TT	SNP24 CA	SNP25 GG	SNP26	SNP27 GG	SNP28 AA	SNP29 AA	SNP30 GG	SNP31 CC	SNP32 AA	SNP33 CG
SNP34 CC	SNP35 CA	SNP36 GA	SNP37 GG	SNP38 TC	SNP39 TT	SNP40 CT	SNP41 TT	SNP42 GG	SNP43 CC	SNP44 GG
SNP45 CC	SNP46 AG	SNP47 CC	SNP48 TC	SNP49 CA	SNP50 GG	SNP51 GT	SNP52 CC	SNP53 GG	SNP54 CT	SNP55 TT
SNP56 CT	SNP57 AA	SNP58 GG	SNP59 CC	SNP60 TT	SNP61 GG	SNP62 CC	SNP63 TT	SNP64 GG	SNP65 GT	SNP66 TA
SNP67 AA	SNP68 CT	SNP69 CC	SNP70 CC	SNP71 GG	SNP72 GA	SNP73 CC	SNP74 AA	SNP75 CC	SNP76 GT	SNP77 TT
SNP78 GC	SNP79 CT	SNP80 GA	SNP81 GG	SNP82 CC	SNP83 CC	SNP84 CC	SNP85 TA	SNP86 CC	SNP87 GA	SNP88 GG



**RESULTS REVIEWED AND CONFIRMED BY:**

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

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DISEASE(S):	EXERCISE INDUCED COLLAPSE (NORMAL / CLEAR - NO MUTATION DETECTED)
	DEGENERATIVE MYELOPATHY (NORMAL / CLEAR - NO MUTATION DETECTED)
	AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY (NORMAL / CLEAR - NO MUTATION DETECTED)
	CENTRONUCLEAR MYOPATHY (NORMAL / CLEAR - NO MUTATION DETECTED)
	CONE-ROD DYSTROPHY 1 - PRA (NORMAL / CLEAR - NO MUTATION DETECTED)
	CYSTINURIA (NORMAL / CLEAR - NO MUTATION DETECTED)
	GLOBOID CELL LEUKODYSTROPHY/KRABBE'S DISEASE (NORMAL / CLEAR - NO MUTATION DETECTED)
	MUCOPOLYSACCHARIDOSIS (NORMAL / CLEAR - NO MUTATION DETECTED)
	MYOTUBULAR MYOPATHY X LINKED (NORMAL / CLEAR - NO MUTATION DETECTED)
	NARCOLEPSY (NORMAL / CLEAR - NO MUTATION DETECTED)
	NEONATAL ENCEPHALOPATHY (NORMAL / CLEAR - NO MUTATION DETECTED)
	PHOSPHOFRUCTOKINASE (PFK) DEFICIENCY (NORMAL / CLEAR - NO MUTATION DETECTED)
	PYRUVATE KINASE (PK) DEFICIENCY (NORMAL / CLEAR - NO MUTATION DETECTED)
	PROGRESSIVE ROD CONE DEGENERATION - PRA (CARRIER/HETEROZYGOUS - ONE COPY DETECTED)
	VON WILLEBRAND'S DISEASE TYPE I (NORMAL / CLEAR - NO MUTATION DETECTED)
TRAIT(S):	LONG HAIR GENE (PHENOTYPE) (CARRIER/HETEROZYGOUS - ONE COPY DETECTED)
	A-LOCUS AGOUTI (at/at - TRICOLOR/BLACK & TAN (MERLE DEPENDENT NO BIFACTOR))
	B (TYRP1 LOCUS) BROWN/CHOCOLATE (NORMAL - BB FULL COLOR DOES NOT CARRY BROWN)
	DILUTE MLPH GENE (BLUE/GREY) (DD - NO COPY OF MLPH-D ALLELE)
	K-LOCUS (DOM BLACK/WILD TYPE) (KK - DOMINANT FOR K WILL NOT BE BRINDLED or EXPRESS AGOUTI)
	EM-LOCUS MELANISTIC BLACK MASK ALLELE (E <sup>m</sup> E - ONE MASK AND ONE NORMAL EXTENSION ALLELE)
	E-LOCUS (EXTENSION - YELLOW/RED/CREAM/APRICOT (AFFECTED - ee YELLOW, GOLDEN, CREAM or APRICOT)
	SPOTTING LOCUS (W GENE) (NORMAL / CLEAR - NO MUTATION DETECTED)
	FOLLICULAR DYSPLASIA - COLOUR DILUTION ALOPECIA (NORMAL / CLEAR - NO MUTATION DETECTED)
	BLACK HAIR FOLLICULAR DYSPLASIA (CARRIER/HETEROZYGOUS - ONE COPY DETECTED)

Please note: The current B-Locus [TYRP1] does not include the stop codon [bs] mutation. This may cause the result shown for some breeds to be incomplete. Dog may be Bb or bb.



## RESULTS REVIEWED AND CONFIRMED BY:

Dr. Noam Pik BVs MDSV

George Sofronidis BSc (Hons)

## AN OVERVIEW OF GENETIC TESTING - GLOSSARY OF TERMS

*The terms below are provided to help clarify certain items on your genetic reports. The genetic results/terms are those as reported by Orivet.*

**NORMAL/CLEAR - NO MUTATION DETECTED** - No presence of the mutation (wild type) is detected. The animal is clear of disease, will not pass on any disease-causing mutation.

**CARRIER/ HETEROZYGOUS - ONE COPY DETECTED** - One copy of the normal gene (wild type) and affected (mutant) gene is present, 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.

**AFFECTED HETEROZYGOUS (ONE COPY)** - One copy of the normal gene (wild type) and affected (mutant) gene is present, yet due to the dominant mode of inheritance of the disease the animal may show symptoms (affected). Appropriate treatment should be pursued by consulting a veterinarian.

**AFFECTED/ POSITIVE - TWO COPIES** - Two copies of the disease gene (mutant) are present, the animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a veterinarian.

**NORMAL BY PARENTAGE HISTORY** - The sample submitted has had its parentage confirmed- by pedigree or DNA. By definition, 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.

**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

**DNA PROFILE** - Also known as a DNA fingerprint 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). The nomenclature **CSNP** identifies the single nucleotide polymorphism (SNP) at a particular site on the chromosome with each number representing a different site.

**FAIL** - The sample submitted has failed to give a conclusive result. Failures are due mainly to quality/quantity of DNA. We strongly advise that another sample be re-collected and submitted. To minimize bacterial contamination you should allow the swab to air dry (stand up) for at least 3 minutes prior to placing them back into the original swab packaging.

**PARENTAGE CONFIRMATION** - A separate parentage report is generated and emailed for any parentage request. Parentage confirmation report can only be generated if a DNA profile has been carried out for dam, offspring and possible offspring.

**PENDING** - Result for this test is still being processed. When completed, the result will be emailed. Certain tests are run on different chips which can lead to results being uploaded and completed separately.

**INDETERMINABLE** - The samples submitted has failed to give a conclusive result, this result may need to be determined via a manual process. If you have submitted a swab sample you may need to recollect and resubmit a blood sample to enable a conclusive result for the test.

**APPROVED COLLECTION METHOD (YES)** - the sample submitted for testing HAS met the requirements recommended by member bodies for the DNA collection process. The animal has been identified via its microchip number (Positive ID) and collected by a Veterinarian or Approved Collection Agent.

**APPROVED COLLECTION METHOD (BLANK)** - the sample submitted for testing HAS NOT met the requirements recommended by member bodies for the DNA collection process.

**TRAIT** - A feature that an animal is born with (genetically determined characteristic). Traits are visual phenotype that range from colour to hair length, 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.

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**Orivet**  
Genetic Pet Care

**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 no 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 continue to increase and we see some of 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, his (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 the 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 and we will be happy to work with you to answer any relevant questions.**

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