# **GENETIC ANALYSIS REPORT**



### **OWNER'S DETAILS**

Jennifer Parker

## **COLLECTION DETAILS**

Case Number : 20ATTC0646 Date of Test : 19th Feb 2020

Collected By :

Approved Collection: NO

## **ANIMAL'S DETAILS**

Registered Name

Pet Name : Ramble

Registration Number:

Breed : Border Collie

Microchip Number

Sex : Intact Male

Date of Birth : 17th Mar 2019

Colour :

Sample with Lab ID Number 20ATTC0646 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

## **GENETIC ANALYSIS SUMMARY**

DERMATOFIBROSIS (GERMAN SHEPHERD TYPE)

## **TESTS REPORTED**

## RESULT 1

Urinary system / Urologic - Associated with the kidneys, bladder, ureters and urethra

2,8-DIHYDROXYADENINE UROLITHIASIS TYPE IA

ALPORT SYNDROME/ HEREDITARY NEPHROPATHY (SAMOYED TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

AUTOSOMAL HEREDITARY RECESSIVE NEPHROPATHY

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CYSTINURIA (MINIATURE PINSCHER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CYSTINURIA (NEWFOUNDLAND TYPE)

CYSTINURIA (SLC3A1) (AUSTRALIAN CATTLE DOG TYPE)

CYSTINURIA (SLC3A1) LABRADOR RETRIEVER TYPE

NEGATIVE / CLEAR [NO VARIANT DETECTED]

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CYSTINURIA (SLC3A1) LABRADOR RETRIEVER TYPE

FAMILIAL NEPHROPATHY

GLOMERULOPATHY (PLN) KIRREL2

GLOMERULOPATHY (PLN) NPHS1

HEREDITARY NEPHROPATHY

HYPERURICOSURIA

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PRIMARY HYPEROXALURIA

RENAL CYSTADENOCARCINOMA AND NODULAR
RENAL CYSTADENOCARCINOMA AND NODULAR
REPRACTOFISHED TYPE

NEGATIVE / CLEAR [NO VARIANT DETECTED]

Ophthalmologic - Associated with the eyes and associated structures

ACHROMATOPSIA (POINTER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CANINE MULTIFOCAL RETINOPATHY CMR1 (COTON DE TULEAR NEGATIVE / CLEAR [NO VARIANT DETECTED]

CANINE MULTIFOCAL RETINOPATHY CMR2 (COTON DU TULEAR NEGATIVE / CLEAR [NO VARIANT DETECTED]

CANINE MULTIFOCAL RETINOPATHY CMR3 (LAPPHUND TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA CARRIER [ONE COPY OF THE VARIANT DETECTED]

CONE DEGENERATION NEGATIVE / CLEAR [NO VARIANT DETECTED]

CONE-ROD DYSTROPHY I - PRA (CORD I)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CONGENITAL STATIONARY NIGHT BLINDNESS

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CURLY COAT DRY EYE SYNDROME (CAVALIER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

FOCAL EPILEPSY NEGATIVE / CLEAR [NO VARIANT DETECTED]

GENERALISED PRA (SCHAPENDOES TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GENERALISED PRA 1 (GOLDEN RETRIEVER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

<sup>&</sup>lt;sup>1</sup> **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.

GENERALISED PRA 2 (GOLDEN RETRIEVER TYPE) GONIODYSGENESIS AND GLAUCOMA (BORDER COLLIE) HEREDITARY CATARACT MACULAR CORNEAL DYSTROPHY (LABRADOR TYPE) MICROPHTHALMIA, ANOPHTHALMIA & COLOBOMA (WHEATEN TERRIER TYPE)	NEGATIVE / SLEAR [NO VARIANT BETESTED] NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED]	
MULTIFOCAL RETINOPATHY CMR1 (MASTIFF/BULL BREEDS TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
PRIMARY GLAUCOMA PRIMARY LENS LUXATION PRIMARY OPEN ANGLE GLAUCOMA (BEAGLE TYPE) PROGRESSIVE RETINAL ATROPHY - LATE ONSET (BASENJI TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
PROGRESSIVE RETINAL ATROPHY - RCD3 (CORGI/CRESTED TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
PROGRESSIVE RETINAL ATROPHY (MASTIFF) PROGRESSIVE RETINAL ATROPHY (PULI TYPE) PROGRESSIVE RETINAL ATROPHY (SHETLAND SHEEPDOG) PROGRESSIVE RETINAL ATROPHY 3 PROGRESSIVE RETINAL ATROPHY DOMINANT (MASTIFF TYPE) PROGRESSIVE RETINAL ATROPHY PRA1 (PAPILLON TYPE) PROGRESSIVE ROD CONE DEGENERATION (PRCD) - PRA RETINAL DEGENERATION (NORWEGIAN ELKHOUND TYPE) RETINAL DEGENERATION RCD1A TYPE A PRA 1 (MINIATURE SCHNAUZER TYPE) X-LINKED PRA (SAMOYED/HUSKY TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
Respiratory - Associated with the lungs and respiratory system		
ACUTE RESPIRATORY DISTRESS SYNDROME (DALMATIAN TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
PRIMARY CILIARY DYSKINESIA (OLD ENGLISH SHEEPDOG TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
Dental - Associated with the teeth and associated s	tructures	
AMELOGENESIS IMPERFECTA (ITALIAN GREYHOUND TYPE) RAINE SYNDROME DENTAL HYPOMINERALISATION (BORDER COLLIE)	NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED]	
Nervous system / Neurologic - Associated with the brain, spinal cord and nerves		
BRAIN HYPOMYELINATION (WEIMARANER TYPE) CANINE MULTIPLE SYSTEM DEGENERATION (CHINESE	NEGATIVE / CLEAR [NO VARIANT DETECTED]  NEGATIVE / CLEAR [NO VARIANT DETECTED]	
CRESTED)  CEREBELLAR ATAXIA (AMERICAN STAFFORDSHIRE TERRIER	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
TYPE) CEREBELLAR ATAXIA (FINNISH HOUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
CEREBELLAR CORTICAL DEGENERATION (HUNGARIAN VIZSLA TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
CONGENITAL MYASTHENIC SYNDROME (JACK RUSSELL TERRIER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
CONGENITAL MYASTHENIC SYNDROME (LABRADOR RETRIEVER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
CONGENITAL MYASTHENIC SYNDROME (OLD DANISH POINTER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
DEGENERATIVE MYELOPATHY ENCEPHALOPATHY (ALASKAN HUSKY TYPE) EPISODIC FALLING SYNDROME (CAVALIER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED]	
EXERCISE INDUCED COLLAPSE (RETRIEVER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
GENERALISED MYOCLONIC EPILEPSY (RHODESIAN RIDGEBACK TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
HEREDITARY ATAXIA (AUTOPHAGY)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
IVERMECTIN SENSITIVITY MDR1 (MULTI DRUG RESISTANCE) JUVENILE EPILEPSY (BENIGN FAMILIAL) - LAGOTTO	NEGATIVE / CLEAR [NO VARIANT DETECTED]  NEGATIVE / CLEAR [NO VARIANT DETECTED]	
ROMAGNOLO TYPE L2- HYDROXYGLUTARIC ACIDURIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
LEUKOENCEPHALOMYELOPATHY (LEONBERGER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	
NARCOLEPSY (DACHSHUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]	

NARCOLEPSY (DOBERMANN TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NARCOLEPSY (LABRADOR)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEONATAL ATAXIA (COTON DU TULEAR TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEONATAL CEREBELLAR CORTICAL DEGENERATION (BEAGLE TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEONATAL ENCEPHALOPATHY (POODLE TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEUROAXONAL DYSTROPHY (CANE CORSO TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEUROAXONAL DYSTROPHY (PAPILLON TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEUROAXONAL DYSTROPHY (ROTTWEILER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURODEGENERATIVE VACUOLAR STORAGE DISEASE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
(LAGOTTO ROMAGNOLOTYPE)	
NEURONAL CEROID LIPOFUSCINOSIS 1 (DACHSHUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS 10 (AMERICAN BULLDOG TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS 2 (DACHSHUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS 5 (BORDER COLLIE TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS 6 (AUSTRALIAN SHEPHERD TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS 8 (ENGLISH SETTER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
NEURONAL CEROID LIPOFUSCINOSIS A (TIBETAN TERRIER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POLYNEUROPATHY (NDRG1) (ALASKAN MALAMUTE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POLYNEUROPATHY (NDRG1) (GREYHOUND)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POLYNEUROPATHY AND NEURONAL VACUOLATION (JLPP)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POLYNEUROPATHY GJA9 (LEONBERGER/ST BERNARD TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
·	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SANFILIPPO SYNDROME TYPE A / MUCOPOLYSACCHARIDOSIS IIIA (DACHSHUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SPINOCEREBELLAR ATAXIA (CAPN1)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SPINOCEREBELLAR ATAXIA (JACK RUSSELL TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SPONGY DEGENERATION SDCA2	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SPONGY DEGENERATION WITH CEREBELLAR ATAXIA (KCNJ10)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
STARTLE HYPEREKPLEXIA (WOLFHOUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Haemolymphatic - Associated with the blood and lymph	
CANINE CHONDRODYSPLASIA (NORWEGIAN ELKHOUND,	
KARELIAN BEAR)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
CONGENITAL MACROTHROMBOCYTOPENIA	NEGATIVE / CLEAR [NO VARIANT DETECTED]
ELLIPTOCYTOSIS B-SPECTRIN (LABRADOR RETRIEVER/POODLE TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
FACTOR VII DEFICIENCY	NEGATIVE / CLEAR [NO VARIANT DETECTED]
GLOBOID CELL LEUKODYSTROPHY/KRABBE'S DISEASE	NEGATIVE / CLEAR [NO VARIANT DETECTED]
GREY COLLIE SYNDROME (CYCLIC HEMATOPOIESIS) AP3	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HAEMOPHILIA A / FACTOR VIII (GERMAN SHEPHERD TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HAEMOPHILIA B / FACTOR IX (CAIRN TERRIER TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
HAEMOPHILIA B / FACTOR IX G418E	NEGATIVE / CLEAR [NO VARIANT DETECTED]
MACROTHROMBOCYTOPENIA (CAIRN/NORFOLK TERRIER TYPE	
MAY-HEGGLIN ANOMALY (PUG TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
PLATELET DYSFUNCTION	NEGATIVE / CLEAR [NO VARIANT DETECTED]
POST OPERATIVE HAEMORRHAGE / PLATELET DISORDER (MOUNTAIN DOG TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
PREKALLIKREIN DEFICIENCY (SHIH TZU TYPE) SCOTT SYNDROME (GERMAN SHEPHERD TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE / CLEAR [NO VARIANT DETECTED]
THROMBASTHENIC THROMBOPATHIA (OTTERHOUND TYPE)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
VON WILLEBRAND'S DISEASE TYPE I	NEGATIVE / CLEAR [NO VARIANT DETECTED]
VON WILLEBRAND'S DISEASE TYPE II	HETEROZYGOUS [ONE COPY OF THE POLYMORPHIC MUTATION DETECTED]
VON WILLEBRAND'S DISEASE TYPE II (GERMAN WIREHAIRED POINTER)	NEGATIVE / CLEAR [NO VARIANT DETECTED]
VON WILLEBRAND'S DISEASE TYPE III	NEGATIVE / CLEAR [NO VARIANT DETECTED]
Immunologic - Associated with the organs and cells of the immune system	
CANINE LEUKOCYTE ADHESION DEFICIENCY TYPE I (IRISH	NEGATIVE / CLEAR [NO VARIANT DETECTED]
SETTER TYPE)	NEONINE / OLLAN INO VANIANT DETECTED

CANINE LEUKOCYTE ADHESION DEFICIENCY TYPE III (GERMAN NEGATIVE / CLEAR [NO VARIANT DETECTED]
SEFERE COMBINED IMMUNODEFICIENCY (FRISIAN WATER DOG)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

TRAPPED NEUTROPHIL SYNDROME (BORDER COLLIE TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

Metabolic - Associated with the enzymes and metabolic processes of cells

CATALASE DEFICIENCY (BEAGLE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

COBALAMIN MALABSORPTION (BEAGLE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

COBALAMIN MALABSORPTION: CUBILIN DEFICIENCY (BORDER NEGATIVE / CLEAR [NO VARIANT DETECTED]

COLLIE TYPE)

FUCOSIDOSIS (ENGLISH SPRINGER SPANIEL TYPE)

NEGATIVE / GLEAR [NO VARIANT DETECTED]

GANGLIOSIDOSIS (PORTUGUESE WATER DOG TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GANGLIOSIDOSIS GM1 GLB1 (SHIBA INU TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GANGLIOSIDOSIS GM2 (JAPANESE CHIN TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GANGLIOSIDOSIS GM2 (POODLE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GANGLIOSIDOSIS GM2 (FOODEL TITE)

REGATIVE / CLEAR [NO VARIANT DETECTED]

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GLYCOGEN STORAGE DISEASE IA (MALTESE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

GLYCOGEN STORAGE DISEASE III NEGATIVE / CLEAR [NO VARIANT DETECTED]

GLYCOGEN STORAGE DISEASE IIIA (CURLY COAT RETRIEVER NEGATIVE / CLEAR [NO VARIANT DETECTED]

MALIGNANT HYPERTHERMIA NEGATIVE / CLEAR [NO VARIANT DETECTED]

MUCOPOLYSACCHARIDOSIS (HUNTAWAY TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MUCOPOLYSACCHARIDOSIS TYPE I (PLOTT HOUND TYPE)

MUCOPOLYSACCHARIDOSIS VI (GREAT DANE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MUCOPOLYSACCHARIDOSIS VI (POODLE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MUCOPOLYSACCHARIDOSIS VII - TYPE II (GERMAN SHEPHERD/BELGIAN SHEPHERD TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PHOSPHOFRUCTOKINASE DEFICIENCY (GERMAN SPANIEL)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PHOSPHOFRUCTOKINASE DEFICIENCY (SPANIEL TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

POMPES DISEASE (LAPPHUND TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY
(CLUMPER SPANIEL TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

(CLUMBER SPANIEL TYPE)

PYRUVATE KINASE DEFICIENCY (BEAGLE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PYRUVATE KINASE DEFICIENCY (CANINE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PYRUVATE KINASE DEFICIENCY (LABRADOR TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

PYRUVATE KINASE DEFICIENCY (PUG) NEGATIVE / CLEAR [NO VARIANT DETECTED]

Musculoskeletal - Associated with muscles, bones and associated structures

CENTRONUCLEAR MYOPATHY (LABRADOR RETRIEVER TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

CENTRONUCLEAR MYOPATHY /INHERITED MYOPATHY (GREAT DANE TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CHONDRODYSPLASIA ITGA10 (ELKHOUND TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

CRANIOMANDIBULAR OSTEOPATHY (TERRIER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MILD DISPROPORTIONATE DWARFISM (LABRADOR TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MUSCULAR DYSTROPHY (LANDSEER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTONIA CONGENITA (MINIATURE SCHNAUZER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

TOTOMA CONGENITA (MINIATORE SCHNAUZER TIFE)

MYOTONIA CONGENITA CLCN1 (CATTLE DOG TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTONIA HEREDITARIA (CATTLE DOG TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTUBULAR MYOPATHY X-LINKED NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTUBULAR MYOPATHY X-LINKED (LABRADOR RETRIEVER NEGATIVE / CLEAR [NO VARIANT DETECTED]

MYOTUBULAR MYOPATHY X-LINKED (ROTTWEILER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

OSTEOGENESIS IMPERFECTA (CHOW CHOW)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

OSTEOGENESIS IMPERFECTA (GOLDEN RETRIEVER TYPE)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

OSTEOGENESIS IMPERFECTA SERPINH1 (DACHSHUND TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

SKELETAL DYSPLASIA 2 (MILD DISPROPORTIONATE DWARFISM)

NEGATIVE / CLEAR [NO VARIANT DETECTED]

Ontogeny / ontogenesis (or simply developmental) - Developmental (Diseases associated with in-utero development)

CLEFT LIP PALATE (NOVA SCOTIA DUCK TOLLING RETRIEVER NEGATIVE / CLEAR [NO VARIANT DETECTED]

CONGENITAL HYPOTHYROIDISM WITH GOITER (TENTERFIELD NEGATIVE / CLEAR [NO VARIANT DETECTED] TERRIERITALETYPOTHYROIDISM WITH GOITER (TOY FOX NEGATIVE / CLEAR [NO VARIANT DETECTED] TERRIER TYPE) PITUITARY DWARFISM - SINGLE ASSAY TEST NEGATIVE / CLEAR [NO VARIANT DETECTED] Dermatologic - Associated with the skin DYSTROPHIC EPIDERMOLYSIS BULLOSA (ASIAN SHEPHERD NEGATIVE / CLEAR [NO VARIANT DETECTED] TYPE) DYSTROPHIC EPIDERMOLYSIS BULLOSA (GOLDEN RETRIEVER NEGATIVE / CLEAR [NO VARIANT DETECTED] TYPE) ECTODERMAL DYSPLASIA (CHESAPEAKE BAY RETRIEVER NEGATIVE / CLEAR [NO VARIANT DETECTED] TYPE) HEREDITARY FOOTPAD HYPERKERATOSIS NEGATIVE / CLEAR [NO VARIANT DETECTED] HEREDITARY NASAL PARAKERATOSIS/DRY NOSE (LABRADOR NEGATIVE / CLEAR [NO VARIANT DETECTED] RETRIEVER TYPE) ICHTHYOSIS (AMERICAN BULLDOG) NEGATIVE / CLEAR [NO VARIANT DETECTED] ICHTHYOSIS (GERMAN SHEPHERD TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED] ICHTHYOSIS (GREAT DANE) NEGATIVE / CLEAR [NO VARIANT DETECTED] ICHTHYOSIS (NORFOLK TERRIER) NEGATIVE / CLEAR [NO VARIANT DETECTED] ICHTHYOSIS A (GOLDEN RETRIEVER) NEGATIVE / CLEAR [NO VARIANT DETECTED] MUSLADIN-LUEKE SYNDROME (BEAGLE TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED] Digestive system / Gastrointestinal - Associated with the organs and structures of the digestive system GALL BLADDER MUCOCELE FORMATION (SHETLAND NEGATIVE / CLEAR [NO VARIANT DETECTED] SHEEPDOG TYPE) Reproductive - Associated with the reproductive tract MULLERIAN DUCT SYNDROME (MINIATURE SCHNAUZER TYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED] Trait (Associated with Phenotype) E/e - BLACK CARRIES EXTENSION E LOCUS - (CREAM/RED/YELLOW) [YELLOW/WHITE/APRICOT/RUBY/RED] E<sup>2</sup>/E<sup>2</sup> - DOMINANT BLACK DOES NOT CARRY "AUSTRALIAN E LOCUS (CATTLE DOG CREAM VARIANT) E2 CATTLE DOG" TYPE CREAM E3/E3 - DOMINANT BLACK DOES NOT CARRY "HUSKY TYPE" E LOCUS (ARTIC BREEDS PALE/YELLOW/WHITE VARIANT) E3 PALE YELLOW/WHITE Em/En - ONE COPY OF MASK ALLELE DETERMINED BY A EM (MC1R) LOCUS - MELANISTIC MASK **SERIES** Eg/Eg - NO GRIZZLE PHENOTYPE EG LOCUS (GRIZZLE) B<sup>d</sup>/b<sup>d</sup> - CARRIER OF BROWN/LIVER/RED/CHOCOLATE **BROWN (345DELPRO) DELETION** [DELETION] BS/BS - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE BROWN (GLNT331STOP) STOP CODON [STOP CODON] **BROWN (SER41CYS) INSERTION CODON** bc/bc - BROWN/CHOCOLATE, LIVER OR RED [INSERTION] D/D - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS D (DILUTE) LOCUS **NORMAL** DILUTE D2 VARIANT (CHOW CHOW TYPE) D<sup>2</sup>/D<sup>2</sup> - NO COPY OF d2 ALLELE (DILUTE) - PIGMENT IS NORMAL KB / ky or kbr- ONE COPY DOMINANT BLACK (KB) and ONE COPY K LOCUS (DOMINANT BLACK) OF NON-BLACK (ky) dog MAY be brindled at/at - TAN POINTS/BLACK & TAN or TRICOLOUR MAY BE A LOCUS (FAWN/SABLE;TRI/TAN POINTS) BRINDLED [SEE K LOCUS] SPOTTING (W) LOCUS (MASTIFF TYPE) NEGATIVE - NOT SHOWING THE PHENOTYPE POSITIVE TWO COPIES of TAN SADDLE VARIANT - MAY BLACK AND TAN/SADDLE COAT COLOUR EXPRESS THE PHENOTYPE [See E and K Locus] HARLEQUIN (H) PATTERN (GREAT DANE TYPE) h/h - DOES NOT CARRY ANY HARLEQUIN PATTERN LONG HAIR GENE (CANINE C95F) CARRIER - CARRYING ONE COPY OF THE PHENOTYPE NATURAL BOB TAIL (SHORT TAIL PHENOTYPE) NEGATIVE / CLEAR [NO VARIANT DETECTED] NEGATIVE FOR THE SHORTENED HEAD (BRACHYCEPHALY) SKULL DIVERSITY (ALL BREEDS) VARIANT **OCULOCUTANEOUS ALBINISM\*** NEGATIVE / CLEAR [NO VARIANT DETECTED] OCULOCUTANEOUS ALBINISM (BULLMASTIFF) **NEGATIVE - NOT SHOWING THE PHENOTYPE** 

**NEGATIVE - NOT SHOWING THE PHENOTYPE** 

**NEGATIVE - NOT SHOWING THE PHENOTYPE** 

Dermatologic - Associated with the skin

BLACK HAIR FOLLICULAR DYSPLASIA

COAT COLOUR DILUTION ALOPECIA

## **RESULTS REVIEWED & CONFIRMED BY:**

Dr. Noam Pik BVSc, BMVS, MBA, MACVS

George Sofronidis BSc(Hons)

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#### **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.

This report has been generated by Orivet Genetic Pet Care (Case Number : 20ATTC0646)