



# Orivet

# Genetic Comprehensive Report

**Animal Name:** Friska

**Owner:**

Amelie Martin

Membership Number : Not assigned

Member Body/Breed Club: Not assigned

Approved Collection Method:  Yes



[orivet.com](https://orivet.com)

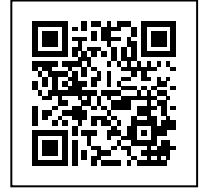
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Harmonization of  
Genetic Testing  
for Dogs



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## Owner's details

Name: Amelie Martin

## Animal's Details

Registered Name :

Pet Name : Friska

Registration Number :

Breed : Goldendoodle

Microchip Number : 96700000056327

Sex : Female

Date of Birth : 16th Dec 2020

Colour : Abricot

## Sample Collection Details

Case Number : 22G03771

Collected By : Dre Ariane Lalonde-Larve

Approved Collection : Yes

Sample Type : SWAB

## Test Details

Test Requested : Goldendoodle - Full Breed Profile

Pet Name : Friska

Date of Test : 8th Apr 2022

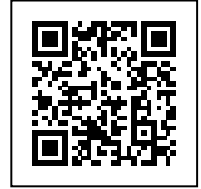
## Authorisation

Sample with Lab ID Number 22G03771 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 :

Pet Name : Friska

Registration Number :

Breed : Goldendoodle

Microchip Number : 967000000056327

Sex : Female

Date of Birth : 16th Dec 2020

Colour : Abricot

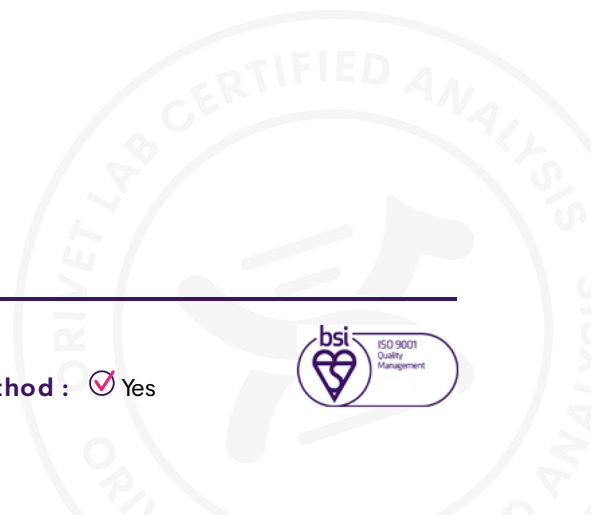
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 P19\_1 A T P19\_2 G G P5\_2 G G P19\_3 G G P2\_1 G G P2\_3 A C P27\_3 A T P20\_1 G G  
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 P8\_2 A G P8\_3 A G P23\_1 C C P9\_3 T T P23\_2 C C P23\_3 A G P24\_1 A G P3\_1 A G

Owner's Name : Amelie Martin

Pet Name : Friska

Microchip Number 967000000056327

Approved Collection Method :  Yes



# Genetic Comprehensive Report

## Animal's Details

Registered Name :	
Pet Name :	Friska
Registration Number :	
Breed :	Goldendoodle
Microchip Number :	96700000056327
Sex :	Female
Date of Birth :	16th Dec 2020
Colour :	Abricot

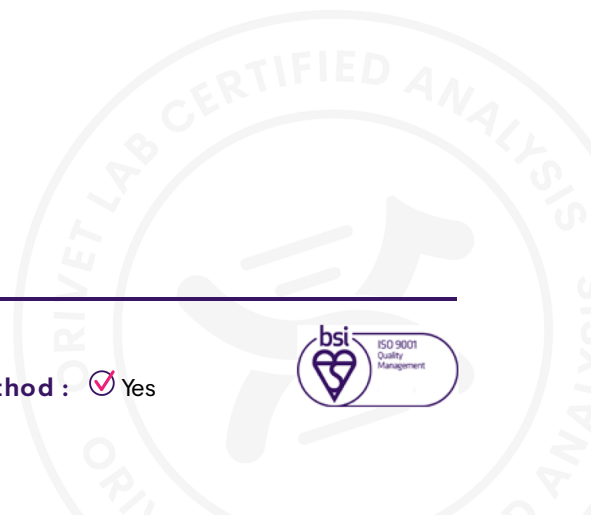
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Owner's Name : Amelie Martin

Pet Name : Friska

Microchip Number 96700000056327

Approved Collection Method :  Yes



# Genetic Comprehensive Report

## Animal's Details

Registered Name :	
Pet Name :	Friska
Registration Number :	
Breed :	Goldendoodle
Microchip Number :	96700000056327
Sex :	Female
Date of Birth :	16th Dec 2020
Colour :	Abrirot

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Owner's Name : Amelie Martin

Pet Name : Friska

Microchip Number 96700000056327

Approved Collection Method :  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** CENTRONUCLEAR MYOPATHY (LABRADOR RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** 3-hydroxyacyl-CoA dehydratase 1 (HACD1) also known as PTPLA on chromosome 2

**Variant Detected :** 236 bp SINE repeat insertion in exon 2 of HACD1

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.

**Test Reported :** CONGENITAL EYE MALFORMATION (GOLDEN RETRIEVER)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** SIX6, chr8

**Variant Detected :** c.487C>Tp.Gln163\*

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.

**Test Reported :** CONGENITAL MACROTHROMBOCYTOPENIA

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Tubulin beta 1 class VI (TUBB1) on Chromosome 24

**Variant Detected :** Base Substitutionc.745G>Ap.Asp249Asn

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.

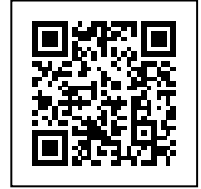
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** CONGENITAL MYASTHENIC SYNDROME (LABRADOR RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** 2-hydroxyacyl-CoA lyase 1 (COLQ) on chromosome 23

**Variant Detected :** Base Substitutionc.1010T>Cp.Ile337Thr

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.

**Test Reported :** CYSTINURIA (SLC3A1) LABRADOR RETRIEVER TYPE

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Solute carrier family 3 member 1 (SLC3A1) on chromosome 10

**Variant Detected :** Nucleotide Deletionc.350delGp.Gly117Alafs\*41

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.

**Test Reported :** DEGENERATIVE MYELOPATHY

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Superoxide dismutase 1 (SOD1) on chromosome 31

**Variant Detected :** Base Substitutionc.118G>Ap.Glu40Lys

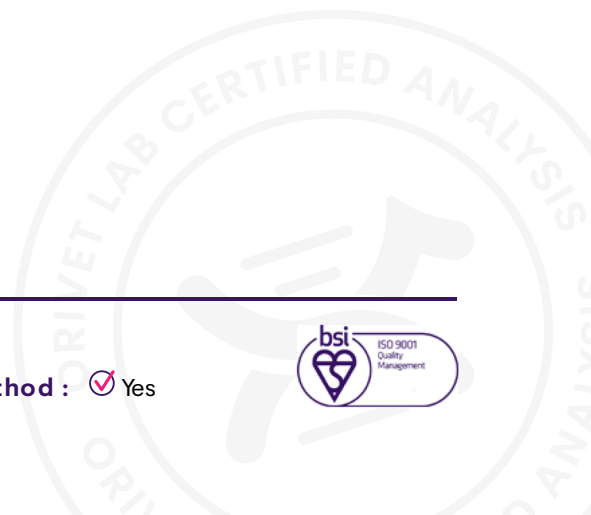
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

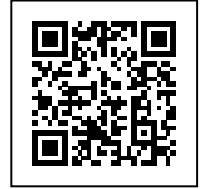
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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## Genetic Comprehensive Report

Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** DYSTROPHIC EPIDERMOLYSIS BULLOSA (GOLDEN RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Collagen type VII alpha 1 chain (COL7A1) Chromosome 20

**Variant Detected :** Base Substitutionc.5797G>Ap.Gly1906Ser

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.

**Test Reported :** ELLIPTOCYTOSIS B-SPECTRIN (LABRADOR RETRIEVER/POODLE TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Spectrin beta erythrocytic (SPTB) Chromosome 8

**Variant Detected :** Base Substitutionc.6384C>TThr2110Met

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.

**Test Reported :** EXERCISE INDUCED COLLAPSE (RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** DNM1

**Variant Detected :** Base Substitution c.767 G>T

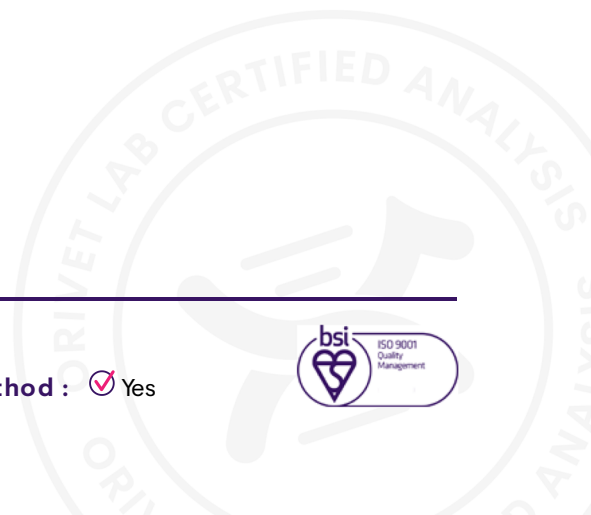
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes







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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** GENERALISED PRA 1 (GOLDEN RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Solute carrier family 4 member 3 (SLC4A3) on chromosome 37

**Variant Detected :** C.2601-2602 Insertion Cp.Glu868Arg-frameshiftX104

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.

**Test Reported :** GENERALISED PRA 2 (GOLDEN RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Tetratricopeptide repeat domain 8 (TTC8) on chromosome 8

**Variant Detected :** c.669delAp.Lys223Arg-frameshiftX15

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.

**Test Reported :** HEREDITARY NASAL PARAKERATOSIS/DRY NOSE (LABRADOR RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Suppressor of variegation 3-9 homolog 2 (SUV39H2) on chromosome 2

**Variant Detected :** Base Substitutionc.972T>Gp.Asn324Lys

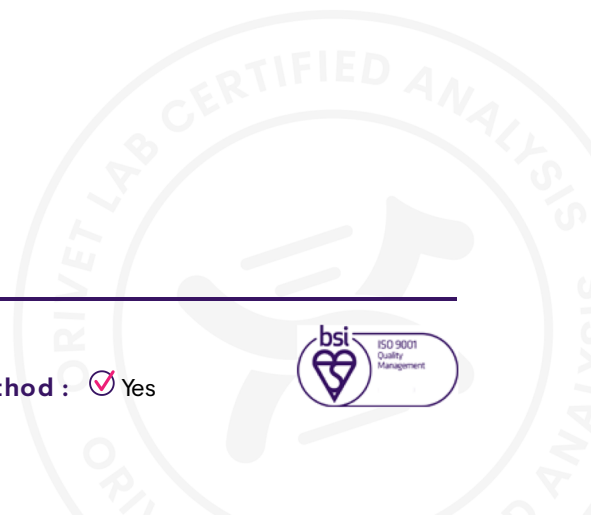
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** HYPERURICOSURIA

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Solute carrier family 2 member 9 (SLC2A9) on chromosome 3

**Variant Detected :** Base Substitutionc.563G>Tp.Cys188Phe

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.

**Test Reported :** ICHTHYOSIS A (GOLDEN RETRIEVER)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Patatin like phospholipase domain containing 1 (PNPLA1) on Chromosome 12

**Variant Detected :**

Nucleotide Insertion and Nucleotide Deletionc.1445-1447delACC and c.1447insTACTACTAp.Asn482Ilefs9X

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.

**Test Reported :** MACULAR CORNEAL DYSTROPHY (LABRADOR TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** LOC4

**Variant Detected :** c.814C>A

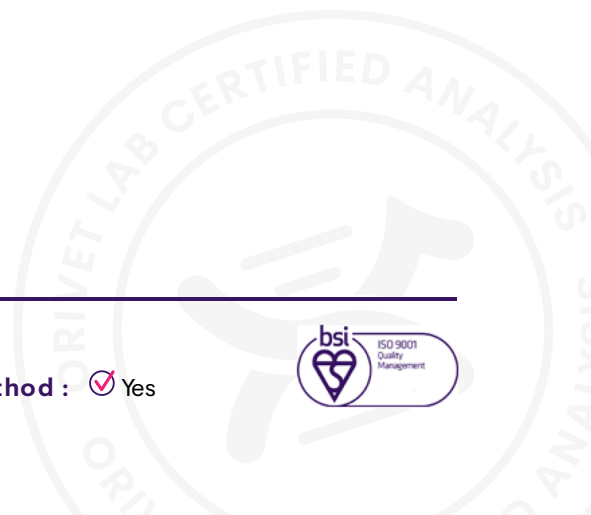
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** MALIGNANT HYPERTHERMIA

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Ryanodine receptor 1 (RYR1) on Chromosome 1

**Variant Detected :** Base Substitutionc.1640T>Cp.Val547Ala

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.

**Test Reported :** MUCOPOLYSACCHARIDOSIS VI (POODLETYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :**

**Variant Detected :**

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.

**Test Reported :** MYOTUBULAR MYOPATHY X-LINKED (LABRADOR RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Myotubularin 1 (MTM1) on Chromosome X

**Variant Detected :** Base Substitutionc.465C>Ap.Asn155Lys

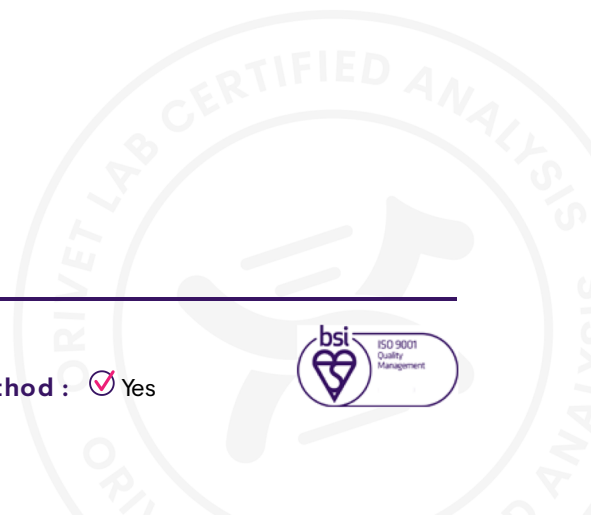
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

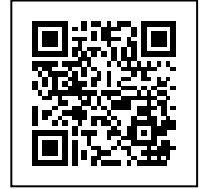
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** NARCOLEPSY (LABRADOR)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Hypocretin receptor 2 (HCRT2) on Chromosome 12

**Variant Detected :** Base Substitutionc.1105+5G>Asplice site mutation

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.

**Test Reported :** NEONATAL ENCEPHALOPATHY (POODLE TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Activating transcription factor 2 (ATF2) on Chromosome 36

**Variant Detected :** Base Substitutionc.152T>Gp.Met51Arg

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.

**Test Reported :** NEURONAL CEROID LIPOFUSCINOSIS NCL (GOLDEN RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** CLN5 intracellular trafficking protein (CLN5) on Chromosome 22

**Variant Detected :** Nucleotide Deletionc.934\_935delAGp.E312Vfs\*6

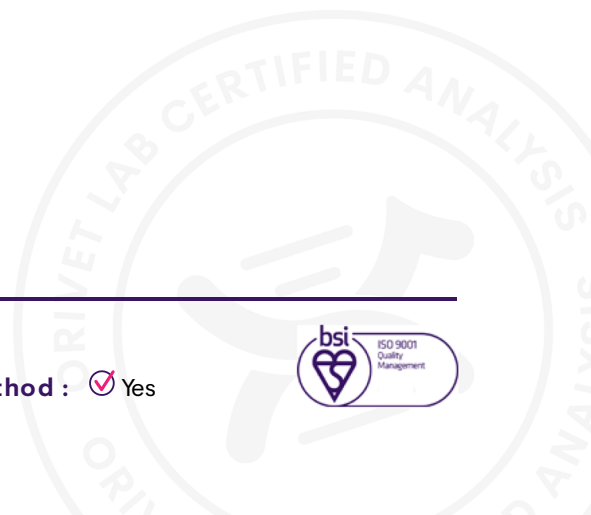
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

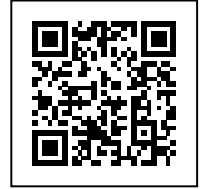
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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## Genetic Comprehensive Report

Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** OSTEOCHONDRODYSPLASIA (MIN POODLE TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** SLC13A1

**Variant Detected :** g.63600045\_63729942del129897bp

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.

**Test Reported :** OSTEOGENESIS IMPERFECTA (GOLDEN RETRIEVER TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Collagen type I alpha 1 chain (COL1A1) Chromosome 9

**Variant Detected :** Base Substitutionc.1276G>Cp.Gly381Ala

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.

**Test Reported :** PROGRESSIVE ROD CONE DEGENERATION (PRCD) - PRA

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Photoreceptor disc component (PRCD) on Chromosome 9

**Variant Detected :** Base Substitutionc.5 G>Ap.Cys2Tyr

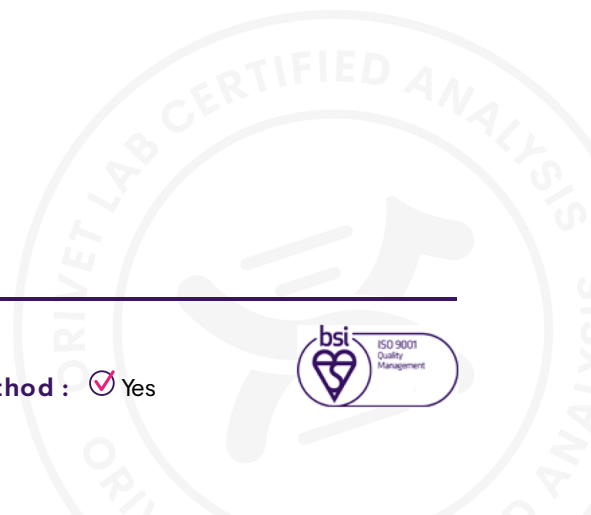
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

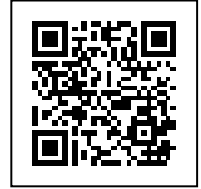
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** PYRUVATE KINASE DEFICIENCY (LABRADOR TYPE)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** PKLR

**Variant Detected :** c.799C>T

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.

**Test Reported :** RCD4-PRA (LATE ONSET)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** C2orf71 on Chromosome 17

**Variant Detected :** c.3149\_3150insCp.Cys1051ValfsX90

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.

**Test Reported :** SKELETAL DYSPLASIA 2 (MILD DISPROPORTIONATE DWARFISM)

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** Collagen alpha-2(XI) chain gene (COL11A2) on chromosome 12

**Variant Detected :** Base Substitutionc.143G>Cp.Arg48Pro

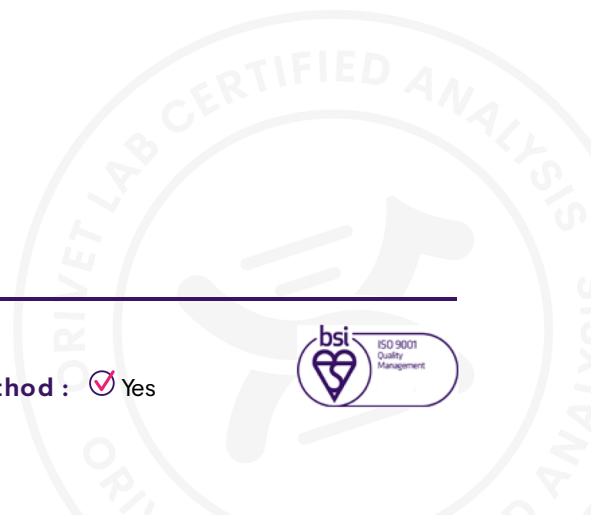
We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** VON WILLEBRAND'S DISEASE TYPE I

**Result :** **NEGATIVE / CLEAR [NO VARIANT DETECTED]**<sup>1</sup>

**Gene :** VWF

**Variant Detected :** c.7437G>A

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.

**Test Reported :** E LOCUS - (CREAM/RED/YELLOW)

**Result :** **e/e - HOMOZYGOUS FOR NON-EXTENSION [WHITE/YELLOW/APRICOT/WHEATEN]**<sup>1</sup>

**Gene :** MC1R

**Variant Detected :** Em (point mutation) > E (wild type) > e (point mutation) chr5:63694334-63694334: C>T

2 copies of red/yellow are present referred to as "non-extension". Dog's coat is entirely phaeomelanin based ie. red/yellow/cream/apricot/white/wheaten. Please note in some breeds an "ee" phenotype can often Colours can be cream to white rather than yellow to red. Shades can vary between littermates.

**Test Reported :** I LOCUS COLOUR INTENSITY

**Result :** **I/I - NO COPY OF MFSD12 INTENSITY ALLELE (NOT LIKELY TO SHOW EXTREME DILUTION)**<sup>1</sup>

**Gene :** MFSD12

**Variant Detected :** c.151C>T (p.Arq51Cys)

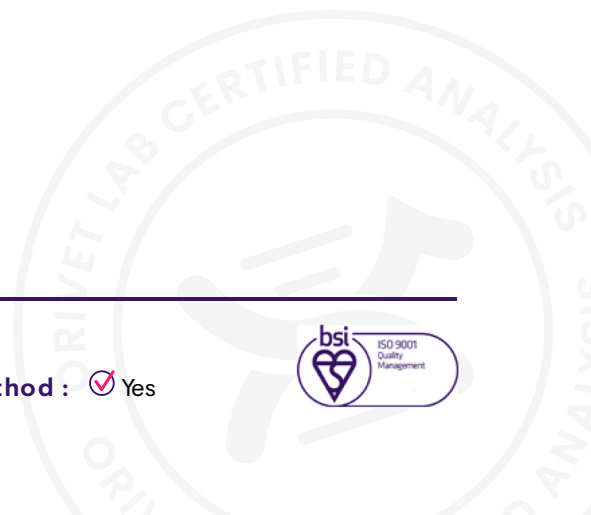
This variant is associated with the dilution of phaeomelanin which is involved in the cream/white/apricot color in dogs. Degree of intensity (dilution) will vary within and between breeds.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** BROWN (345DELPRO) DELETION

**Result :** B<sup>d</sup>/B<sup>d</sup> - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [DELETION]<sup>1</sup>

**Gene :** TYRP1

**Variant Detected :** Base Substitution (Point Mutation)

Does not carry the brown deletion codon. Please refer to the other brown variants to clarify potential colour for offspring.

**Test Reported :** BROWN (GLNT331STOP) STOP CODON

**Result :** B<sup>s</sup>/B<sup>s</sup> - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [STOP CODON]<sup>1</sup>

**Gene :** TYRP1

**Variant Detected :** Point Mutation

Does not carry the brown stop codon. Please refer to the other brown variants to clarify potential colour for offspring.

**Test Reported :** BROWN (SER41CYS) INSERTION CODON

**Result :** B<sup>c</sup>/B<sup>c</sup> - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [INSERTION]<sup>1</sup>

**Gene :** TYRP1

**Variant Detected :** Base Substitution (Point Mutation)

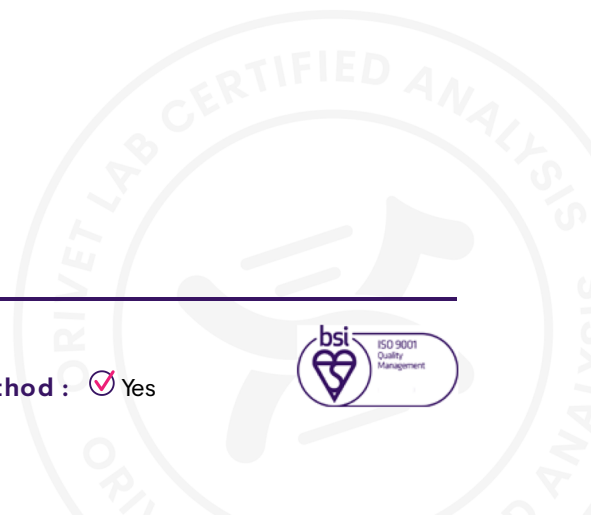
Does not carry the brown insertion codon. Please refer to the other brown variants to clarify potential colour for offspring.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes







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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** LIVER [TYRP1] (LANCASHIRE HEELER TYPE)

**Result :** B<sup>e</sup>/B<sup>e</sup> - DOES NOT CARRY BROWN/LIVER [TYRP1]<sup>1</sup>

**Gene :**

**Variant Detected :**

**Test Reported :** D (DILUTE) LOCUS

**Result :** D/D - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL<sup>1</sup>

**Gene :** MLPH

**Variant Detected :** Base Substitution

Full colour, no dilute gene present. The D allele modifies the Melanophilin (MLPH) gene. This animal cannot produce "dilute" offspring. Please Note: There are other dilute variants d2 (Sloughi, Chow Chow & Thai Ridgeback) and rare d3 (Italian Greyhound & Chihuahua) so this test/result may not identify dilute in these breeds.

**Test Reported :** DILUTE D2 VARIANT (CHOW CHOW TYPE)

**Result :** d<sup>2</sup>/d<sup>2</sup> - NO COPY OF d2 ALLELE (DILUTE) - PIGMENT IS NORMAL<sup>1</sup>

**Gene :** MLPH

**Variant Detected :** c.705G>C

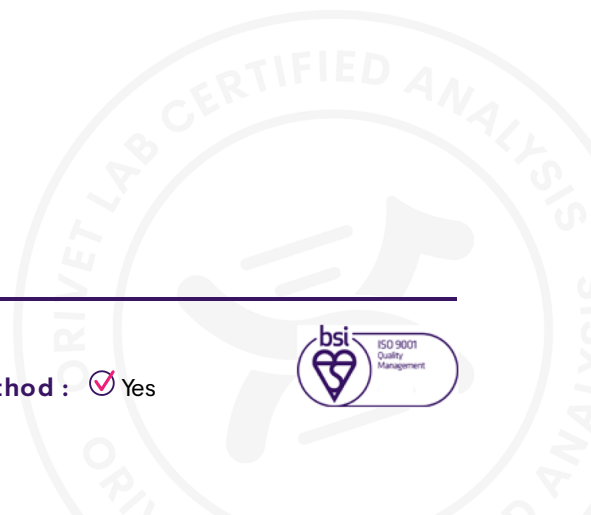
This d2 variant has been shown to be associated with the blue/dilute seen in the Chow Chow, Sloughi, Thai Ridgeback and any mixes of these breeds.

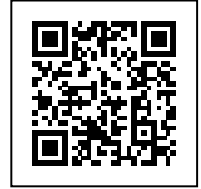
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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## Genetic Comprehensive Report

Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

### Test Reported : K LOCUS (DOMINANT BLACK)

**Result : K/K - DOMINANT BLACK - SOLID [WILL NOT BE BRINDLED or EXPRESS AGOUTI]<sup>1</sup>**

**Gene : CBD103**

**Variant Detected :** Deletion of GGG

Two copies of dominant black (K) are present. No brindle/red or fawn offspring will be produced. Will not express Agouti phenotype. This can also be referred to as KB. In some breeds the K locus is fixed so all dogs will be KK. This (the K Locus) can be modified by other genes eg. liver, dilute, greying or merle. Red can only be added through the e locus.

### Test Reported : A LOCUS (FAWN/SABLE; TRI/TAN POINTS)

**Result : a<sup>y</sup>/a<sup>t</sup> - FAWN/RED/SABLE CARRIES TRICOLOUR/TAN POINTS<sup>1</sup>**

**Gene : ASIP**

**Variant Detected :** Base Substitution 246 G>T(A82S); G>A (R83H): C>T (p.R96C)

Dog has fawn/sable and carries black and tan (hidden colour tri or tan points). Tri factored (Sable & White). Also referred to as "sabled red". Produces fawn or sable coat and the majority of the coat is red/yellow with some black usually intermingled within the coat. Coat colour shown is dependent on the E, K and B Locus. the ay allele is dominant over at.

### Test Reported : MERLE

**Result : m [171bp] / m [171bp] - NON MERLE SOLID COAT (NO CHANGE TO COAT or EYE COLOUR)<sup>1</sup>**

**Gene : SILV**

**Variant Detected :**

250 base pair SINE insertion, oligo(dA)-rich tails with length polymorphism. Detects and reports all the 7 alleles on the M Locus (Mh, M, Ma+, Ma, Mc+, Mc and m)

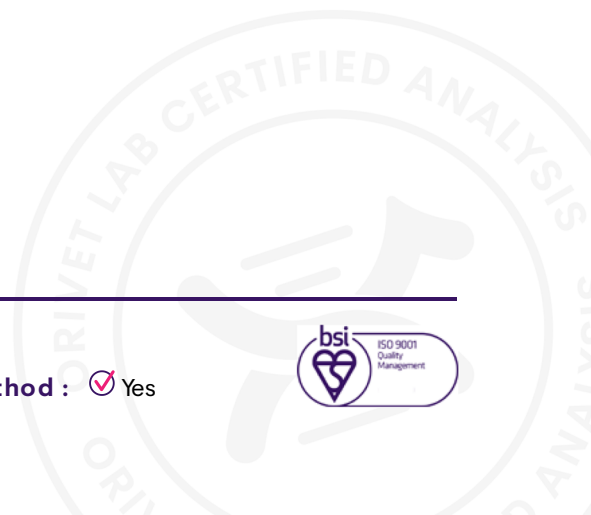
There are many factors that may influence a Merle result, these include mosaicism (merle expressed in different cell types) or the amount of circulating merle copies within the sample type. If this result does not match your phenotype please contact Orivet to request retest or re-analysis of the sample.

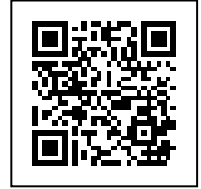
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** SPOTTING (W) LOCUS (MASTIFF TYPE)\*

**Result :** **NEGATIVE - NOT SHOWING THE PHENOTYPE**<sup>1</sup>

**Gene :**

**Variant Detected :**

**Test Reported :** LONG HAIR GENE (CANINE C95F)

**Result :** **POSITIVE - SHOWING THE PHENOTYPE**<sup>1</sup>

**Gene :** FGF5

**Variant Detected :** p.Cys95Phe c284G>T (Point Mutation)

The phenotype/trait tested is present. Please Note this can vary from breed to breed and within breed.

**Test Reported :** SHEDDING (MC5R)

**Result :**

**SHD/SHD [LOW SHEDDING] - NO COPIES OF THE SHEDDING (MC5R) VARIANT DETECTED [REFER TO R151W (IC) FOR LEVEL]**

<sup>1</sup>

**Gene :** MC5R

**Variant Detected :**

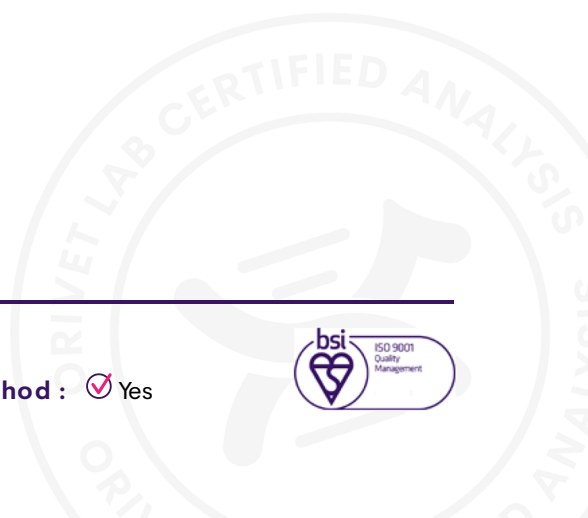
The dog will (may) exhibit low or no levels of shedding. Please Note: this level is also dependent on the furnishing allele. If the dog has no IC (R151W) phenotype will be low shedding.

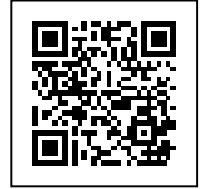
**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes





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Sample with Lab ID Number 22G03771 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** COAT COMPOSITION CFA28 GENE (DOUBLE/SINGLE COAT)

**Result :** UDC/udc - ONE COPY OF THE DOUBLE COAT (DENSE UNDERCOAT) PHENOTYPE DETECTED<sup>1</sup>

**Gene :** CFA28

**Variant Detected :**

Moderate to Low Shedding please refer to IC result to clarify level of shedding

**Test Reported :** CURLY COAT/HAIR CURL (KRT71 R151W)

**Result :**

**ONE COPY OF THE KRT71 R151W (CU/Cu) VARIANT DETECTED - MOST LIKELY TO HAVE MODERATE 'WAVY' CURLY COAT PHENOTYPE**

<sup>1</sup>

**Gene :** KRT71 (R151W)

**Variant Detected :** chr27:2539211-2539211: c.451C>T

Please note there are other additional curly coat genes/variant that will impact the curly coat phenotype.

**Test Reported :** IMPROPER COAT (RSPO2)

**Result :** IC2/ic2 - CARRIES ONE COPY THE IMPROPER COAT RSPO2 (DELETION) VARIANT DETECTED<sup>1</sup>

**Gene :** RSPO2

**Variant Detected :** 167 bp insertion in 3'UTR region

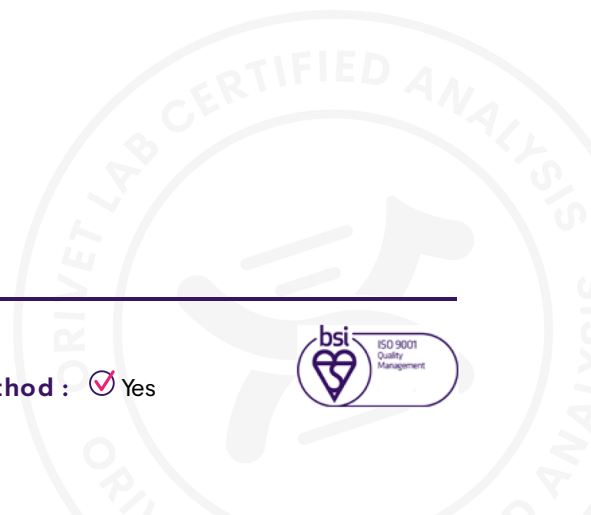
Please Note: This is one of the 3 IC variants that are associated with IC. There may be other causes of this condition in dogs and a carrier result does not exclude a different mutation in this gene or any other gene that may result in a similar genetic trait.

**Owner's Name :** Amelie Martin

**Pet Name :** Friska

**Microchip Number** 96700000056327

**Approved Collection Method :**  Yes



# 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**

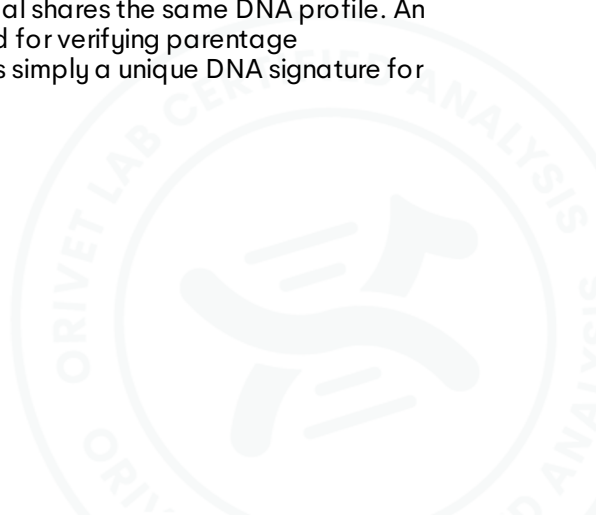
Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss.

## **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](mailto:admin@orivet.com) and we will be happy to work with you to answer any relevant questions.

