

**Animal Name: Merlin** 

**Owner:** 

**Amanda Broderick** 

Membership Number: Not assigned

Member Body/Breed Club: Not assigned

**Approved Collection Method: No** 



















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### Owner's details

**Amanda Broderick** Name: **Animal's Details** Registered Name: First's Flight Of Merlin The White Pet Name: Merlin Registration Number: SS23269704 Breed: Golden Retriever Microchip Number: 985113004247345 **Neutered Male** Sex: Date of Birth: 30th Dec 2020

### Sample Collection Details

Case Number: 24A106350

Collected By:

Approved Collection: No

White Gold

**SWAB** 

**Test Details** 

Sample Type:

Colour:

Test Requested : Golden Retriever - Full Breed Profile

Pet Name : Merlin

Date of Test : 11th Jul 2024

### **Authorisation**

Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

(huel \_\_\_\_

George Sofronidis BSc (Hons)

N. M.

bsi ISO 9001 Quality Management

Dr Noam Pik BVSc, MAVS





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### **Animal's Details**

Registered Name :	First's Flight Of Merlin The White
Pet Name :	Merlin
Registration Number :	SS23269704
Breed :	Golden Retriever
Microchip Number:	985113004247345
Sex:	Neutered Male
Date of Birth :	30th Dec 2020
Colour:	White Gold

### **ISAG Profile 1**

Cfam_1:106430955	АА	Cfam_1:119414584	АА	Cfam_1:20842130		Cfam_1:3962719	A G	Cfam_1:70238933	АА	Cfam_1:80971770	
(BICF2S23111132) Cfam_10:10652659	A G	(BICF2P157421) Cfam_10:22409408		Cfam_10:30034450		(BICF2G630708384) Cfam_10:66922269	A G	(BICF2P554817) Cfam_11:23907101	A C	Cfam_11:5318488	A G
(BICF2P237994) Cfam_11:65603333		Cfam_12:35306641		Cfam_12:55201839	GG	(BICF2S23049416) Cfam_12:5579055	АА	(BICF2P1308802) Cfam_12:68125319	АА	(BICF2S2338108) Cfam_13:59896033	A C
Cfam 13:8704192	GG	Cfam 14:50063321	ΑА	(BICF2G630122583) Cfam_14:58465266	A G	(BICF2P382742) Cfam 15:19299365	ΑА	(BICF2P1344095) Cfam 15:22834903	СС	(BICF2P561057) Cfam 16:29634940	GG
(BICF2P182473)		(BICF2P624936) Cfam 16:57958947		(P24_2) Cfam 17:10649078	АА	(BICF2P105070) Cfam 17:34462308	A G	(BICF2G630437783)		(BICF2G630111735)	GG
(BICF2P774003)		(BICF2P635478) Cfam 19:15926130		(BICF2G630220326) Cfam 19:27288167	ΔΔ	(BICF2G630209373) Cfam 19:47470564	A C	(BICF2P998036)		(B1CF2G630689403) Cfam_2:2610859	GG
(BICF2S23535154)		(P13_3) Cfam 2:77806065		(BICF2P251850) Cfam 20:13740894		(BICF2S23214514) Cfam_20:49900586		(BICF2S23737033) Cfam_20:57167714		(P32_3) Cfam_21:15558670	A A
(BICF2P1159837)		(BICF2P878175) Cfam 21:35719434		(BICF2S23246455) Cfam 22:26694580		(BICF2P347679) Cfam 22:55308193		(P26_1) Cfam_22:641125		(B1CF2G630653298) Cfam_23:42886681	AC
(BICF2S23018785)		(BICF2S23326150) Cfam_24:23393510		(BICF2G630326688) Cfam 24:29909901		(BICF2S23329382) Cfam 24:47381908		Cfam 25:2073511	A C	(P34_1)	GG
(BICF2P277987)		Cfam 26:20004896	ΔА	(TI GRP2P316532_rs8597522) Cfam 26:35071515	A G	(BICF2P990814) Cfam 27:22599860	GG	(P15_3)		(BICF2G630102146) Cfam 27:41049333	A C
(BICF2G630159183)		(BICF2G630798972) Cfam 28:38885325		(BICF2P1192522) Cfam 28:9877730	GG	(BICF2G630149030) Cfam 29:17561258	GG	(BICF2S236196)		(TI GRP2P356245_rs8830240) Cf am 29:36319325	A C
(BICF2G630271966)		(TI GR P2P 362535_rs9130694) Cfam 3:1252765		(BICF2G630276039) Cfam 3:24757939		(BICF2S23713161) Cfam 3:73570828		Cfam 30:15542105	A G	(B1CF2G630634836) Cfam 30:32852404	A G
(P17_3)		(P27_2) Cfam_31:21068798		Cfam 31:39391935	A G	Cfam_32:17792284	A G	(BICF2G630409193) Cfam 32:32382778		(TI GRP2P372104_rs9153277) Cfam 32:679380	GG
(BICF2S23124313)		(BICF2P1454500) Cfam_33:23742061		(BICF2G630200354) Cfam_34:195313	СС	(BICF2G630594648) Cfam 34:24396298		(BICF2P885380) Cfam_35:15345329		(G1425f16S28)	GG
(BICF2P516667)		Cfam_36:3565500	ΔА	(P2_3) Cfam_37:15436615		· · · <del>-</del> · · · · ·	GG	(TIGRP2P407751_rs8803124) Cfam_37:9398945		(BICF2P1226745) Cfam_38:17657161	
(BICF2P935470)		(BICF2P728698) Cfam 38:9224942		(P21_3) Cfam 4:31301072		(BICF2G630133028) Cfam 4:64121754		Cfam 4:75910211	ΔG	Cfam 4:86049027	GG
(BICF2P600196) Cfam 5:26320165		(BICF2P615597) Cfam 5:5410890		(BICF2P805553) Cfam 5:85451804			АА	(BICF2P1357746) Cfam_6:33976751		(BICF2S23126079) Cfam 6:64006720	
	G G	(BICF2S23648905) Cfam 7:36555518		(BICF2P1346673) Cfam_7:76294		(P8_1) Cfam 8:18121580		Cfam 8:45852939		Cfam 8:5291824	GG
(BICF2G630552597)		(BICF2G630558437) Cfgm 9:22610227		Cfam 9:40096141	G C	(BICF2P65087) Cfam_9:52710991		Cfam 9:60437147	GG	(P23_3)	
(BICF2S23449478)	3 6	(BICF2P1010945)	AG	(BICF2P1216677)	3 3	(P24_1)	3 6	(BICF2S22943825)	66		

Owner's Name : Amanda Broderick Pet Name : Merlin



### **Animal's Details**

Registered Name :	First's Flight Of Merlin The White
Pet Name :	Merlin
Registration Number :	SS23269704
Breed :	Golden Retriever
Microchip Number:	985113004247345
Sex:	Neutered Male
Date of Birth :	30th Dec 2020
Colour:	White Gold

### **ISAG Profile 2**

Cfam_1:119306331	GG	Cfam_1:72613047	A G	Cfam_1:74450772		Cfam_10:14685262	АА	Cfam_10:39548483	АА	Cfam_10:47923623	
(BICF2P635172) Cfam_10:57954366	GG	(P1_2) Cfam_10:8085469		Cfam_11:1161870		(BICF2G630666362) Cfam_11:62157625	GG	(BICF2G630488267) Cfam_11:70698603	A C	Cfam_12:23059939	АА
(BICF2P963969) Cfam_12:40681020	GG	Cfam_12:6337286	A G	Cfam_12:70657733	АА	(BICF2G630306265) Cfam_12:8532712	A G	(BICF2G630307199) Cfam_13:40616856	GG	(B1CF2P465276) Cfam_14:55735620	GG
(TI GR P 2P 164720_rs8839809) Cf am_16:29675662	СС	(BICF2P1193353) Cfam_16:58093031	СС	(BICF2P1183665) Cfam_17:12787849		(BICF2P496466) Cfam_17:57371669	GG	(BICF2P651575) Cfam_17:9407683	АА	(BICF2P1369088) Cfam_18:10189759	GG
(BICF2S23250041) Cfam_18:16385020	GG	(P24_3) Cfam_18:16388978	СС	Cfam_18:31579269	АА	(BICF2S2351979) Cfam_18:47325586	АА	(BICF2G630221287) Cfam_19:30246414	GG	(BICF2P46604) Cfam_19:40189405	СС
(B1CF2S23529290) Cfam_19:42756283	A G	(BICF2P250787) Cfam_20:45777531	A G	(BICF2P184963) Cfam_20:48602465	GG	(TI GR P 2P 255960_rs9030578) Cf am_20:6046176	АА	(P25_2) Cfam_21:22581321		(BICF2P1310805) Cfam_21:29796784	GG
(BICF2P401677) Cfam_21:31751817	GG	(BICF2P345488) Cfam_22:20498421		(BICF2P840653) Cfam_22:33934047	A G	(BICF2S22910736) Cfam_22:37522364	АА	Cfam_22:39647748	GG	(TI GRP2P283310_rs8881748) Cfam_22:61153661	GG
(BICF2P42825) Cfam_23:44497217		Cfam_23:48055836	СС	(BICF2G630328323) Cfam_24:18599997	A G	(BICF2P345056) Cfam_24:27925354		(BICF2S23519644) Cfam_24:30954773	A G	(P26_3) Cfam_24:43589304	A G
Cfam_24:45191477	АА	(BICF2G630365778) Cfam_25:4614777		(BICF2G630504410) Cfam_27:20948372	АА	Cfam_27:34444177	GG	(B1CF2G630499189) Cfam_27:42526114	A G	(B1CF2S23138418) Cfam_28:12804225	A G
(P15_2) Cfam_28:34478533	A G	(BICF2P1362405) Cfam_28:35104850	A G	(BICF2S2359809) Cfam_28:9703418	GG	(TI GR P 2P 354499_rs9162547) Cf am_29:19681270		(BICF2S22913753) Cfam_29:22992304	A G	(B1CF2G630274628) Cfam_29:4020192	GG
(B1CF2G630264994) Cfam_29:4022252	АА	(BICF2P1226838) Cfam_3:10255068	A G	(BICF2G630276136) Cfam_3:37849557	A G	Cfam_3:43055696	A G	(BICF2P950116) Cfam_3:43063677	A G	(BICF2P464536) Cfam_3:64084413	АА
(B1CF2S22912385) Cfam_3:90291255	G G	(BICF2S2399705) Cfam_3:91626907		(BICF2P643134) Cfam_30:10012939		(B1CF2G630340940) Cfam_30:11735245	A G	(B1CF2G630340944) Cfam_30:27619023	АА	(P4_3) Cfam_31:20912553	
(BICF2P285489) Cfam_32:13183511	A G	Cfam_33:15233992	GG	Cfam_33:22070526	GG	(BICF2P103615) Cfam_33:22472901	A C	(BICF2S22926284) Cfam_33:22648231	GG	Cfam_34:24351570	A G
(BICF2P1019402) Cfam_34:34993916		(BICF2S23356653) Cfam_34:37323213	GG	(BICF2G63078341) Cfam_34:41703614		(BICF2P378969) Cfam_35:15283717	GG	(TI GR P2P389035_rs9038546) Cfam_36:10084888	АА	(B1CF2S23649947) Cfam_36:12723744	СС
Cfam_36:18627936		(BICF2P590440) Cfam_36:288045	АА	Cfam_36:9241262		(B1CF2S23429022) Cfam_37:18338930		(BICF2P129670) Cfam_37:26611359	АА	(BICF2P70891) Cfam_37:28611801	A G
Cfam_37:30110473		(P6_2) Cfam_37:30902202		Cfam_38:13098194		Cfam_38:15271384	G G	(BICF2P129347) Cfam_38:19172567	СС	(B1CF2G630133994) Cfam_38:20930997	A C
Cfam_4:42104780	GG	Cfam_4:67040898		Cfam_4:70217695		(B1CF2S22928800) Cfam_5:13080303		(BICF2S23031254) Cfam_5:36642434		(B1CF2S23614068) Cfam_5:44650576	GG
(BICF2P1286728) Cfam_5:55349573	GG	Cfam_5:64611038	GG	Cfam_7:15017979	GG	Cfam_7:3318809	GG	Cfam_7:6423299		(B1CF2G630187658) Cfam_7:76487265	GG
(BICF2P496837) Cfam_8:19076567	A G	(BICF2P414351) Cfam_8:24614720	GG	(BICF2G630552598) Cfam_8:52381322		(BICF2P1173491) Cfam_8:6188937	АА	Cfam_8:67183794	АА	(B1CF2P798404) Cfam_9:20867959	
(BICF2P1391407) Cfam_9:32506288		(BICF2P1141966) Cfam_9:50114927		Cfam_9:56021221 (BICF2G630474528)	GG	(TI GR P 2P 116826_rs8741680)		(BICF2P789367)			

Owner's Name : Amanda Broderick Pet Name : Merlin



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Sex:	Neutered Male
Date of Birth :	30th Dec 2020
Colour:	White Gold

### **DNA Profile**

B1CF2G630102146	G G B1CF2G630149581	G G B1 CF2G630187649	A T BICF2G630187658	G G B1 CF2G630209373	A G B1CF2G630209508	A G
(BICF2G630102146)	(B1CF2G630149581)	(BICF2G630187649)	(BICF2G630187658)	(BICF2G630209373)	(B1CF2G630209508)	СС
BICF2G630255439	A G B1CF2G630274628	A G BICF2G630307199	A C BICF2G630340940	A G BICF2G630340944	A G B1CF2G630365778	
(BICF2G630255439)	(B1CF2G630274628)	(BICF2G630307199)	(B1CF2G630340940)	(BICF2G630340944)	(BICF2G630365778)	A G
BICF2G630382763	A G B1CF2G630437783	C C BICF2G630449851	A G B1CF2G630467607	C C BICF2G630488267	A A BICF2G630504410	
(B1CF2G630382763)	(B1CF2G630437783)	(BICF2G630449851)	(BICF2G630467607)	(BICF2G630488267)	(BICF2G630504410)	GG
B1CF2G630552598	G G B1CF2G630558437	A G BICF2G630594648	A G BICF2G630634836	A C BICF2G630641678	A G BICF2G630689403	
(B1CF2G630552598) B1CF2G630798972	(BICF2G630558437) A A BICF2G630814422	(BICF2G630594648) A C BICF2G63090019	(BICF2G630634836) A T BICF2P1019402	(BICF2G630641678) A G BICF2P103615	(BICF2G630689403) A G BICF2P1104630	АА
(BICF2G630798972)	(BICF2G630814422)	(BICF2G63090019)	(BICF2P1019402)	(BICF2P103615)	(BICF2P1104630)	A G
BICF2P1141966	G G BICF2P1173491	G G BICF2P1183665	A A BICF2P1193353	A G BICF2P1216677	G G BICF2P1226838	
(BICF2P1141966)	(BICF2P1173491)	(BICF2P1183665)	(BICF2P1193353)	(BICF2P1216677)	(BICF2P1226838)	СС
BICF2P1232055	G G BICF2P1271174	G G BICF2P129347	A A BICF2P129670	A A BICF2P1308802	A C BICF2P1310805	
(BICF2P1232055)	(BICF2P1271174)	(BICF2P129347)	(BICF2P129670)	(BICF2P1308802)	(BICF2P1310805)	АА
BICF2P1344095	A A BICF2P1346673	G G BICF2P1357746	A G BICF2P1454500	A A BICF2P155421	A A BICF2P157421	
(BICF2P1344095)	(BICF2P1346673)	(BICF2P1357746)	(BICF2P1454500)	(BICF2P155421)	(BICF2P157421)	TT
BICF2P182473	G G BICF2P224656	C C BICF2P237994	A G BICF2P246592	A C BICF2P250787	C C BICF2P25730	
(BICF2P182473)	(BICF2P224656)	(BICF2P237994)	(BICF2P246592)	(BICF2P250787)	(BICF2P25730)	АА
BICF2P283440	A G BICF2P285489	G G BICF2P345056	A A BICF2P347679	A G BICF2P378969	A C BICF2P382742	
(BICF2P283440)	(BICF2P285489)	(BICF2P345056)	(BICF2P347679)	(BICF2P378969)	(BICF2P382742)	АА
BICF2P415783	A G BICF2P422152	A G BICF2P508740	G G BICF2P516667	G G BICF2P553317	A A BICF2P554817	
(BICF2P415783)	(BICF2P422152)	(BICF2P508740)	(BICF2P516667)	(BICF2P553317)	(BICF2P554817)	АА
BICF2P561057	A C BICF2P585943	G G BICF2P624936	A A BICF2P635172	G G BICF2P65087	A G BICF2P651576	
(BICF2P561057)	(BICF2P585943)	(BICF2P624936)	(BICF2P635172)	(BICF2P65087)	(BICF2P651576)	GG
BICF2P717226	A A BICF2P751654	A G BICF2P774003	A A BICF2P798404	G G BICF2P842510	G G BICF2P856893	
(BICF2P717226)	(BICF2P751654)	(BICF2P774003)	(BICF2P798404)	(BICF2P842510)	(BICF2P856893)	GG
BICF2P878175	G G BICF2P935470	A G BICF2S22910736	A A BICF2S22913753	A G BICF2S22928800	G G BICF2S22943825	
(BICF2P878175)	(BICF2P935470)	(BICF2S22910736)	(BICF2S22913753)	(BICF2S22928800)	(BICF2S22943825)	GG
BICF2S23028732	TT BICF2S23031254	C C BICF2S23049416	A G BICF2S23057560	G G BICF2S23124313	G G BICF2S23126079	
(BICF2S23028732)	(BICF2S23031254)	(BICF2S23049416)	(B1CF2S23057560)	(BICF2S23124313)	(BICF2S23126079)	GG
BICF2S23246455	A A BICF2S23250041	C C BICF2S23333411	G G B1CF2S23356653	G G BICF2S23429022	G G BICF2S23449478	
(BICF2S23246455)	(BICF2S23250041)	(BICF2S23333411)	(BICF2S23356653)	(BICF2S23429022)	(BICF2S23449478)	A G
BICF2S23519644	G G BICF2S2351979	G G BICF2S2359809	A A BICF2S236196	A A BICF2S23626625	G G BICF2S23648905	
(BICF2S23519644)	(BICF2S2351979)	(BICF2S2359809)	(BICF2S236196)	(B1CF2S23626625)	(BICF2S23648905)	580 A A
BICF2S23649947	A G BICF2S23713161	G G BICF2S23737033	A G BICF2S24511913	A G TI GR P2P 106843_rs88588	16 GGTIGRP2P116826_rs87416	
(B1CF2S23649947) TIGRP2P164720_rs8839809	(BICF2S23713161) G G TIGRP2P177606_rs8886563	(B1CF2S23737033) G G TIGRP2P215708_rs8686029	(BICF2S24511913) A A TIGRP2P316532_rs859752	(TI GRP2P106843_rs88588 22 A A TI GRP2P372104_rs91532	316) (TI GR P 2 P 1 1 6 8 2 6 _ rs 8 7 4 1 7 7 A G TI GR P 2 P 4 0 2 0 4 2 _ rs 9 1 2 1 0	
(TI GR P 2P 164720_rs883980 TI GR P 2P 406551_rs9235397			9) (TI GRP2P316532_rs85975 G G BI CF2G630159183	522) (TI GRP2P372104_rs91532 A A BI CF2G630170631	277) (TI GR P 2 P 4 0 2 0 4 2 _ rs 9 1 2 1 A A B I CF 2 G 6 3 0 6 4 6 4 3 1	006) G G
(TI GR P 2P 406551_rs923539) B I CF 2P 1060087	7) (TI GR P 2 P 4 0 7 7 5 1 _ rs 8 8 0 3 1 2 G G B I C F 2 P 6 4 3 1 3 4	4) (BICF2G630271966) A G BICF2P990814	(BICF2G630159183) A A	(BICF2G630170631)	(BICF2G630646431)	
(BICF2P1060087)	(BICF2P643134)	(BICF2P990814)				

Owner's Name : Amanda Broderick Pet Name : Merlin







Scan to authenticate this Report online

Sample with Lab ID Number 24A106350 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] 1

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: 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.

Test Reported: DYSTROPHIC EPIDERMOLYSIS BULLOSA (GOLDEN RETRIEVER TYPE)

Result: NEGATIVE / CLEAR [NO VARIANT DETECTED] 1

**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.

Owner's Name: Amanda Broderick Pet Name: Merlin







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Sample with Lab ID Number 24A106350 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] 1

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 (GOLDEN RETRIEVER TYPE 2)

Result: NEGATIVE / CLEAR [NO VARIANT DETECTED] 1

Gene: ABHD5

Variant Detected: chr23:2274932-2274945 (canFam4): 14 bp deletion (GACTTCAACCAGAA)

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: Amanda Broderick Pet Name: Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported: ICHTHYOSIS A (GOLDEN RETRIEVER)

Result: CARRIER [ONE COPY OF THE 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.Asn482llefs9X

We have scanned your animal's DNA and one copy of the normal gene and copy of the positive (mutant) gene has been detected. The genotype of the animal tested is CARRIER this result may also be referred to as HETEROZYGOUS or A/N or "+/-". Being an autosomal recessive disease the animal will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal as it may produce affected offspring if mated to another carrier. It is recommended to have any breeding partner tested before breeding.

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

Result: NEGATIVE / CLEAR [NO VARIANT DETECTED] 1

**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.

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.

Owner's Name: Amanda Broderick Pet Name: Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

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

Result: NEGATIVE / CLEAR [NO VARIANT DETECTED] 1

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.

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.

Test Reported: VON WILLEBRAND'S DISEASE TYPE I

Result: NEGATIVE / CLEAR [NO VARIANT DETECTED] 1

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.

Owner's Name : Amanda Broderick Pet Name : Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

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

Result: E<sup>n</sup>/E<sup>n</sup> - NO MELANISTIC MASK (E<sup>n</sup>) 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 PHEOMELANIN 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 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 : Amanda Broderick Pet Name : Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported: BROWN DELETION = BD

Result: Bd/Bd - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [DELETION]

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 STOP CODON = BS

Result: Bs/Bs - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [STOP CODON]

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 INSERTION = BC

Result: BC/BC - DOES NOT CARRY BROWN/RED/LIVER or CHOCOLATE [INSERTION] 1

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 : Amanda Broderick Pet Name : Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

Test Reported: BROWN TYRP1 [LANCASHIRE HEELER TYPE] = BL

Result: BL/BL - 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 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: K/K - DOMINANT BLACK - SOLID [WILL NOT BE BRINDLED or EXPRESS AGOUTI] 1

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 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.

Owner's Name : Amanda Broderick Pet Name : Merlin







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Sample with Lab ID Number 24A106350 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported

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

**Result**:  $a^t/a$  - TRI COLOUR / TAN POINTS CARRIES SOLID COLOUR/BICOLOUR<sup>1</sup>

Gene: ASIP

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

Dog has black & tan and carries recessive black. Bi factored/white factored. Tan points include tan tips over the eyes, tan on the side of the muzzle, tan patches on the chest & tan on the legs. In some breeds white markings may "over ride" the tan markings known as tri. Please note that any genes on the "A" series will only be expressed if the K locus is kk, kkbr or kbrkbr.

Test Reported: SHEDDING (MC5R)

Result:

shd/shd [HIGH SHEDDING] - TWO COPIES OF THE shd (MC5R) VARIANT DETECTED REFER TO R151W (IC) FOR LEVEL OF SHEDDING

1

Gene: MC5R

**Variant Detected:** 

The dog will (may) exhibit a low leves 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 : Amanda Broderick Pet Name : Merlin



### 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 HET EROZYGOUS [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.

### **INDET ERMINABLE**

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.