



# Orivet

# Genetic Comprehensive Report

**Animal Name:** RAI 03-22

**Owner:**

**Peter Howie**

**Membership Number : Not assigned**

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

**Approved Collection Method: No**



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Harmonization of Genetic Testing for Dogs

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## Genetic Comprehensive Report

### Owner's details

Name: Peter Howie

### Animal's Details

Registered Name :

Pet Name : RAI 03-22

Registration Number :

Breed : Standard Poodle

Microchip Number : 953010006019275

Sex : Intact Male

Date of Birth : 19th Oct 2022

Colour : BLUE MERLE

### Sample Collection Details

Case Number : 23A128273

Collected By :

Approved Collection : No

Sample Type : SWAB

### Test Details

Test Requested : Groodle - Full Breed Profile

Pet Name : RAI 03-22

Date of Test : 19th Jan 2023

### Authorisation

Sample with Lab ID Number 23A128273 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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# Genetic Comprehensive Report

## Animal's Details

Registered Name :	Labracadabra
Pet Name :	RAI 03-22
Registration Number :	
Breed :	Standard Poodle
Microchip Number :	953010006019275
Sex :	Intact Male
Date of Birth :	19th Oct 2022
Colour :	BLUE MERLE

P1\_2 G G P3\_2 A A P3\_3 G G P11\_3 C C P12\_1 G G P24\_2 A G P12\_3 G G P30\_3 A T  
 P13\_1 C C P24\_3 C C P31\_1 A A P28\_3 A T P31\_3 G G P25\_1 G G P32\_2 C G P13\_2 A A  
 P13\_3 A C P25\_2 A G P25\_3 A C P32\_3 G G P33\_1 G G P14\_1 A T P10\_1 G G P26\_1 A G  
 P33\_3 A G P26\_2 A A P14\_2 G G P26\_3 G G P14\_3 A C P15\_1 A G P34\_1 A C P34\_2 G G  
 P34\_3 A A P10\_3 C C P15\_2 G G P15\_3 C C P16\_3 C G P35\_1 G G P35\_2 A G P36\_1 A C  
 P17\_1 A A P36\_2 C C P37\_2 A G P17\_2 A A P29\_1 G G P37\_3 G G P38\_1 C C P38\_2 G G  
 P27\_1 G G P17\_3 A A P27\_2 A A P4\_3 A A P18\_2 A C P18\_3 A C P5\_1 G G P11\_1 G G  
 P19\_1 A T P19\_2 G G P5\_2 G G P19\_3 G G P2\_1 G G P2\_3 A A P27\_3 A A P20\_1 A A  
 P20\_3 A A P5\_3 A A P11\_2 C G P6\_2 A G P6\_3 C C P21\_1 A G P21\_3 A A P22\_2 A C  
 P28\_1 G G P7\_1 C C P7\_2 A G P28\_2 G G P7\_3 A G P29\_2 G G P8\_1 A G P22\_3 G G  
 P8\_2 G G P8\_3 G G P23\_1 C G P9\_3 A T P23\_2 C C P23\_3 G G P24\_1 G G P3\_1 A G

Owner's Name : Peter Howie

Pet Name : RAI 03-22

Microchip Number 953010006019275

Approved Collection Method : No



# Genetic Comprehensive Report

## Animal's Details

Registered Name :	
Pet Name :	RAI 03-22
Registration Number :	
Breed :	Standard Poodle
Microchip Number :	953010006019275
Sex :	Intact Male
Date of Birth :	19th Oct 2022
Colour :	BLUE MERLE

BICF2G630103624	C C	BICF2G630111735	A G	BICF2G630122583	A G	BICF2G630133028	G G
BICF2G630133994	A G	BICF2G630149030	A G	BICF2G630200354	A A	BICF2G630209886	A A
BICF2G630220326	G G	BICF2G630221287	A G	BICF2G630264994	A A	BICF2G630276039	A G
BICF2G630276136	G G	BICF2G630306265	G G	BICF2G630326688	A G	BICF2G630328172	A A
BICF2G630328323	A G	BICF2G630367177	A C	BICF2G630409193	G G	BICF2G630453264	C G
BICF2G630474528	G G	BICF2G630499189	A G	BICF2G630539759	A G	BICF2G630552597	G G
BICF2G630653298	A A	BICF2G630666362	A G	BICF2G630691635	C G	BICF2G630704611	G G
BICF2G630708384	G G	BICF2G630762459	C C	BICF2G63078341	A A	BICF2G630881115	A A
BICF2P1010945	G G	BICF2P105070	A G	BICF2P1138733	A G	BICF2P1159837	G G
BICF2P1181787	A G	BICF2P1192522	A A	BICF2P1226745	G G	BICF2P1286728	G G
BICF2P1362405	G G	BICF2P1369088	A G	BICF2P1391407	A A	BICF2P164304	G G
BICF2P184963	A A	BICF2P251850	A C	BICF2P277987	A G	BICF2P345488	A G
BICF2P401677	G G	BICF2P414351	G G	BICF2P42825	A G	BICF2P452541	A G
BICF2P457665	A G	BICF2P464536	A G	BICF2P465276	A A	BICF2P466604	A A
BICF2P46672	G G	BICF2P496466	A A	BICF2P496837	A G	BICF2P567552	G G
BICF2P590440	G G	BICF2P600196	A A	BICF2P615597	A A	BICF2P635478	A A
BICF2P651575	G G	BICF2P651577	A A	BICF2P70891	C C	BICF2P725743	C G
BICF2P728698	G G	BICF2P789367	A A	BICF2P805553	G G	BICF2P840653	A G
BICF2P885380	G G	BICF2P923421	G G	BICF2P950116	A G	BICF2P963969	A G
BICF2P998036	A C	BICF2S22912385	A G	BICF2S22926284	A A	BICF2S22953709	A A
BICF2S23018785	G G	BICF2S23111132	G G	BICF2S23138418	G G	BICF2S23141330	A T
BICF2S23214514	C C	BICF2S23326150	A G	BICF2S23329382	C C	BICF2S23357186	C G
BICF2S2338108	A G	BICF2S23434277	C G	BICF2S23529290	G G	BICF2S23535154	A G
BICF2S23614068	A C	BICF2S2399705	G G	G1425f16S28	A G	TIGRP2P255960_rs9030578	A G
TIGRP2P283310_rs8881748	A G	TIGRP2P328303_rs8531882	A A	TIGRP2P354499_rs9162547	A G	TIGRP2P356245_rs8830240	A C
TIGRP2P362535_rs9130694	A G	TIGRP2P389035_rs9038546	A A				

Owner's Name : Peter Howie

Pet Name : RAI 03-22

Microchip Number 953010006019275

Approved Collection Method : No



# Genetic Comprehensive Report

## Animal's Details

Registered Name :	
Pet Name :	RAI 03-22
Registration Number :	
Breed :	Standard Poodle
Microchip Number :	953010006019275
Sex :	Intact Male
Date of Birth :	19th Oct 2022
Colour :	BLUE MERLE

BICF2G630102146	A G	BICF2G630149581	A G	BICF2G630159183	A G	BICF2G630170631	A C
BICF2G630187649	S T	BICF2G630187658	A A	BICF2G630204463	A A	BICF2G630209373	A G
BICF2G630209508	A A	BICF2G630255439	A G	BICF2G630271966	A G	BICF2G630274628	A A
BICF2G630307199	A C	BICF2G630340940	G G	BICF2G630340944	G G	BICF2G630365778	A A
BICF2G630382763	G G	BICF2G630437783	C C	BICF2G630449851	A G	BICF2G630467607	C C
BICF2G630488267	A A	BICF2G630504410	G G	BICF2G630552598	G G	BICF2G630558437	A A
BICF2G630594648	A A	BICF2G630634836	C C	BICF2G630641678	G G	BICF2G630689403	A G
BICF2G630798972	G G	BICF2G630814422	C C	BICF2G630900019	A T	BICF2P1019402	G G
BICF2P103615	A G	BICF2P1060087	A A	BICF2P1104630	A A	BICF2P1141966	A G
BICF2P1173491	A A	BICF2P1183665	A A	BICF2P1193353	A G	BICF2P1216677	A A
BICF2P1226838	A G	BICF2P1232055	A G	BICF2P1271174	G G	BICF2P129347	A G
BICF2P129670	A G	BICF2P1308802	A C	BICF2P1310805	A A	BICF2P1344095	A G
BICF2P1346673	A G	BICF2P1357746	A G	BICF2P1454500	A G	BICF2P155421	A C
BICF2P157421	A A	BICF2P182473	G G	BICF2P224656	A C	BICF2P237994	A A
BICF2P246592	A C	BICF2P250787	C C	BICF2P25730	A A	BICF2P283440	A A
BICF2P285489	G G	BICF2P345056	A A	BICF2P347679	G G	BICF2P378969	A A
BICF2P382742	A A	BICF2P415783	G G	BICF2P422152	A G	BICF2P508740	C G
BICF2P516667	G G	BICF2P553317	A G	BICF2P554817	A A	BICF2P561057	A A
BICF2P585943	G G	BICF2P624936	A G	BICF2P635172	G G	BICF2P643134	G G
BICF2P65087	A G	BICF2P651576	A A	BICF2P717226	A C	BICF2P751654	A A
BICF2P774003	C C	BICF2P798404	A A	BICF2P842510	A G	BICF2P856893	A G
BICF2P878175	A G	BICF2P935470	A A	BICF2P990814	A G	BICF2S22910736	A G
BICF2S22913753	A G	BICF2S22928800	G G	BICF2S22943825	A A	BICF2S23028732	A T
BICF2S23031254	A C	BICF2S23049416	G G	BICF2S23057560	G G	BICF2S23126079	A A
BICF2S23246455	A A	BICF2S23250041	A C	BICF2S23333411	G G	BICF2S23356653	G G
BICF2S23429022	G G	BICF2S23449478	A A	BICF2S23519644	A G	BICF2S2351979	G G
BICF2S2359809	G G	BICF2S236196	A A	BICF2S23626625	C C	BICF2S23648905	G G
BICF2S23124313	A G	BICF2S23649947	A G	BICF2S23713161	A G	BICF2S23737033	A G
BICF2S24511913	A G	TIGRP2P106843_rs8858816	A G	TIGRP2P116826_rs8741680	A G	TIGRP2P164720_rs8839809	A G
TIGRP2P177606_rs8886563	G G	TIGRP2P215708_rs8686029	A A	TIGRP2P316532_rs8597522	A G	TIGRP2P372104_rs9153277	G G
TIGRP2P402042_rs9121006	G G	TIGRP2P406551_rs9235397	A G	TIGRP2P407751_rs8803124	A A	BICF2G630646431	A A

Owner's Name : Peter Howie

Pet Name : RAI 03-22

Microchip Number 953010006019275

Approved Collection Method : No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**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 :** **CLEAR BY PARENTAGE - DNA VERIFIED [PARENTS NORMAL/CLEAR FOR DISEASE]**<sup>1</sup>

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

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

The sample submitted has had its parentage verified by DNA. By interrogating the DNA profiles of the Dam, Sire and Offspring, parentage has been confirmed and this information together with the disease history submitted for the parents excludes this animal from having the disease. The controls run confirm that the dog is NORMAL/CLEAR for the disease requested.

**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 :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 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>TTThr2110Met

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 :** GANGLIOSIDOSIS GM2 (POODLE TYPE)

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

**Gene :** Hexosaminidase subunit beta (HEXB) on Chromosome 2

**Variant Detected :** Nucleotide Deletionc.391delGp.Val95fsX

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 :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 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 :** 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 Deletion c.1445-1447delACC and c.1447insTACTACTAp.Asn482llefs9X

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 :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No







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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**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 :** 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 :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 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 :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**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 :** 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 - DOMINANT BLACK DOES NOT CARRY YELLOW/RED/WHITE**<sup>1</sup>

**Gene :** MC1R

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

2 copies of black E or "extension". All areas of the coat colour eumelanin will not produce any "e" offspring.

The Extension loci is responsible for the majority of non-agouti patterns.

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** EM (MC1R) LOCUS - MELANISTIC MASK

**Result :**  $E^n/E^n$  - NO MELANISTIC MASK ( $E^n$ ) EXTENSION ALLELE<sup>1</sup>

**Gene :** MC1R

**Variant Detected :** Base Substitution G>A

Dog tested negative for the melanistic mask allele. The dog will not have a black mask, and cannot pass a copy on to any offspring..

**Test Reported :** I LOCUS COLOUR INTENSITY

**Result :** i/i - TWO COPIES OF THE MFSD12 INTENSITY ALLELE (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 pheomelanin which is involved in the cream/white/apricot color in dogs. Degree of intensity (dilution) will vary within and between breeds.

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

**Result :**  $B^d/B^d$  - 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.

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**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>o</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.

**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 :**

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**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 Melanophillin (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 :** K LOCUS (DOMINANT BLACK)

**Result :**

**KB / k<sup>y</sup> or k<sup>br</sup> - ONE COPY DOMINANT BLACK (KB) and ONE COPY OF NON-BLACK (k<sup>y</sup>) dog MAY be brindled**

<sup>1</sup>

**Gene :** CBD103

**Variant Detected :** Deletion of GGG

One copy of non black and one copy of ky or kbr is present. This KB will cover the A locus and all you will visualise is the base colour. Dog will express the alleles on the A locus but any and all pheomelanin (red) in the coat will be brindled. This allele overrides the ASIP (A) locus. The agouti phenotype may be altered for some breeds and therefore be brindle. There are three alleles at the K Locus with the following dominance hierarchy KB > Kbr > k. The first KB represents dominant black, the second allele Kbr represents brindling and may display A locus gene. Brindle in most breeds appears as black stripes on a red base. Please Note: At this stage no commercial genetic testing can distinguish brindle so breeders should rely on their pedigree or breed standard to exclude or include brindle phenotype.

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

**Result :** **a<sup>t</sup>/a<sup>t</sup> - TAN POINTS/BLACK & TAN or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]**<sup>1</sup>

**Gene :** ASIP

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

Homozygous for black and tan/tricolour (no hidden colours) allele. Tri factored/white factored in dogs that have white points. No Bi Factoring (Black White & Tan). Animals are primarily black and have areas of pheomelanin (tan) which tends to be seen on the leg and stomach areas, the side of the head and spots above the eyes. Please note the colour and distribution of pheomelanin "tan" will be dependent on the breed and other colour genes. Please note that any genes on the "A" series will only be expressed if the K locus is kk, k<sup>br</sup> or k<sup>br</sup>k<sup>br</sup>.

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** PIED (BOTH SINE AND REPEAT VARIANTS)

**Result :** S/sp - CARRIER OF PIEBALD [LIMITED WHITE SPOTTING, FLASH OR PARTI]<sup>1</sup>

**Gene :** MITF-M on Chromosome 20

**Variant Detected :**

g.chr20:21836563insSINELength polymorphism (repeat CAGA) chr20:21839332-21839366 MITF-M

Carries a single copy of the Melanocyte Inducing Transcription Factor (MITF) "sp" allele. In some breeds the dog may have limited random coat colour deletion, this can vary from a few white hairs up to half white. For some breeds pied is any amount of white on the dog at all, for others it is a dog that is predominantly white. The dog may pass on the "sp" allele to offspring. If no other white-causing genes are at play (such as Irish white head, pseudo irish, etc.) then most will end up with white chest/toes or less white. Some S/sp appear phenotypically solid in color. It has also been shown that sp/sp does not present as piebald in many wolves and nordic dog breeds

**Test Reported :** MERLE

**Result :** m/Mc/M [171/221/267bp] - MOSAIC RESULT [MUTED MERLE]<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)

Mosaicism - "Somatic Mosaicism" or "Somatic Mutation" - is the presence of two or more types of cells with different genotypes present in the body of one individual dog.

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

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

**Gene :**

**Variant Detected :**

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No





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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** SHEDDING (MC5R)

**Result :**

**SHD/shd [MODERATE SHEDDING] - ONE COPY OF THE SHD (MC5R) VARIANT DETECTED [REFER TO R151W (IC) FOR LEVEL]**

<sup>1</sup>

**Gene :** MC5R

**Variant Detected :**

The dog will (may) exhibit a moderate (average) level 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.

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

**Result :** UDC/UDC - NO COPY OF THE DOUBLE COAT (DENSE UNDERCOAT) PHENOTYPE DETECTED<sup>1</sup>

**Gene :** CFA28

**Variant Detected :**

Dog has a single coat usually associated with no undercoat. Hair length can be short or long.

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

**Result :**

**POSITIVE FOR THE KRT71 R151W (Cu/Cu) VARIANT - LIKELY TO HAVE CURLY (TIGHT) HAIR 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.

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No







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## Genetic Comprehensive Report

Sample with Lab ID Number 23A128273 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

**Test Reported :** CURLY COAT PHENOTYPE(KRT71 p.SER422ARGFSTER)

**Result :**

**NEGATIVE FOR THE KRT71 (p.Ser422ArgfsTer) VARIANT - NOT SHOWING THE CURLY COAT (C2) PHENOTYPE**

1

**Gene :** KRT71

**Variant Detected :** c.1266\_1273delCCTGAAGCinsACA p. Ser422ArgfsTer

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

**Result :** IC2/IC2 - NO 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 normal result does not exclude a different mutation in this gene or any other gene that may result in a similar genetic trait.

**Test Reported :** BODY SIZE IGSF1 "BULKY GENE"

**Result :**

**HOMOZYGOUS FOR INSULIN LIKE GROWTH FACTOR (IGF1R) - ASSOCIATED WITH AN INCREASE OF BODY (BULKY) SIZE**

1

**Gene :** IGSF1

**Variant Detected :** chrX.g.102369488-102369489insAAC, p. Asp376\_Glu377insAsn, Chromosome X

Please Note: This variant is found on the X-Chromosome and will present as 2 copies in females and one copy in males. The IGF1R allele in an ancestral allele found in larger-sized breeds.

**Owner's Name :** Peter Howie

**Pet Name :** RAI 03-22

**Microchip Number** 953010006019275

**Approved Collection Method :** No



# Glossary of Genetic Terms (Results)

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## **NEGATIVE / CLEAR [NO VARIANT DETECTED]**

No presence of the variant (mutation) has been detected. The animal is clear of the disease and will not pass on any disease-causing mutation.

## **CARRIER [ONE COPY OF THE VARIANT DETECTED]**

This is also referred to as HETEROZYGOUS. One copy of the normal gene and copy of the affected (mutant) gene has been detected. The animal will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal - if breeding with another carrier or affected or unknown then it may produce an affected offspring.

## **POSITIVE / AT RISK [TWO COPIES OF THE VARIANT DETECTED]**

Two copies of the disease gene variant (mutation) have been detected also referred to as HOMOZYGOUS for the variant. The animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a Veterinarian.

## **POSITIVE HETEROZYGOUS [ONE COPY OF THE DOMINANT VARIANT DETECTED]**

Also referred to as POSITIVE ONE COPY or POSITIVE HETEROZYGOUS. This result is associated with a disease that has a dominant mode of inheritance. One copy of the normal gene (wild type) and affected (mutant) gene is present. Appropriate treatment should be pursued by consulting a Veterinarian. This result can still be used to produce a clear offspring.

## **NORMAL BY PARENTAGE HISTORY**

The sample submitted has had its parentage verified by DNA. By interrogating the DNA profiles of the Dam, Sire and Offspring this information together with the history submitted for the parents excludes this animal from having this disease. The controls run confirm that the dog is NORMAL for the disease requested.

## **NORMAL BY PEDIGREE**

The sample submitted has had its parentage verified by Pedigree. The pedigree has been provided and details (genetic testing reports) of the parents have been included. Parentage could not be determined via DNA profile as no sample was submitted.

## **NO RESULTS AVAILABLE**

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

## **INDETERMINABLE**

The sample submitted has failed to give a conclusive result. This result is mainly due to the sample failing to "cluster" or result in the current grouping. A recollection is required at no charge.

## **DNA PROFILE**

Also known as a DNA fingerprint. This is unique for the animal. No animal shares the same DNA profile. An individual's DNA profile is inherited from both parents and can be used for verifying parentage (pedigrees). This profile contains no disease or trait information and is simply a unique DNA signature for that animal.

# Glossary of Genetic Terms (Results)

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## **PARENTAGE VERIFICATION/ QUALIFIES/CONFIRMED OR DOES NOT QUALIFY/EXCLUDED**

Parentage is determined by examining the markers on the DNA profile. A result is generated and stated for all DNA parentage requests. Parentage confirmation reports can only be generated if a DNA profile has been carried out for Dam, Offspring and possible Sire/s.

## **PENDING**

PENDING

## **TRAIT (PHENOTYPE)**

A feature that an animal is born with (a genetically determined characteristic). Traits are a visual phenotype that range from colour to hair length, and also includes certain features such as tail length. If an individual is **AFFECTED** for a trait then it will show that characteristic eg. **AFFECTED** for the B (Brown) Locus or bb will be brown/chocolate.

## **POSITIVE – SHOWING THE PHENOTYPE**

The animal is showing the trait or phenotype tested.

## **CLARIFICATION OF GENETIC TESTING**

The goal of genetic testing is to provide breeders with relevant information to improve breeding practices in the interest of animal health. However, genetic inheritance is not a simple process, and may be complicated by several factors. Below is some information to help clarify these factors.

The goal of genetic testing is to provide breeders with relevant information to improve breeding practices in the interest of animal health. However, genetic inheritance is not a simple process, and may be complicated by several factors. Below is some information to help clarify these factors.

1) Some diseases may demonstrate signs of what Geneticists call "genetic heterogeneity". This is a term to describe an apparently single condition that may be caused by more than one mutation and/or gene

2) It is possible that there exists more than one disease that presents in a similar fashion and segregates in a single breed. These conditions -although phenotypically similar - may be caused by separate mutations and/or genes.

3) It is possible that the disease affecting your breed may be what Geneticists call an "oligogenic disease". This is a term to describe the existence of additional genes that may modify the action of a dominant gene associated with a disease. These modifier genes may for example give rise to a variable age of onset for a particular condition, or affect the penetrance of a particular mutation such that some animals may never develop the condition.

The range of hereditary diseases continues to increase and we see some that are relatively benign and others that can cause severe and/or fatal disease. Diagnosis of any disease should be based on pedigree history, clinical signs, history (incidence) of the disease and the specific genetic test for the disease. Penetrance of a disease will always vary not only from breed to breed but within a breed, and will vary with different diseases. Factors that influence penetrance are genetics, nutrition and environment. Although genetic testing should be a priority for breeders, we strongly recommend that temperament and phenotype also be considered when breeding.

Orivet Genetic Pet Care aims to frequently update breeders with the latest research from the scientific literature. If breeders have any questions regarding a particular condition, please contact us on (03) 9534 1544 or admin@orivet.com and we will be happy to work with you to answer any relevant questions.